POSTER SESSIONS

EXHIBIT HALL, SOUTH HALL A, LOWER LEVEL

The program number and the abstract/poster board number are one and the same. It appears in bold print followed by the letter **W** (Wednesday), **T** (Thursday), or **F** (Friday). The title and first author's name follow.

Session Topic/Title	**Abstract/Poster Board Numbers**	
	Start #	**End #**
Genome Structure and Function	373	438
Prenatal, Perinatal, and Reproductive Genetics	439	498
Genetic Counseling, ELSI, Education, and Health Services Research	499	593
Cancer Genetics	594	845
Mendelian Phenotypes	846	1180
Bioinformatics and Computational Approaches	1181	1457
Omics Technologies	1458	1541
Epigenetics and Gene Regulation	1542	1691
Developmental Genetics and Gene Function	1692	1795
Complex Traits and Polygenic Disorders	1796	2280
Evolution and Population Genetics	2281	2369
Molecular and Cytogenetic Diagnostics	2370	2601
Cardiovascular Phenotypes	2602	2760
Statistical Genetics and Genetic Epidemiology	2761	3020

REVIEWERS' CHOICE ABSTRACTS

(see website for a full list of authors)

The top 10% of posters, as determined by the reviewers' scores of the submitted abstracts, will receive a Reviewers' Choice Abstract ribbon. Look for the ‡ symbol in the online poster listings and keep an eye out for these high-scoring posters as you make your way through the poster hall.

POSTER SCHEDULE

Refer to the schedule below for presentation times and for the poster mounting/removal schedule. **Authors must put up and take down their posters according to the schedule below.** Authors must be present at their boards based on their odd or even abstract/program/board number, and must remain at their boards for the duration of their scheduled presentation times. **Posters should remain on the boards for all three days.**

WEDNESDAY

10:00 AM–11:00 AM	All poster authors (Wednesday, Thursday, and Friday) place posters on boards
10:00 AM–5:00 PM	Posters available for general viewing
2:00 PM–4:00 PM	**Poster Session I (Wednesday** Authors Present**)**
2:00 PM–3:00 PM	**(*odd* *poster board numbers; author* ***must be present*****)**
3:00 PM–4:00 PM	**(*even* *poster board numbers; author* ***must be present*****)**

THURSDAY

10:00 AM–5:00 PM	Posters available for general viewing
2:00 PM–4:00 PM	**Poster Session II (Thursday** Authors Present)
2:00 PM–3:00 PM	**(*odd* *poster board numbers; author* ***must be present*****)**
3:00 PM–4:00 PM	**(*even* *poster board numbers; author* ***must be present*****)**

FRIDAY

10:00 AM–2:00 PM	Posters available for general viewing
11:30 AM–1:30 PM	**Poster Session III (Friday** Authors Present**)**
11:30 AM–12:30 PM	***(odd*** *poster board numbers; author* ***must be present*****)**
12:30 PM–1:30 PM	***(even*** *poster board numbers; author* ***must be present*****)**

2:15-2:30 PM	All authors remove posters from boards
2:30 PM	Posters closed
2:30 PM	Exhibit Hall closed

Genome Structure and Function

373W The effect on the infection of Plasmodium falciparum by the increased Young's Modulus of G6PD deficient erythrocyte membrane. W. Jiang.

374T ‡ High-throughput functional genomic assessment of known and novel *HNF1A* missense variants to support clinical diagnostic interpretation. S. Althari.

375F A novel IDS gene mutation in two Japanese patients with severe mucopolysaccharidosis type II and correlation between developmental outcomes. S. Kasuga.

376W Novel frame-shift mutation in the extracellular domain of WNT coreceptor, low-density lipoprotein receptor-related protein 6 in a Japanese family with autosomal dominant oligodontia and early onset metabolic syndrome. H. Goto.

377T Primary bilateral macronodular adrenocortical hyperplasia due to mutations in *ARMC5*: New mutations in humans and modeling in zebrafish. A.G. Maria.

378F Cargo of miRNAs from synovial fluid exosomes contributes to pathogenesis of primary osteoarthritis. M. Czarny-Ratajczak.

379W Capture Hi-C identifies chromatin interactions between psoriasis-associated genetic loci and disease candidate genes. H.F. Ray-Jones.

380T Exome-wide copy number association study for arsenic-induced skin lesions: A prospective study. M.G. Kibriya.

381F Gene correction of epidermolysis bullosa simplex mutations using CRISPR/Cas9 technology. M. Bchetnia.

382W *ATP7B* gene variants for Alzheimer's disease affects ATP7B structure and markers of copper status. M. Rongioletti.

383T Age-related structural changes of the olfactory receptor subgenome in human blood cells and autologous brain regions. K. Szigeti.

384F Molecular mechanisms in C9orf72 ALS/FTD using the BAC transgenic mouse model with behavioral, neuropathological and molecular features of disease. A. Pattamatta.

385W Genomic architecture predisposes the *PLP1* region to the formation of complex genomic rearrangements. H. Hijazi.

386T Immunotherapy development for *C9ORF72* ALS/FTD using a BAC transgenic mouse model and human antibodies targeting RAN proteins. L. Nguyen.

387F Prolonged pentylenetetrazole exposure modulates the Rho Family GTPases pathway in the zebrafish brain. M.C.S. Nunes.

388W Chromosome 16q22-q24 uniparental disomy unmasks a rare recessive cause of early infantile onset epileptic encephalopathy 28. M. Davids.

389T Gene discoveries in autism are biased towards intellectual disability. M. Jensen.

390F Marker chromosome architecture and temporal origin revealed in a family with pleiotropic psychiatric phenotypes. C.M. Grochowski.

391W Local and global chromatin interactions are altered by large genomic deletions associated with human brain development. A. Urban.

392T Clinical features in a pediatric population due to chromosome deletions at a third level pediatric Mexican hospital in 24 years period of time: Five case reports. M.D. Hurtado-Hernandez.

393F Loss of Kctd13 in mice causes short-term memory deficiency. T. Arbogast.

394W The use of NGS 10X Genomics linked-reads to solve complex breakpoints mapping in individuals with balanced translocation. F. Mafra.

395T Identification of a novel frameshift mutation in the *MCPH1* gene causes primary microcephaly in a Saudi family. M.I. Naseer.

396F Single cell allele specific expression (ASE) in T21 and common trisomies: A novel approach to understand Down syndrome and gene dosage effects in aneuploidies. G. Stamoulis.

397W A sequential screening strategy for efficient rare gene discovery in small families. X.Z. Liu.

398T Using a combined approach of comparative genomic analysis, luciferase assay and CRISPR/Cas9 to identify and characterize cis-regulatory elements in the critical region of the 9p deletion syndrome. X. Hauge.

399F Trisomy 21 and chromatin. S.E. Antonarakis.

400W A comprehensive portrait of human somatic mosaicism. S. Vattathil.

401T The commitment complex *in vivo* over long intron genes. Y. Leader.

402F Exon-intron architecture in high and low GC-content genes affects alternative splicing. L. Tamer.

403W Chitinases mRNA levels quantified by qPCR in crab-eating monkey tissues. M. Uehara.

404T Comparison of chitinolytic activities among mammalian chitinases and bacterial chitinase. M. Kimura.

405F Revisiting the mouse reference genome: Single molecule sequencing of C57BL/6J "Eve". A. Srivastava.

406W Heat regulation of germ cell transcriptome: A bid to identify novel targets for contraception. A. Pandey.

407T Breakpoint analysis of chromosomes having inverted duplication with terminal deletion by NGS. H. Inagaki.

408F MethylHiC reveals long-range genetic-epigenetic and epigenetic-epigenetic interactions within the same single molecule. Y. Liu.

409W Complementary long and short-read sequencing techniques identify rearrangement structures and concurrent single nucleotide variant formation on chromosome 17p11.2. C.R. Beck.

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

410T Method for deciding detection limit of droplet digital PCR for rare mutations. T. Takahashi.

411F Targeted next-generation sequencing for identifying genes related to horse temperament. S. Song.

412W Cross-tissue protein expression and genetic regulation of transcription factors and cell signaling genes in enhancing Genotype-Tissue Expression (eGTEx) samples. M. Oliva.

413T Hanwoo-specific structural variations generate genomic diversity in the cattle genome. J. Park.

414F Study of G2 phase cell cycle arrest and its application in mediating *SOX9* mutagenesis in pluripotent stem cells (PSCs). T.Y. Ha.

415W Detection of novel L1 insertions in the chimpanzee (*Pan troglodytes*) genome. S. Lee.

416T Detection of non-reference L1 insertions in the human genome by using a target enrichment method. W. Shin.

417F Gene expression analysis and enzyme function of pig mammalian chitinase. E. Tabata.

418W Gene expression signature as a potential treatment monitoring biomarker for active tuberculosis in Thailand. R. Miyahara.

419T Heterogeneity of human ribosomes inferred from rDNA and rRNA sequencing. R. Nagaraja.

420F Discovery of rare, diagnostic *Alu*Yb8/9 elements in diverse human populations. J. Feusier.

421W Identification of active LINE-1 retrotransposons in the baboon genome. W. Lee.

422T Extending and improving GENCODE gene annotation. J.E. Loveland.

423F Updates to the human reference genome assembly (GRCh38). T. Rezaie.

424W Deep characterization of the contribution of short tandem repeats to gene expression across tissues. S. Feudjio Feupe.

425T The effect and mechanism of inhibiting G6PD activity on the proliferation of *Plasmodium falciparum*. Z. Zhang.

426F Integration of Hi-C chromatin loop calls across multiple resolutions identifies loops that are consistent across cell types and functionally associated. H. Li.

427W Moving into the darkness: Improving variant analysis with linked-reads. A.W. Xu.

428T Detection of rare copy number variants (CNV) in Kaiser cohort using DNA microarray data from Affymetrix axiom array. T. Haldar.

429F Genomic and structural integrity of human induced pluripotent stem cells. K. Kanchan.

430W *FusorSV:* An algorithm for optimally combining data from multiple structural variation detection methods. A. Malhotra.

431T ‡ Pervasive transcriptional dosage compensation buffers impact of autosomal structural variation. R.C. McCoy.

432F ‡ Germline *de novo* mutation clusters arise during oocyte aging in genomic regions with increased double-strand break incidence. C. Gilissen.

433W Linked-reads for high resolution individual genome analysis via haplotype reconstruction. S. Williams.

434T Characterisation and genotyping structural variation at the malaria-associated human glycophorin A-B-E cluster. W. Algady.

435F Low coverage sequencing of inbred animal backcrosses to check and correct genome assemblies. G.W. Nelson.

436W The mechanism and function of targeting lincRNAs by NMD in mammals. L. Hu.

437T Human germline mutation hotspots are characterized by a transversion-rich mutation signature. J. Carlson.

438F ‡ Predicting hotspots of *Alu*/*Alu*-mediated rearrangements in the human genome. X. Song.

Prenatal, Perinatal, and Reproductive Genetics

439W Chromosomal microarray analysis in fetuses with congenital heart disease: 5 years of clinical experience. P. Hu.

440T Do fetal endothelial nitric oxide synthase (eNOS) gene haplotypes influence prolonged preterm rupture of fetal membranes (PPROM)? K. Yanamandra.

441F Alteration of the expression and methylation modification of RAS in cardiomuscular tissue of the mice conceived by in-vitro fertilization. Q. Wang.

442W ‡ Ancestral disparities in genetic architecture of life course correlations between early growth and adulthood cardiometabolic disorders. F. Tekola Ayele.

443T PheWAS analysis of 13,000 individuals detects a common genetic variant that tags the Rh D blood group system in the European population. J. Fadista.

444F Compromised DNA repair and genomic imbalances in human male infertility. V. Singh.

445W Validation of a novel copy number variant detection algorithm for CFTR from targeted next-generation sequencing data. K. Kosheleva.

446T Genetic diagnosis of disorders of sex development (DSD): A national registry for disease-specific precision health. E.C. Delot.

447F Transethnic meta-analyses from genome-wide association studies of fibroid characteristics in African and European American women. M.J. Bray.

448W Non-immune hydrops fetalis with long bones fragility: A new subtype of osteogenesis imperfecta type II or a new form of skeletal dysplasia? G.A. Molfetta.

449T Prenatal diagnosis of diastrophic dysplasia: Importance of prenatal approach with NGS panel. M. Tamayo.

450F Neurodevelopment in Japanese singletons, aged 4—6 years, conceived by assisted reproductive technologies. T. Shimada.

451W Perinatal outcomes have little influence on FSIQ in children with 22q11.2DS. T. Crowley.

452T Independent head-to-head comparative diagnostic accuracy of NIPT methods in a prospective Canadian cohort of high-risk and low-risk pregnant women: The PEGASUS study. F. Rousseau.

453F Single-cell RNA sequencing in sperm from fathers of autistic children. J.A. Rosenfeld.

454W Unprogrammed presentation number

455T The Dutch TRIDENT studies: Implementing NIPT as part of the national prenatal screening program. E.A. Sistermans.

456F ‡ Large-scale cytogenomic analysis of samples from conception to childhood: A comprehensive assessment of the landscape of unbalanced genomic abnormalities. T. Sahoo.

457W ‡ Perinatal features and genotype-phenotype correlations in a large cohort of 355 patients with Prader-Willi syndrome. V. Kimonis.

458T Next generation sequencing based carrier screening study in Chinese population. G. Chen.

459F Carrier screening for 316 monogenic recessive diseases revealed high carrier frequency of rare known pathogenic mutations. D. Bercovich.

460W The utility of exome sequencing in prenatal diagnosis. E.A. Normand.

461T A genomic autopsy of perinatal death: Diagnosis and discovery by whole exome and whole genome sequencing. A.B. Byrne.

462F The Genomic Autopsy Study: Data from the first 50 cases indicates that whole exome sequencing/whole genome sequencing is a powerful adjunct to standard autopsy in the investigation of complex fetal and neonatal presentations. C.P. Barnett.

463W Novel pathogenic point mutation of KDM6A identified in a Chinese woman with Kabuki Syndrome type 2. W. Shi.

464T Congenital infection-like phenotypes: Are there any specific particularities available for prenatal diagnosis of rare diseases? N. Bourgon.

465F Prenatal diagnosis of Roberts syndrome suspected on sonogram in a consanguineous couple: A case report and a review of the literature. J.R. King.

466W Identifying the genetic causes underlying prenatally diagnosed structural congenital anomalies (SCAs) by whole exome sequencing (WES). G.K.C. Leung.

467T Increased nuchal translucency and Noonan Spectrum Disorders – A Mount Sinai hospital experience. P. Sinajon.

468F WES identifies likely pathogenic *FANCG* variants in a fetus with multiple congenital anomalies. B.D. Webb.

469W Large genome-wide meta-analysis of age at menopause including X chromosome, gene–environment interactions and Mendelian randomization analysis. F. Day.

470T HDP associated gene analysis in Japanese pregnant women in Maternity Log Study. Y. Tsunemoto.

471F Prenatal evaluation of a fetal cystic hygroma: An unexpected finding of a de novo fetal BRCA1 deletion. N.S. Seligman.

472W Detection of fetal subchromosomal aberration with cell-free DNA screening led to diagnosis of parental translocation: Review of 11344 consecutive cases in a university hospital. Y.Q. Qian.

473T Fetal cell-free DNA fraction in maternal plasma is affected by fetal trisomy. N. Suzumori.

474F ‡ Uncovering novel cytogenetic and molecular etiologies for infertility. S.L.P. Schilit.

475W Differential miR-346 and miR-582-3p expression in association with selected maternal and fetal complications. M. Su.

476T Challenges associated with increasing the predictive power of AMH in controlled ovarian stimulation. S.E. Parets.

477F Association of multiple TLR6-TLR1-TLR10 gene cluster SNPs with preterm birth in a Wisconsin cohort suggests a potential evolutionary selection bias. C. Hoffman.

478W Off the street phasing (OTSP): Free no hassle haplotype phasing for molecular PGD applications. G. Altarescu.

479T Novel genes for male infertility: Genetic and functional perspectives. D.V.S. Sudhakar.

480F Modeled fetal disease risk of a 175 condition expanded carrier screening panel. K.A. Beauchamp.

481W Analysis of blood plasma metabolomic profile analysis of pregnancy and the association analysis with pregnancy-related diseases in Maternity Log Study. Y. Harada.

482T Newborn metabolomics: Accurate prediction of gestational age from cord blood. E. Jasper.

483F Bacterial taxonomic analysis of oral microbiome in spontaneous preterm birth for Maternity Log Study. D. Ochi.

484W Maternal oral microbiota profile associated with hypertensive disorders of pregnancy. T. Yamauchi.

485T Prenatal screening for 22q11.2 deletions using a targeted microarray-based cell-free DNA (cfDNA) test. J. Shabbeer.

486F Correlation between nutrigenetic variants, adherence to the Mediterranean diet and obesity in male infertility. M. La Rovere.

487W Knowledge and attitudes on non-invasive prenatal pharmacogenetic testing among pregnant and preconception women. M.J. Ross.

488T Association between *MTHFR*, *MTHFD1* and *RFC1* gene polymorphisms and unexplained spontaneous pregnancy loss in Korean women. S. Shim.

489F Lessons from prenatal CMAs in low- and high-risk pregnancies: Is the incidental truly incidental? A.E. Eilat.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

490W ‡ Revealing transcriptome and methylome landscapes in a human oocyte by parallel sequencing. T. Lee.

491T Targeted next generation sequencing-based preimplantation genetic screening can enable detection of uniparental isodisomy, familial relationships, and polyploidy. M. Umbarger.

492F The revelation of complex chromosomal rearrangements through genome-wide cfDNA testing. T. Boomer.

493W Comparing maternal malignancies and multiple aneuploidies on prenatal cell-free DNA (cfDNA). E. Soster.

494T Broad spectrum of prenatal cultural artifacts detected by microarray. V. Potluri.

495F Prenatal NGS testing confounded by low levels of maternal cell contamination. Z. Wolf.

496W ‡ FXPOI: Modifying factors may play a larger role among the most vulnerable mid-range premutation group of women. E.G. Allen.

497T ‡ Novel application of fragile X CGG repeat analysis in a clinical setting: Outcomes and data derived from 716 embryos undergoing preimplantation genetic diagnosis (PGD). R. Cabey.

498F CarrierTest: The first experience with expanded preconception carrier screening. M. Koudová.

Genetic Counseling, ELSI, Education, and Health Services Research

499W Potential impact of predictive genetic testing among at-risk female relatives of *ATM* heterozygotes on breast cancer surveillance recommendations. D. Almanza.

500T Genetic testing in adult cancer patients in palliative care: What they understand, want, and may need. J. Bodurtha.

501F Educational and support needs of Lynch Syndrome probands and their relatives. W. Kohlmann.

502W ‡ *MSH6* and *PMS2* germline pathogenic variants implicated in Lynch syndrome are associated with breast cancer. M.E. Roberts.

503T ‡ Could population-level genomic screening be cost effective? An economic analysis informed by a large sequencing study. D.L. Veenstra.

504F Clinical predictors of gene panel choice when selected by the patient. E.K. Courtney.

505W *BRCA1/BRCA2* population screening in Ashkenazi Jews: Long term impact and familial communication. S. Lieberman.

506T Cancer susceptibility mutations in very young Israeli women with breast cancer. B. Nehoray.

507F Anticipated responses of genetic specialists and non-genetic specialists to unsolicited genomic secondary findings. K.D. Christensen.

508W Experiences and next steps in utilizing The Jackson Laboratory Clinical Knowledgebase (JAX-CKB), a relational database, for clinical and educational purposes. K. Sanghavi.

509T Hereditary colorectal cancer screening: A 10 year longitudinal study following an educational intervention. J.C. Carroll.

510F The Tell Me More Study: Returning medically actionable genomic variants in the setting of a neonatal genome sequencing study. T. DeMarco.

511W Genetic counseling assistants in a cancer genetics clinic: Genetic counselor time utilization and impact on patient volume. M.L.G. Hallquist.

512T A reporting of secondary findings in whole exome sequencing in Japan: The Project of HOPE. Y. Horiuchi.

513F Factors influencing sharing of genetic information: An in-depth interview study of patient who underwent cancer genetic testing in Singapore. S.T. Li.

514W Uptake of polygenic risk information among women at potentially high breast cancer risk. B. Meiser.

515T Assessing the possibility of *RUNX1*, *ETV6* and *GATA2* related germline predisposition in myeloid neoplasms in a somatic cancer setting. K. Barber.

516F Quality Improvement Project: Shared decision making in genetic cancer risk assessment and patient barriers to risk assessment and reduction in primary care. D.M. Person.

517W All in the Family: How family history affects diagnostic yield of hypertrophic aardiomyopathy multigene panel testing. S.J. Martin.

518T Potential impact of statin pharmacogenetic testing in an integrated healthcare system: The Integrating Pharmacogenetics in Clinical Care (I-PICC) Study. S.J. Miller.

519F Diabetes incidence after a polygenic risk intervention: Five-year follow-up in the Genetic Counseling/Lifestyle Change for Diabetes Prevention (GC/LC) Study. J.L. Vassy.

520W Acid sphingomyelinase deficiency (ASMD): Disease impact on families and caregivers. R. Avetisyan.

521T Screening African Americans for *APOL1*-associated kidney disease risk: Stakeholder views. S.M. Fullerton.

522F Association between lower folic acid level and unbalanced neuron excitability in Chinese pediatric patients. Y. Liu.

523W Providing genomic medicine to the Hispanic population at the Stanford Center for Undiagnosed Diseases. L. Fernandez.

524T Physicians attitudes, knowledge and barriers towards the inborn errors of the metabolism in a university hospital. F. Suarez.

525F A 25-year experience of Fragile X syndrome molecular diagnosis from a laboratory in Thailand. P. Limprasert.

526W Detection of genomic causes of developmental brain disorders among research participants in large-scale sequencing initiatives: Results disclosure, cascade testing, and psychosocial implications. B. Finucane.

527T ‡ Maternal origin of familial 22q11.2 deletions negatively impacts FSIQ scores. D.E. McGinn.

528F The significance of *KIAA2022* gene in development of early epileptic encephalopathy and intellectual disability in a girl. S. Zhilina.

529W ‡ Earlier answers are better answers: Family-centered utility of genome-scale sequencing for children with intellectual disability. K.B. Brothers.

530T Early diagnosis of Usher syndrome in non-syndromic young patients with hearing loss, the importance of pretest counseling. M.F. van Dooren.

531F Cri du Chat syndrome: Characteristics of 69 Brazilian patients. C.A. Kim.

532W Over FISHing: Findings in 1400 patients with 22q11.2 deletion syndrome. D. McDonald-McGinn.

533T Educational approach for TSC families at LeBonheur Children's Hospital Tuberous Sclerosis Center of Excellence. N. Urraca.

534F Genetic evaluation and counselling of couples who lost children affected by rare disorders. C. Skrypnyk.

535W The burden and economic impact of pediatric rare and undiagnosed genetic disease. N. Gonzaludo.

536T Lessons learned about expanded carrier screening using genome sequencing: Implications for research and practice. K.A.B. Goddard.

537F Data sharing to advance understanding of genetic disease: A model for data sharing agreements from the Newborn Screening Translational Research Network. A. Brower.

538W Cases of patients and informal caregivers who decided to handle their own health condition. V.P.F. Francisco.

539T Demystifying biomedical big data analysis through a massive open online course (MOOC). B.R. Haddad.

540F Section of Genetic Counseling at CHOP: Organizing for advocacy and professional development. M.H. Harr.

541W Genome Gateway: An online platform to increase communication between patients, providers and researchers. A. Hott.

542T Lumping and splitting: An age old dilemma with new age implications for disease classification. C. Thaxton.

543F Diversity and inclusion in genomic research: Why the uneven progress? S. Callier.

544W Genetic testing in the criminal justice system: Human rights perspectives. A. de Paor.

545T ‡ Fine-scale demography and behavior of male and female human geneticists. E. Glassberg.

546F Re-writing regulation? A comparative ethics and policy study of "natural" vs. "synthetic" cells, organoids and human genomes. R. Isasi.

547W Actions and reactions to negative results from genome sequencing in a healthy preconception population. T. Kauffman.

548T All, some or none: How wanting to learn genomic results differs within a national survey sample. J.B. McCormick.

549F Usability of family health history tools among underserved patients. C. Wang.

550W ‡ Why patients decline genomic sequencing studies: Experiences from the CSER consortium. L.M. Amendola.

551T Prostate cancer risk follow-up among *BRCA1/2* mutation carriers in Finland. O. Kajula.

552F ‡ Sociodemographic influences on attitudes and beliefs about genetic research and services. P.L. Bussies.

553W Views of experts and the public on genome editing and its issues: A literature review. I. Taguchi.

554T The Rare Genomes Project: Improving our ability to diagnose rare genetic conditions through a nationwide partnership with families. H. Brooks.

555F ‡ Impact of personal microbiome information on research volunteers. C. Bloss.

556W Establishing variant frequencies for pharmacogenomic data in a community health system based pharmacogenomics program. P.J. Hulick.

557T Primary care physician views on direct access pharmacogenomic testing in a community health system. A.A. Lemke.

558F ‡ Pharmacogenomic counseling: Exploring participant questions about PGx test results. T. Schmidlen.

559W Implementation of precision medicine intiatives: Special considerations for underserved communities. C.W. Brown.

560T Integration of a tool for electronic education and consenting within primary care to enable precision prevention. M. Moore.

561F The missing and the vulnerable: Developing responsible science policy for applying DNA to cross-border humanitarian causes. S.H. Katsanis.

562W Tipping the scales: Participants make healthy dietary changes in response to direct-to-consumer genetic test results. S.B. Laskey.

563T Genotypes associated with phenotypes: A human genetics laboratory exercise. D. Caporale.

564F f-treeGC: Questionnaire-based pedigree chart creation software in compliance with recommendations for standardized human pedigree nomenclature. T. Tokutomi.

565W Everything is actionable: Patient values and perceived utility of incidental genome sequencing results. Y. Bombard.

566T Repatriation of out-of-country molecular testing for disorders targeted by the provincial newborn screening program: Improving access to diagnostic testing in a cost and time efficient manner in Ontario, Canada - Our first year experience. B. Bélanger.

567F Developing a genomics ready clinical workforce: From raising awareness to the establishment of a faculty of genomic medicine. M. Bishop.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

568W Use of problem-based team learning to improve success of underrepresented students in an undergraduate genetics course. B. Bowling.

569T Phenotate: Crowdsourcing phenotype annotations of genetic disorders through student exercises. W.H. Chang.

570F The Genomics Education Partnership: Authentic big data course-based research projects for undergraduates. R.L. Glaser.

571W Utility of video-based education in the genetics clinic and beyond: Developing alternative service delivery models. P. Magoulas.

572T An interactive video vignette successfully teaches pedigree analysis to undergraduates. D.L. Newman.

573F RGEOde: Mining big data in the high school or undergraduate biology classroom. K. Pirc-Hoffman.

574W No one's genome is more interesting than your own: Understand Your Genome® through experiential education. E. Ramos.

575T Breakthroughs and barriers in secondary/post-secondary partnerships designed to promote research in molecular genetics, human genetics and GMOs. T.N. Turley-Stoulig.

576F Effectiveness of a dedicated rotation in genomics, genetic medicine, and undiagnosed diseases for internal medicine residents. M. Wheeler.

577W Perspectives and barriers to adoption of infobutton-related technologies for genomic medicine. L.V. Rasmussen.

578T Evaluating nonresponse bias in a longitudinal study of healthy adults receiving genome sequencing. S.D. Crawford.

579F MyCode participants' research priorities for precision health. E. Huang.

580W A conceptual framework for genome sequencing: Adaptation of the Theory of Planned Behavior. L. Hull.

581T Special considerations for genomic sequencing research in the United States Air Force: Development of the MilSeq Project. M.D. Maxwell.

582F "It's not an easy language for me to understand": Australians' expectations of support from health professionals for interpretation of personal genomic test results. S. Metcalfe.

583W The duty to warn at-risk relatives: The experience of genetic counselors and medical geneticists. T.J. Perry.

584T A legal analysis of the loss of chance doctrine in the context of genomics and precision medicine. J. Wagner.

585F Medical relevant findings from whole exome sequencing for apparently healthy individuals in physical examination of Chinese people. Z. Xia.

586W How Filipino parents cope with having a child with Maple Syrup Urine Disease. M.R. Tumulak.

587T ‡ Experience and specialty impact processes for interpretation and clinical application of genetic test results. C. Berrios.

588F A comparison of international policies on CRISPR and gene modification technologies and the risk of lag in Canadian science. Z. Master.

589W What determines value for genomics-informed healthcare? Evidence from a discrete choice experiment. D. Regier.

590T Measuring health outcomes in telegenetics. J. Stock.

591F Reproductive decision making process, medical concerns and special needs of preimplantation genetic diagnosis (PGD) users: Lessons from qualitative and quantitative analysis. S. Zuckerman.

592W Next-generation sequencing experience: Impact of early diagnosis of Usher syndrome. C. Wright.

593T Targeted genomic screening in unselected adults. K.R. Muessig.

Cancer Genetics

594F Burkitt lymphoma and skeletal dysplasia. S.L. Campbell.

595W *RB1* gene mutations in retinoblastoma patients from Central America. M. Dean.

596T Male breast cancer susceptibility due to FANCM mutation: A case report. R. Janavicius.

597F Beyond the NMD boundary: Characterizing the phenotypes of C-terminal *CDH1* mutations. K. Krempely.

598W Prevalence of the *UGT1A1*28* polymorphism in a population-based African American breast cancer cohort: A pilot study. A. Starlard-Davenport.

599T Muir-Torre syndrome presenting with juvenile astrocytoma. G.E. Tiller.

600F In-frame germline deletion of exons 23 and 24 results in DICER1 syndrome. M. Apellaniz-Ruiz.

601W Improving variant classification by incorporating pre-curated gene-specific knowledge into hereditary cancer multi-gene panel testing. H. Kang.

602T Evaluation of QIAGEN Clinical Insight as a content resource for variant curation in a CLIA laboratory. K. Karimi.

603F Genotoxic chemotherapies and radiotherapy contribute to the development of multiple primary tumors in patients with Li-Fraumeni syndrome. E. Kasper.

604W *FLCN* gene pathogenic variants in individuals presenting with lung cysts without pneumothorax, skin features or renal tumours typical of Birt-Hogg-Dube syndrome: A case series and review of the literature. K. Kohut.

605T Screening of over 1000 Indian patients with breast and/or ovarian cancer with a multi-gene panel: Prevalence of *BRCA1/2* and non-BRCA mutations. A.U. Mannan.

606F Cancer following radiotherapy for primary cancers in Li-Fraumeni syndrome patients. L. Oba.

607W Germline mutations in DNA repair genes are overrepresented in children and young adults with rhabdomyosarcoma: A discovery and validation cohort study. T. Wegman-Ostrosky.

608T Unexpected cancer-predisposition gene mutations in Cowden syndrome and Bannayan-Riley-Ruvalcaba syndrome patients without underlying germline *PTEN* mutations. L. Yehia.

609F Clinical and genetic analysis of patients with hamartomatous polyposis syndromes. J. Oh.

610W ‡ WGS in pediatric neuroncology patients shows a preponderance of germline Mendelian disease gene mutations. M. Bainbridge.

611T Identification of three novel loss of function mutations within *APC*, the causal gene for classical familial adenomatous polyposis. A. Gupta.

612F Functional characterization of germline *TMEM127* mutations in familial pheochromocytoma. S.K. Flores.

613W Systematic characterization of germline variants in endometrial carcinoma from the DiscovEHR study. J.E. Miller.

614T The HBV receptor gene *SLC10A1* is frequently down-regulated in hepatocellular carcinoma and is associated with poor survival. P. An.

615F Germline variants in mismatch repair genes are associated with microsatellite instability in sporadic tumors. A.R. Buckley.

616W Genetic polymorphisms of *CD40* ligand gene and susceptibility to cervical cancer. T. Chang.

617T The progression of global gene expression in melanoma: From normal skin to metastatic disease. L. Cordeiro.

618F CDKN2A germline polymorphisms demonstrate parallel associations of disease risk and clinical outcome in melanoma patients. S. Fang.

619W Association of cytokine polymorphisms with gastric cancer prognosis in Santiago, Chile. P. Gonzalez-Hormazabal.

620T Multi-gene hereditary cancer panel testing for *BAP1*. S. Hiraki.

621F Role of *HOXB13* in breast and ovarian cancer: Preliminary data from a laboratory-based multigene panel testing cohort. C. Horton.

622W Genome-wide analysis of shared and distinct germline genetic risk variants in colorectal cancer stratified by primary tumor site. J.R. Huyghe.

623T Birth order affects risk of multiple lymphoid cancers in lymphoid cancer families. S.J. Jones.

624F Rare DNA repair gene mutations predispose to young onset and lethal prostate cancer in the UK. Z. Kote-Jarai.

625W Genome-wide scan of single-nucleotide polymorphisms associated with prostate cancer specific death. W. Li.

626T Analysis of circulating tumor cells in multiple myeloma patients reveals mutations in proto-oncogenes and tumor supressor genes of NF-kB, Ras/MAPK, and PI3K/Akt pathways. D.S. Manjegowda.

627F BRA-STRAP: BRCA Refined Analysis of Sequence Tests: Risk And Penetrance. T. Nguyen-Dumont.

628W Genome-wide association study to identify the novel biomarker for response to tamoxifen. H. Ohnishi.

629T Gene environment interactions in the context of lobular breast cancer. C. Petridis.

630F Oral findings of cancer predisposition conditions: "Red flags" that dental, oral and craniofacial providers should recognize for early diagnosis, referral and management. A.M. Pham.

631W Exome sequencing of individuals with testicular germ gell tumor and family history reveals novel germline variants. L.C. Pyle.

632T Exome sequencing provides evidence of novel genes associated with colorectal cancer and polyps. E.A. Rosenthal.

633F Identification of novel prostate cancer susceptibility loci in Finnish population. C. Sipeky.

634W Co-heritability between aggressive and non-aggressive prostate cancer: Are germline risk loci the same for all prostate cancers? C.G. Tai.

635T A rare variant in GOLM1 predisposes to cutaneous malignant melanoma. C. Teerlink.

636F Association of TLL1 variant with hepatocellular carcinoma developed after eradication of hepatitis C virus. K. Tokunaga.

637W Biology and clinical implications of the 19q13 aggressive prostate cancer susceptibility locus. G.-H. Wei.

638T Cisplatin induced gene expression in lung and ovarian cancer cell lines reveal tissue specificity. S.W. Williams.

639F No association between 135G>C polymorphism of *RAD51* and colorectal cancer in Iranian population. N. Yazdanpanahi.

640W Functional evaluation of the correlation of gene expression changes in pathogenesis and platinum chemotherapy in lung and breast cancer cell lines. M.L. Yoder.

641T A genetic analysis of the change in prostate-specific antigen concentrations over time. S. Rashkin.

642F Characterization of global molecular architecture and regulatory mechanisms underlying hepatocellular carcinoma. H.M. Natri.

643W Molecular characterization of Brazilian patients with hereditary breast and ovarian cancer syndrome: What can we find beyond *BRCA1* and *BRCA2* genes? S.C.S. Carvalho.

644T Fine-mapping analysis of 152 breast cancer risk loci from OncoArray and iCOGS data. L. Fachal.

645F Gene expression imputation identifies novel candidate genes and susceptibility loci associated with risk of cutaneous squamous cell carcinoma. N.M. Ioannidis.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

646W Novel susceptibility loci associated with *BRCA*-negative *BRCA-like* breast cancer ("BRCAX") for Korean women. J.Y. Lee.

647T Melanoma genetics: Larger sample size identifies novel loci and enables causal inference. S. Macgregor.

648F A genome-wide association study of cisplatin-induced tinnitus in adult cancer survivors. B.L. Mapes.

649W Genetic association analysis of advanced neoplasia in a colon cancer screening cohort. X. Qin.

650T Genome-wide association study identifies novel loci for mammographic breast density. W. Sieh.

651F Phenome wide association study of breast cancer genetics reveals novel association with seborrheic keratosis. J. Liu.

652W Profiling of genomic alterations of mitochondrial DNA in gingivobuccal oral squamous cell carcinoma indicates that somatic mutations modulate prognosis in patients. A. Palodhi.

653T Prognostic inherited genetic variation in non-small cell lung cancer. F. Abbas Aghababazadeh.

654F Replication study and functional analysis identify a novel gene associated with gemcitabine-induced leukopenia/neutropenia. C. Udagawa.

655W Risks of melanoma in melanoma-prone families with and without *CDKN2A/CDK4* mutations over four decades. A.M. Goldstein.

656T Impact of interaction between single nucleotide polymorphisms (SNPs) in neural repair genes and patient characteristics on cognitive outcomes in patients with hematological malignancies treated with hematopoietic cell transplantation (HCT). N. Sharafeldin.

657F Novel analysis incorporating multiple tumor characteristics provide evidence of highly heterogeneous associations for known breast cancer risk loci. T.U. Ahearn.

658W Development of breast cancer risk prediction for the UK population using the UK Biobank dataset. K. Alajmi.

659T Integrative genomic analyses revealed candidate susceptibility genes in GWAS identified loci for colorectal cancer risk. J. Bao.

660F Genome-wide association study of Waldenström macroglobulinemia identifies novel loci. S.I. Berndt.

661W Evaluation of the impact of rare variants on glioblastoma susceptibility. R. Bohlender.

662T Familial-aggregation of somatic mutations in lung cancers. Y. Chang.

663F Classification of lung adenocarcinoma using optimized support vector machines applied to gene expression data. J.S. Diaz.

664W Whole-genome DNA methylation profiling in breast cancer by the Illumina MethylationEPIC array and the TruSeq EPIC sequencing platforms. C. He.

665T Fine mapping of the 6q25 breast cancer risk locus among Latinas reveals additional variants associated with risk. J. Hoffman.

666F Prevalence of cancer predisposition gene mutations among unselected pancreatic cancer patients. C. Hu.

667W Identification of hereditary mutations of breast cancer susceptibility and candidate genes in high-risk patients. M. Janatova.

668T Results of *BRCA1/2* testing in 1339 high risk Israeli patients with breast and/or ovarian cancer who were tested negative for the founder mutations. I.M. Kedar.

669F Whole exome sequencing reveals genes with elevated germline rare variants burden in myeloid malignancy patients. S. Li.

670W Large rearrangement analysis in *GREM1* and the identification of novel deletions and duplications. D. Mancini-DiNardo.

671T Identification of genetic variants associated with lung cancer risk among European and African Americans with COPD. V.L. Martucci.

672F Genome-wide homozygosity and risk of four non-Hodgkin lymphoma subtypes. A. Moore.

673W Whole-genome sequencing analysis of HPV31 and HPV35 reveals variability in cervical cancer risk. M. Pinheiro.

674T Height and body mass index as modifiers of breast cancer risk among 22,588 carriers of *BRCA1* or *BRCA2* mutations: A Mendelian randomization study. F. Qian.

675F A *cis*-eQTL genetic variant of the cancer-testis gene *CCDC116* is associated with risk of multiple cancers. N. Qin.

676W Multiple-gene sequencing revealed novel mutation characteristics beyond BRCA1/2 in Chinese women with familial breast cancer. Y. Shi.

677T Genetic variants in the 8q24 region are associated with prostate cancer risk in Mexican men. B. Silva.

678F Genetic variation and gastric cancer susceptibility in Western Honduras. G. Tavera.

679W A model averaging approach for improved *in silico* variant prediction. Y. Tian.

680T SNP-SNP interactions associated with prostate cancer aggressiveness in African Americans. H. Tung.

681F DNA methylation reveals distinct signatures in profiling tumor-infiltrating immune cells. X. Wang.

682W Risk factors associated with primary cutaneous melanoma of the scalp and neck. S.V. Ward.

683T Tumor profiling of separated carcinomatous and sarcomatous components from uterine carcinosarcoma biopsies provides insights into their development. Z. Weber.

684F Rare germline variants in known cancer predisposition genes in sporadic chordoma. Y. Xiao.

685W *BRIP1*, *RAD51C*, and *RAD51D* ovarian and breast cancer associations observed in a multi-gene panel testing cohort. L. Yackowski.

686T Rare germline variants in the promoter region of *CDKN2B* may cause melanoma predisposition. R. Yang.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

687F Combining weak signals by rank truncated products of p-values with application to detection of simultaneous closely-spaced mutations in cancer genomes. D. Zaykin.

688W Significance of secondary genetic findings in a large prospective population sample. K. Kristiansson.

689T A esophageal adenocarcinoma susceptibility locus at 9q22 also confers risk to esophageal squamous cell carcinoma by regulating the function of BARX1. C.W. Yan.

690F Detection of epigenetic field defects using weighted epigenetic distance-based method. Y. Wang.

691W Genome-wide interaction study of smoking behavior and non-small cell lung cancer risk in Caucasian population. Y. Li.

692T Type 2 diabetes susceptibility variants contribute to breast cancer risk. M.H. Black.

693F Novel approach to construction of causal gene regulatory network. R. Jiao.

694W Rate of reclassification of cancer genetic variants differs by race/ethnicity, depending on gene. L.R. Van Tongeren.

695T Genes associated with pancreatic cancer predict survival and prognosis. A. Gonzalez-Reymundez.

696F Efficient gene-by-treatment interactions test to develop predictive biomarkers on genome-wide studies using multidimensional hierarchical mixture models. T. Otani.

697W Two susceptibility variants associated with osteosarcoma in the HLA class II region. C. Zhang.

698T HPV16 genomic insights into cervical carcinogenesis: Viral oncogene E7 conservation is critical to carcinogenesis, and relation to human APOBEC3 activity. L. Mirabello.

699F Proteomics and genomics integration to predict ovarian cancer survival. U. Ozbek.

700W Survey of microRNA SNPs identifies novel breast cancer susceptibility loci in a case-control population-based study of African American women. J. Bensen.

701T Mosaic truncating *PPM1D* mutations are age-related but lack a strong association with breast cancer risk. T.A. Myers.

702F Gender differences in germline mutations in bladder cancer participants from the DiscovEHR study. L. Bang.

703W Integrative Bayesian group bridge regularization analysis in multiple heterogeneous high-dimensional survival data. Y. Li.

704T Inflated genome-wide *de novo* mutation rate in carriers of *TP53* germline mutations. X. Pan.

705F Detection of signal regions in whole genome genotyping and sequencing association studies using scan statistics. Z. Li.

706W A novel Bayesian multiple testing approach for region-based analysis of next generation sequencing (NGS) data. J. Xu.

707T Population-based breast cancer risk estimates associated with mutations in cancer predisposition genes from the CARRIERS study. F.J. Couch.

708F Clinicopathological and prognostic significances of EGFR, KRAS, BRAF and PI3KCA mutations in biliary tract cancer. H. Lee.

709W Evolutionary genomic analysis of a longitudinal series of prostate cancer bone metastases and xenografts from one patient revealed selection of progressively therapy resistant metastatic clone. T. Gaasterland.

710T The early evolutionary signatures of clonal hematopoiesis leading to blood based cancers and cardiometabolic conditions. K. Skead.

711F Similar frequency of ACMG-59 gene secondary findings in a large whole exome sequenced cancer cohort and ethnicity-matched controls. J. Kim.

712W Genetic diversity and rare mutations in the Puerto Rican population. E.J. Torres Gonzalez.

713T ‡ Functional role of intragenic methylation in alternative splicing in cancer. Y. Lee.

714F Systematic evaluation of copy number variations: Towards rational personalized cancer therapy. S. Appenzeller.

715W Single-molecule resolution of haplotype-specific, megabase-scale and complex oncogenic rearrangements in metastatic cancers. S. Greer.

716T NGSEA: Network-augmented Gene Set Enrichment Analysis. H. Han.

717F Methodologies for FMS-like tyrosine kinase (FLT3) internal tandem duplication (ITD) detection from two types of next generation sequencing data such as whole exome sequencing and amplicon sequencing. D. Kim.

718W Genomic analysis of PDX sequencing data. J. Kim.

719T A tailored topic model integrates both nucleotide context and genomic location heterogeneity in mutational process profiling. S. Li.

720F Copy number segmentation with left-to-right hierarchical Dirichlet process hidden Markov model and segment clustering. K. Liao.

721W Heterogeneous deconvolution of mixed tumor expression - DeMix-Py. R. Liu.

722T ‡ Identification of germline copy number variations (CNVs) using targeted sequencing data on 6q in hereditary lung cancer families. D. Mandal.

723F ViFi: Virus integration and fusion identification in tumor samples. N. Nguyen.

724W Expanding GEMINI to annotate and prioritize subclonal mutations in heterogeneous tumors. T. Nicholas.

725T Identification and characterization of novel oncogene candidates in invasive breast carcinoma. D.G. Piqué.

726F Pathogenicity of Mutation Analyzer (PathoMAN): A fast, automation of germline genomic variant curation in clinical sequencing. V. Ravichandran.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

727W Weighted similarity network fusion through integrating genomic functional annotation. P. Ruan.

728T Telomere length dynamics from whole genome sequencing using Telomeasure in progressing and non-progressing Barrett's esophagus. J.M. Shelton.

729F Practical noninvasive biomarkers: Identification of blood gene signatures for diagnosis of lung cancer. B. Song.

730W Identification of somatic tumor-only variants on on 1120 solid tumor cases through intelligent variant filtration. S. Van Vooren.

731T Integrated somatic mutation detection from tumor-normal sequencing data using multiple calling methods. Y. Wang.

732F Access and discover pathways from Pathway Commons. J.V. Wong.

733W Identification of germline copy number variations (CNVs) using whole-exome sequencing data in Caucasian and African American men with hereditary prostate cancer. K. Wood.

734T Optimal design of single cell studies for detecting and quantifying clonal subpopulations. J. Yu.

735F Evaluating relationships between pseudogenes and genes: From pseudogene evolution to their functional potentials. Y. Zhang.

736W Prediction of genome-wide DNA methylation in locus-specific repetitive elements. Y. Zheng.

737T Incorporating multiple NGS read features enables detection of transposon insertions across the genome. A. Zimmer.

738F A novel algorithm to identify somatic copy-number alterations which delivers high accuracy in targeted resequencing of cancer genes from tumor specimens. F.M. De La Vega.

739W AmpliconArchitect: On the fine structure of focal amplifications in cancer. V.B. Deshpande.

740T Tumor mutation burden (TMB) as a marker for DDR and IO combination. Z. Lai.

741F Genomic instability phenotypes in multidimensional genomic cancer studies. B.N. Lasseigne.

742W ‡ Predictive, discriminative versus associated or prognostic biomarker? Comparisons of discriminant, predictive and association and network analysis methods for mass spectrometry data from ovarian cancer. Y. Liang.

743T Tissue-specific feature of whole genome sequencing aids tissue-mapping in plasma. H. Liang.

744F Discovering resistance mechanisms of mutant *NRAS* melanoma towards MEK inhibitor treatment from patient derived tumors. G. Moriceau.

745W Access, visualize and analyze pediatric genomic data on St Jude Cloud. S. Newman.

746T Expression variability is associated with breast tumour subtype. J.F. Pearson.

747F Transcriptome-based classification of primary melanoma identifies tumor subtypes that predict outcome in stage I. R. Thakur.

748W A novel framework for tumor classification which uses sufficient dimension reduction for feature selection and Bayesian networks for integrating CT image and epigenomic. Y. Wang.

749T ‡ Multiregion high-depth whole exome sequencing of matched primary and metastatic tumors revealed inter- and intra-individual genomic heterogeneity and polyclonal seeding in colorectal cancer metastasis. Q. Wei.

750F The Seven Bridges Cancer Genomics Cloud: Enabling discovery from petabyte-scale human genomic data resources. E.H. Williams.

751W CliP: Fast subclonal architecture reconstruction from whole-genome sequencing data. K. Yu.

752T Dissecting tumor-immune system interaction in non-small cell lung cancer using TCGA data. X. Yu.

753F RADAR: A *RNA* bin*D*ing protein regulatory network resource for c*A*ncer *R*esearch. J. Zhang.

754W Domain retention in transcription factor fusion genes and its biological and clinical implications: A pan-cancer study. Z. Zhao.

755T Integrative approach to cancer driver gene discovery from somatic mutations. S. Zhao.

756F Identification of tissue-specific regulatory networks. M. Bilow.

757W A comprehensive characterization of tumor profiles using custom SureSelect targeted panels. A. Ashutosh.

758T GATK4 adds germline and somatic *copy number variant* plus somatic *SNV and indel* calling. S.H. Lee.

759F Detecting pathogenic structural variants with long-read PacBio SMRT Sequencing. A.M. Wenger.

760W Integrated search for multi-omics data using extended GA4GH Genomics API. S. Kawano.

761T MicroRNA eQTL analysis in pancreatic cancer with efforts towards functional validation. A. Jermusyk.

762F Designing for success: The right CRISPR design strategies for the right experiment. L. Brody.

763W Whole genome sequencing signatures for early detection of cancer via liquid biopsy. B.G. Kermani.

764T ‡ Developing validated phenotypic cancer cohorts for molecular stratification and susceptibility assessment, a use case: Patients diagnosed with early versus late stage non-small cell lung cancer. B.R. Johnson.

765F Somatic mutation identification through haplotype discrepancy in tumor-only sequencing without matched normal DNA. W. Chen.

766W Leveraging protein coding gene expression profiles to accurately impute lncRNA transcriptome of uncharacterized samples. A. Nath.

767T Subtype-specific expression of long noncoding RNAs in b-cell acute lymphoblastic leukemia. C. Nodzak.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

768F The NantOmics Pharmacogenomics Test: An integrative panomic approach to pharmacogenomics screening. C. Schwartz.

769W Cepip: Context-dependent epigenomic weighting for prioritization of regulatory variants and disease-associated genes. J. Wang.

770T Negative binomial model-based clustering: Discover novel molecular subtypes of ovarian cancer. Q. Li.

771F Identification of potential LREA regions in prostate cancer cell lines using multi-omic analysis features of Strand NGS software. P. Karuna.

772W Matched tumor/germline samples aid in detecting genomic instability in multiple myeloma using linked-read whole genome sequencing without the need for high molecular weight DNA. C. Ashby.

773T Expression-based Variant Impact Phenotyping (eVIP) for determination of somatic mutation function in cancer. A. Berger.

774F ‡ Fix-C: A novel experimental and computational method for structural variation detection and *in silico* long range phasing from FFPE tumor tissue. H.A. Costa.

775W Using liquid biopsies for low frequency variant detection and tissue-of-origin exploration. K. Cunningham.

776T Obtaining high quality transcriptome data from formalin-fixed, paraffin-embedded diagnostic prostate tumour specimens. L.M. FitzGerald.

777F Digital gene expression from low sample input: Highly multiplexed and robust profiling of formalin-fixed paraffin-embedded (FFPE) and fresh frozen samples from as little as 1 ng of RNA using the nCounter® Platform. D. Hanson.

778W Multiplexed molecular characterization of non-small cell lung cancer PDX models with NanoString's nCounter® Vantage 3D™ DNA:RNA:Protein Solid Tumor assay. D. Hinerfeld.

779T Development of a lysate-based multiplex proteomics platform using nCounter. J. Lee.

780F Functional validation of pleiotropic suspectibility loci for breast and ovarian cancer using chromosome conformation capture technology. J. Plummer.

781W Hereditary predisposition to asynchronous bilateral breast cancer: Going beyond BRCA1, BRCA2 and PALB2. M. Tischkowitz.

782T Genomic features of gastric cancer patient-derived xenograft (PDX) models. C. Zhang.

783F Comparative analysis between gene expression profile and genomic profile in adrenocortical carcinoma samples. F.P. Fortes.

784W Using NGS to detect mutations below 1% allele frequency in circulating cell free DNA and associated tumors. A. Wood.

785T Functional integration of genomic and transcriptomic data using Strand NGS explains drug resistance in basal cell carcinoma. S. Kapoor.

786F DNA repair improves sequencing accuracy in FFPE DNA samples. F. Stewart.

787W ‡ Longitudinal integrative omics of rituximab treatment on primary B cells. L.R.K. Brooks.

788T Use of Bionano Optical Maps to identify medically-relevant genomic variation. A.W.C. Pang.

789F Familially-inherited fusion genes as a new-class of cancer predisposition genes. D. Zhuo.

790W The identification of biomarkers for EGFR-TKI-induced interstitial lung disease through whole genome sequencing analysis. H. Zembutsu.

791T Novel sequencing adapters resolve index-hopping with unique, dual-matched barcoding and enable low frequency mutation detection with consensus analysis. M. Light.

792F Highly efficient double-stranded molecular tagging empowers improved accuracy of ultra-low frequency mutation detection. J. Wang.

793W New methods for high-throughput nucleic sequencing and diagnostics using a thermostable group II intron reverse transcriptase (TGIRT). C.D. Wu.

794T Cell cycle specific copy number profiling from parallel single cell genomics and transcriptomics. R. Rahbari.

795F A novel NGS target enrichment technology: Improved speed, selectivity, and uniformity. J. Pel.

796W Cryptic forms of mutant splicing detected by cBROCA. S. Casadei.

797T Exome sequencing reveals a novel germline gain-of-function *EGFR* mutation in a young adult with bilateral adrenocortical carcinoma. S. Akhavanfard.

798F Trans-eQTLs in prostate cancer risk. M. Bicak.

799W Interrogating key RECQL4 related genomic and epigenomic alterations in osteosarcoma. H. Horn.

800T Ion AmpliSeq™ TERT promoter sequencing. J.M. Kilzer.

801F The role of antioxidants in the context of carcinogen induced chromosome aberrations. Y.C. Li.

802W ‡ HPV16 integrated genomic and molecular characterization of cervical cancer in Guatemala. H. Lou.

803T Effects of 744ins20 - ter240 *BRCA1* mutation on breast/ovarian carcinogenesis and role of curcumin in telomerase inhibition. M. Pongsavee.

804F Rhesus macaques with mutations in MLH1 and MSH6 develop Lynch syndrome colorectal cancers. M. Raveendran.

805W Detection of viral sequences and integration sites in HPV-positive (HPV^{+}) recurrent/metastatic head and neck cancer (RMHNC) patients. D. Thach.

806T An integrative detection and analysis of structural variation in cancer genomes. J. Xu.

807F ‡ Functional characterization of a novel prostate cancer candidate gene at 2q37 in normal human prostate cell line. C. Cieza-Borrella.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

808W Optical mapping reveals a higher level of chained fusion events in human cancer. V.M. Hayes.

809T Comprehensive whole-genome analysis of the primary ENCODE cell line K562. B. Zhou.

810F De novo inference of enhancer-gene networks in diverse cellular contexts reveals the long-range regulatory impact of disease-associated variants. J. Wang.

811W Exosomes in cancer: Small vesicular transporters for cancer development and metastasis, biomarkers in cancer therapeutics. A. Abak.

812T Epigenetic regulation of *POLG1* in breast cancer. P. Bajpai.

813F In-depth analysis of genomics and epigenomics identifies a novel susceptibility lncRNA *GCLET* for gastric cancer. M. Du.

814W ‡ Clinical relevance of non-coding A-to-I RNA editing in multiple human cancers. T. Gu.

815T Epigenetic regulation of the Runx2 gene in lung cancer. A. Herreno.

816F Molecular mechanisms underlying serrated polyps: Comprehensive DNA methylation analysis reveals new targets in the serrated neoplasia pathway. V. Khammad.

817W Differential DNA methylation aspect of L1-chimeric transcripts in various cancers. S. Kim.

818T The effect of expression of glycosylation genes, regulators, and targets on cancer cell line sensitivity to drug treatment. J. Krushkal.

819F Motif disruption domains lead to cancer gene expression rewiring. F.C. Lamaze.

820W Effect of DNA methylation on expression of drug response genes. J.M. Oh.

821T Evaluation of circulating cell free DNA in bisulfite sequencing applications. M. Poulin.

822F Genetic polymorphism and gene expression of SHH & PI3K gene in ameloblastoma. H. Singh.

823W MiRNA profiling of pre-cancerous and cancerous condition of stomach by next-generation sequencing. J. Skieceviciene.

824T Epigenetics modification and gene expression studies upon human amniotic fluid stem cells treated with chemotherapeutic drugs. P. Upadhyaya.

825F Integrating chromatin and expression variation in statistical fine-mapping. M. Roytman.

826W Mismatch repair-associated mutations reprogram the colorectal cancer enhancer epigenome. S. Hung.

827T The genetic diversity affects the cell-fate in genotoxicity test. C.C. Lin.

828F Genome-wide map of APA in lung cancer: A pilot study. A. Zingone.

829W A genome-wide association study (GWAS) implicates NR2F2 in lymphangioleiomyomatosis pathogenesis. K. Giannikou*.

830T *In silico* reanalysis reveals novel prognostic miRNAs in pancreatic neuroendocrine tumors. V.K. Grolmusz.

831F Colorectal cancer-upregulated long non-coding RNA linc-DUSP regulates DNA damage response genes and promotes resistance to apoptosis. M.E. Forrest.

832W Antisense long non-coding RNAs in breast cancer: A transcriptome-wide disruption. S. Wenric.

833T Single cell RNA sequencing identifies novel gene expression signatures in a mouse model of multiple myeloma treated with the cIAP antagonist LCL161. N.E. Banovich.

834F Investigation of a transcription factor network involved in exocrine pancreatic development and homeostasis reveals a putative tumor suppressor role and a novel genetic interaction. J. Hoskins.

835W Disruption of Mi2b and MBD2/3 corepressor functions mediates LINE-1 reactivation andtumorigenicity in human bronchial epithelial cells challenged with benzo(a)pyrene. P. Bojang.

836T Genetic variations in alcohol-metabolizing genes (*GSTM1, GSTT1, CYP2E1, ADH2 and ADH3*) and pancreatitis risk in alcoholics. V. Aaren.

837F Determinants and consequences of ribosomal poverty and subsistence in *C. elegans*. E. Cenik.

838W Genetic variations in ERCC2 gene and the risk of developing head and neck cancer in an Indian population. K. Chukka.

839T MiR-450a and miR-450b-5p negatively impact the tumorigenic potential of ovarian epithelial cancer cells. B.R. Muys.

840F Integrative genome analysis of somatic p53 mutant osteosarcomas identifies Ets2 dependent regulation of small nucleolar RNAs by mutant p53 protein. R. Pourebrahimabadi.

841W ‡ NF1 mutation structure-function analyses using a full-length mouse cDNA. D. Wallis.

842T Whole exome sequencing of patient cell lines with highpersensitivity to radiation exposure identifies ATIC as a novel target for chemoradiosensitization. X. Liu.

843F Transcriptomic changes in NF1 deficient cells. C. Skefos.

844W Consequences of miR-122 loss in the development of hepatocellular carcinoma. P.N. Valdmanis.

845T Modeling human cancer syndromes using TALEN and CRISPR/Cas9 mediated genome editing in *Xenopus tropicalis*. K. Vleminckx.

Mendelian Phenotypes

846F Five cases report with maple syrup disease over a period of 16 years: Metabolic screening, detection of inborn errors of metabolism at the Hospital para el Nino Poblano, Mexico. P. Sánchez Meza.

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

847W The use of livers with metabolic disease for domino transplantation: Can a patient with an inherited metabolic disorder act as a liver transplant donor? P.A. Levy.

848T CPT1A p.P479L and long QT syndrome in Northern BC: Evidence for an increased risk for symptomatic hypoglycemia and implications for management. S.A. Collins.

849F Chart review is insensitive to ascertain pathogenicity of MODY gene variants of unknown significance. J. Goehringer.

850W Genotype-phenotype and structure-phenotype correlations of the insulin receptor gene mutations in patients with severe insulin resistance. J. Hosoe.

851T Blue genes or red genes: Using large scale sequencing cohorts to reassess the pathogenicity of monogenic diabetes genes. T.W. Laver.

852F Personalized medicine in diabetes mellitus: Lessons from the US Monogenic Diabetes Registry. M. Sanyoura.

853W *MAT1A* variants in methionine adenosyltransferase deficiency (MATI/III) suggesting dominant inheritance. J. Higgs.

854T Case report of congenital disorder of glycosylation caused by novel variant on COG6 gene diagnosed in early infancy. Z. Wei.

855F The incidence and mutational spectra of hyperphenylalaninemia in the Xinjiang Uygur population. Y. Su.

856W Mucopolysaccharidosis type VI (MPS VI) and molecular analysis: A review of published classified variants in the *ARSB* gene. M. Bailey.

857T Teeth loss and ungueal dysplasia as atypical features in Hunter syndrome. P. Garavito.

858F Type 2 Gaucher disease in an infant despite a non-mutated maternal *GBA1* gene. R. Grey.

859W Comparative plasma proteomic analysis in Korean patients with Fabry disease pre and post enzyme replacement therapy. S. Heo.

860T Evaluation of intracerebroventricular enzyme replacement therapy treatment with rhNAGLU-IGF2 from birth onwards in MPS IIIB mice. S.-h. Kan.

861F Genetic modifiers of *NGLY1* deficiency, a rare deglycosylation disorder, identified by exploiting natural variation in *Drosophila*. C.Y. Chow.

862W Clinical and molecular variability in Niemann-Pick disease type B. I. Focsa.

863T Monoallelic mutations in *OXCT1* in clinically and biochemically proven SCOT deficiency: Evidence of deep intronic mutations? C. Murali.

864F Newborn screening for Hunter Disease: Is the c.103+56_34 dup allele a pathogenic variant or a pseudodeficiency variant? Y.H. Huang.

865W Pharmacokinetics of glycerol phenylbutyrate in pediatric patients 2 months to 2 years of age with urea cycle disorders. S.A. Berry.

866T ‡ Increased expression of *SLC26A9* delays age at onset of diabetes in cystic fibrosis. A. Lam.

867F Genetic causes of hypercholesterolemia in the Emirati population. H. Daggag.

868W *CEBPA* mutation as a potential clinically novel cause of congenital generalized lipodystrophy. R.M. Mostafavi.

869T Biallelic mutations in *GPD1* gene in a Chinese boy mainly presented with obesity, insulin resistance, fatty liver, and short stature. N. Li.

870F Systematic characterization of mutations in familial hypercholesterolemia linked genes in Estonia. M. Alver.

871W Androgens and antioxidants management improve clinical & hematologic response of Fanconi Anemia Egyptian patients to bypass hematopoietic stem cell transplantation unavailability. A. Attia.

872T Very early-onset inflammatory bowel disease in a Mexican patient with an IL10 receptor deficiency due to a novel homozygous *IL10RB* mutation. D.E. Cervantes-Barragan.

873F PIDDGEN: A multi-disciplinary team providing molecular diagnoses of primary immunodeficiency diseases in South Africa. C. Kinnear.

874W Exome sequencing of extreme phenotypes identifies potential novel genes as modifiers of leg ulcer in sickle cell anemia. G.Q. Carvalho-Siqueira.

875T ‡ Genomic characterization of *F8* and *F9* copy number variants in the My Life, Our Future TOPMed hemophilia cohort. M.M. Wheeler.

876F Exome sequencing reveals novel compound heterozygous mutations in FOXN1 in patients with severe immunodeficiency and no alopecia. S. Khan.

877W An update on the diagnostics, phenotype and treatment of deficiency of ADA2 (DADA2). I. Aksentijevich.

878T ‡ Factor (F)VIII gene mutation type and type of FVIII therapeutic influence the risk of developing neutralizing anti-FVIII antibodies independent of genetic relatedness, age, race, hemophilia A (HA) severity, therapeutic exposure days, and haplotype in HA patients of the PATH Study. T.E. Howard.

879F Immuno-genomic association analysis of Factor VIII immunogenicity in hemophilia patients of the PATH Study using the ImmunoChip Array. B.W. Luu.

880W A new patient with common variable immunodeficiency (CVID) and autoinflammation due to biallelic mutations in HOIP. H. Oda.

881T Novel truncating variant in single immunoglobulin Interlekin-1 receptor related (*SIGIRR*) gene in a dominant family with early-onset inflammatory bowel disease. J.E. Horowitz.

882F Utility of whole genome sequencing for population newborn screening for immunodeficiencies. D.L. Bodian.

883W Severe EDS III with cell activation syndrome (MCAS) in infancy and young children. C. Tsai.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

884T Case report of a patient with uncharacterized IFN-γ mediated autoinflammatory disorder. O. Schnappauf.

885F A novel *GFI1B* mutation at the first zinc finger domain causes congenital macrothrombocytopenia. Y. Uchiyama.

886W Identification and characterization of adenosine deaminase 2 variants in pediatric vasculitis. K. Gibson.

887T Variants in *FOXP1* cause syndromic genitourinary tract defects. N. Bekheirnia.

888F Comprehensive analysis using targeted sequencing panel for congenital anomalies of the kidney and urinary tract and nephronophthisis in Japan. N. Morisada.

889W *HES1* gene screening in a cohort of patients with hipopituitarism reveal an allelic variant c.578G→A (p.G193D). R. Kertsz.

890T Rapid paediatric sequencing (RaPS) from patient to variant: A step-by-step workflow and case report. L. Boukhibar.

891F A novel deletion in ABCC9 gene identified through whole-exome sequencing of patient with clinical spectrum of Cantú syndrome. O. Migita.

892W Population-scale linkage mapping in a healthcare system uncovers novel signal for primary biliary cirrhosis. G.M. Belbin.

893T A case of vitamin D-dependent rickets type 1a with a novel-pathogenic variant in a Mexican patient. M. Abreu-González.

894F ‡ From genetics to therapy: CD55 loss of function leads to protein-losing enteropathy responsive to eculizumab. A. Kurolap.

895W High prevalence of PKD2 R803* mutation in Taiwan. D. Hwang.

896T Novel genotype-phenotype correlations in X-linked Alport syndrome: Serum albumin level, age at onset of hematuria and hypertension. L.I. Shagam.

897F Exome sequencing reveals novel candidate genes and potential oligogenic inheritance in patients with hypergonadotropic hypogonadism. A. Jolly.

898W Enzyme replacement therapy during dialysis in a patient with Fabry disease in a community hospital in New York. E. Astiazaran-Symonds.

899T *CFTR* gene mutations in the São Miguel island (Azores, Portugal): 20 years follow-up study. L. Mota-Vieira.

900F *TRPV4* alternative splicing transcripts in metatropic dysplasia. S.M. Kirwin.

901W Nora's Lesion or something less "bizarre": Case report of family with benign bone tumors and review of the literature. E. Carter.

902T A novel mutation in the C-Terminal Associated Peptide (TCAP) region of Teneurin 3 found to co-segregate in all affecteds in a multi-generation family with developmental dysplasia of the hip. G.J. Feldman.

903F Dual genetic diagnoses identified in a large family with brachydactyly type A1 and insulin resistance using whole-exome sequencing. R. Ho.

904W Mutated *DMRT2* causes a distinct type of spondylocostal dysostosis (SCD). Q. Waisfisz.

905T Potential pathogenic variants identified in a Turkish tooth agenesis cohort via whole exome sequencing. R. Du.

906F Clinicaland molecular heterogeneity in VCP autosomal dominant inclusion body myopathy. S. Al-Tahan.

907W Survey of patients with Ollier disease and Maffucci syndrome over Facebook compared to review of clinical literature. C. Smith.

908T Dyggve-Melchior-Clausen syndrome, a case report with typical family tree. L. Mora.

909F Case series of individuals with novel syndromic phenotypes characterized by enchondromas and/or exostoses with or without vascular anomalies. S.M. Robbins.

910W Biallelic mutations in FLNB cause a skeletal dysplasia with 46,XY gonadal dysgenesis by increasing β-catenin expression. K. Upadhyay.

911T ‡ Multi-center cohorts with animal model and genotype-phenotype analyses: deciphering a new and undefined subtype of congenital scoliosis*, TBX6*-associated congenital scoliosis (TACS). N. Wu.

912F Multicentric carpotarsal osteolysis syndrome in mother and daughter misdiagnosed as juvenile rheumatoid arthritis. K. Chen.

913W Recurrence of perinatal lethal osteogenesis imperfecta due to parental mosaicism for a novel dominant mutation in *COL1A1*. A. Ruiz-Herrera.

914T Case report of a mild skeletal phenotype secondary to mutations in *LBR* gene. M.D.F. Carvalho.

915F ‡ Loss of inhibition of mTOR signaling in a new form of a metaphyseal chondrodysplasia due to a recessively inherited mutation in salt inducible kinase 3 (*SIK3*). F. Csukasi.

916W Enzyme replacement therapy in perinatal hypophosphatasia: Case report and recommendations for clinical practice. L. Dupuis.

917T Spinal manifestations of patients with musculocontractural Ehlers-Danlos syndrome caused by CHST14/D4ST1 deficiency (mcEDS-*CHST14*). T. Kosho.

918F Longitudinal growth curves for OI caused by structural mutations in type I collagen. J. Marini.

919W ‡ Gain of function germline mutations in *ABL1* are associated with congenital heart defects, skeletal malformations, and failure to thrive. Y. Yang.

920T Investigation of the molecular basis of familial and isolated Tarlov cysts. M. Muriello.

921F ‡ *TMEPAI* mutation causes MFS/LDS-like phenotypes in 2 Japanese families. H. Morisaki.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

922W ‡ A distinct cutaneous blistering phenotype with multi-system manifestations caused by a mutation in CD151, the 20th causative gene in epidermolysis bullosa. H. Vahidnezhad.

923T Early oro-dental manifestations as a clue for the clinical diagnosis of infantile systemic hyalinosis. I.S.M. Sayed.

924F Novel compound heterozygous variants in the gene *CHUK* associated with AEC syndrome-like phenotype and immune system involvement. M. Cadieux-Dion.

925W Understanding the impact of a novel homozygous nonsense *CAST* gene mutation in a PLACK family. S.G. Temel.

926T Delineation of musculocontractural Ehlers-Danlos syndrome caused by dermatan sulfate epimerase deficiency (mcEDS-*DSE*): Report of additional patients and comprehensive review of reported cases. A. Unzaki.

927F *ANKRD26* loss of function somatic mutation in a female case with Tree Man Syndrome. K.M.F. Uddin.

928W ‡ Mutations in *SULT2B1* cause autosomal recessive congenital ichthyosis in humans. L. Heinz.

929T Cutaneous neurofibromas in neurofibromatosis type 1: A quantitative natural history study. A. Cannon.

930F Abnormal splicing in a case of epidermolysis bullosa with a novel synonymous mutation in the *ITGB4* gene. E. Tan.

931W A disease-associated *REEP1* variant affects splicing of the gene's 3'UTR. C. Beetz.

932T Synonymous variant in *KCTD7* causes alternative splicing in siblings with progressive epilepsy. D.B. Zastrow.

933F NeuroChip genotyping of the Johns Hopkins brain bank reveals common and rare genetic associations. C. Blauwendraat.

934W A *PSEN2* frameshift variant associated with early onset AD in two families. S. Jayadev.

935T Comparison of the mutations that cause Alzheimer's disease on secondary protein misfolding on transgenic mouse models. G. Xu.

936F ‡ Regulatory role of RNA chaperone TDP-43 for RNA misfolding and repeat-associated translation in SCA31. K. Ishikawa.

937W Genetic analysis in pediatric patients with ataxia. J. Lee.

938T A novel *PRRT2* pathogenic variant in a family with paroxysmal kinesigenic dyskinesia and benign familial infantile seizures. J.G. Lu.

939F ‡ Expanded-(TGGAA)n-associated unconventional translation in spinocerebellar ataxia type 31. N. Sato.

940W A novel mutation in eukaryotic elongation factor 2 kinase (eEF2K) decreases phosphorylation of eEF2 in a patient with degenerative ataxia. K.L. Sund.

941T Japan Consortium of Ataxias (J-CAT): A cloud -based national registry for degenerative ataxias providing comprehensive genetic diagnosis and prospective natural history researches. Y. Takahashi.

942F Two patients with *PNKP* mutations presenting microcephaly, seizure, and oculomotor apraxia. M. Taniguchi-Ikeda.

943W Identification of novel *de novo* CHD8 variants associated with autism, language disability and overgrowth. Y. An.

944T *Tmlhe* and *Bbox1* null mouse models of carnitine deficiency. A. Ye.

945F ‡ Severity of *GABBR2* mutations determines neurological phenotypes ranging from Rett-like syndrome to epileptic encephalopathy. Y. Yoo.

946W Characterization of a complex translocation causing 3q28ter duplication and 10q26.2ter deletion in a child with self-injurious behavior. I.M. Adeshina.

947T ‡ The novel aldehyde trap ADX-102 reduces accumulations of GHB and GABA in brain tissue from succinic semialdehyde dehydrogenase-deficient mice. S.G. Macdonald.

948F Identification of mutations in patients from southern Italy with amyotrophic lateral sclerosis using multigene panel testing. G. Annesi.

949W Evaluation of pathogenic non-coding variants within whole genome data using encephalopathies as a model. D. Misceo.

950T Novel mutations in *CLN6* cause late-infantile neuronal ceroid lipofuscinosis in two unrelated patients. B. Behnam.

951F 17p13.3 microdeletions between *YWHAE* and *LIS1* (*PAFAH1B1*) cause a unique leukoencephalopathy. L.T. Emrick.

952W ‡ Large-scale systematic analysis of recessive neurodevelopmental disorders in consanguineous families. A. Gregor.

953T Genetic, clinical, and imaging study of Pelizaeus-Merzbacher disease using the Integrative Brain Imaging Support System (IBISS). K. Inoue.

954F Rare *SOX30* variants in juvenile myoclonic epilepsy. S. Jaishankar.

955W ‡ AOH-mediated recessive mutation burden can result in blended phenotypes. E. Karaca.

956T Characterizing genetic causes of neurodevelopmental disorders with brain malformations in a predominantly Turkish cohort. J. Punetha.

957F c.105C>A [p.(Tyr35Ter)] variant in *AIMP2* causes microcephaly, intellectual disability, seizures and spastic quadriparesis. A. Shukla.

958W Expanding the genetic spectrum in myoclonic astatic epilepsy. S. Tang.

959T Exome sequencing identifies a novel *FBXO38* variant inherited from a mosaic mother to cause distal hereditary motor neuronopathy Type IID with distinct features. S.A. Ugur Iseri.

960F Aspartate supplementation for aspartate-glutamate carrier isoform 1 deficiency. S. Yano.

961W ‡ *De novo* missense variants in *GNAI1* gene are associated with epileptic encephalopathy. M. Liao.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

962T *BICD2*-related arthrogryposis with unexplained cardiomyopathy. R.D. Kastury.

963F A tuberous sclerosis positive case without cortical tubers and subsequent diagnosis of "unaffected" family members. R. Caylor.

964W Characterizing the rare X-linked dominant variant in *ALG13*: A case report. J. Kohler.

965T Clinical presentation and genotype-phenotype correlation of a complex neurodevelopmental disorder caused by mutations in *ADNP*. F. Kooy.

966F A novel homozygous mutation in two sisters diagnosed with Joubert syndrome. A. Sen.

967W Parent-of-origin effects in 15q11.2 BP1-BP2 deletion syndrome. K. Davis.

968T Case report of a patient with a *TANGO2* deletion that provides additional phenotype information. R. Godshalk.

969F Periventricular nodular heterotopia as hallmark of a new ciliopathy related to *CRB2* mutation. G.M.S. Mancini.

970W Clinical management of patients with GLUT1 deficiency syndrome (De Vivo disease). T. Kozhanova.

971T *CAD* mutations and uridine-responsive epileptic encephalopathy. I. Bader.

972F Impact of a targeted next generation sequencing (TNGS) strategy for the genetic diagnosis of early onset epileptic encephalopathies. S. Gobin.

973W Linkage and haplotype analyses of families with benign adult familial myoclonic epilepsy (BAFME). H. Ishiura.

974T New epilepsy genes and variants discovered utilizing patients referred for clinical genetic testing. K. McCarty.

975F ‡ An integrated whole-genome, whole-transcriptome approach to genetic diagnosis in developmental and epileptic encephalopathies. A.M. Muir.

976W The genetic landscape of the epilepsy-aphasia spectrum disorders. C. Myers.

977T *STXBP1* encephalopathy with epilepsy: 6-year-old girl with de novo missense variant in *STXBP1* expands the phenotype. J. Pappas.

978F High-depth multi-gene panel analysis with integrated sequence and copy number detection is a useful first-tier test with a high diagnostic yield and broad mutation spectrum detection in childhood epilepsy. N. Patil.

979W Novel biallelic *SZT2* mutations in three cases of early-onset epileptic encephalopathy. N. Tsuchida.

980T Whole exome sequencing reveals potential oligogenic inheritance and candidate novel genes in patients with arthrogryposis. Y. Bayram.

981F A novel *MTMR2* gene mutation (c.1168 G>T, p.E390*) in a patient with Charcot-Marie-Tooth Disease Type 4B1. M. Erdogan.

982W Exome sequencing in Italian FTD patients reveals probable novel mutations in neurodegeneration associated genes. M. Hammer.

983T Clinical and molecular insights into developmental abnormalities of corpus callosum. M. Hebbar.

984F Heterozygous missense variant in *TDRKH* encoding tudor and KH domain-containing protein associated with autosomal dominant motor neuropathy. K. Kosaka.

985W Familial choreoathetosis: A novel heterozygous mutation in *PDE10A*. D. Narayanan.

986T A novel *de novo* alteration in *SLC12A6* in a patient with early-onset severe progressive sensorimotor polyneuropathy and abnormal EEG. M. Rossi.

987F N98S mutation in *NEFL* gene causing a mild form of Charcot-Marie Tooth disease. A. Sanchez.

988W Missense mutations and multiplications of alpha-synuclein in familial Parkinson's disease: Genotype-phenotype correlation. K. Nishioka.

989T Expanding the clinical spectrum of ARL6IP1-associated hereditary spastic paraplegia. S. Majid.

990F Genomic analysis identifies new loci associated with motor complications in Parkinson's disease. S. Chung.

991W Genome-wide association study identifies potential genetic modifiers in Charcot-Marie-Tooth disease type 1A. F. Tao.

992T ‡ Integrative omics analysis of a cohort of 198 singletons with cerebral palsy. J. Gecz.

993F Novel *TSC1/TSC2* pathogenic variants in Hungarian cohort with tuberous sclerosis complex: Clinical and molecular genetic aspects. E. Kovesdi.

994W Structural and sequence characterization of SMN1 and SMN2 genes in SMA patient collection. C. Sun.

995T Heterozygous *COG4* variant causes a non-lethal type of COG4-CDG (formerly CDG-IIj). R. Hamid.

996F Spectrum of *TTN* variants in a patient cohort of neuromuscular disorders. P.S. Lai.

997W Whole exome sequencing data analysis in hereditary spastic paraplegia patients from Turkey. B. Ozes.

998T ‡ Mutations of the *ZNF292* gene are a novel cause of neurodevelopmental disability, behavioral problems, and autism spectrum disorders (ASD). G. Mirzaa.

999F Correction of *NAGLU* mutation p.R297X using CRISPR/Cas9 gene editing in mucopolysaccharidosis IIIB patient-derived iPSCs. C.L. Christensen.

1000W Progressive abnormal myelination and cerebrospinal fluid volume in canine mucopolysaccharidosis type I: A neuroimaging and neuropathological study. P. Dickson.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

1001T Magnetic resonance spectroscopy and lipid profiling of myelin composition in corpus callosum of mucopolysaccharidosis I mice. S. Le.

1002F Evaluation of sibling pairs with Gaucher disease discordant for Parkinsonism. G. Lopez.

1003W Phenotypic profiles of *GBA1* mutation carriers with and without Parkinson disease: A data-driven approach. A.M. Steward.

1004T The Lysosomal Disease Network. C.B. Whitley.

1005F A novel pathogenic variant of *PURA* in a patient with severe developmental delay, delayed myelination and empty sella. K. Hosoki.

1006W Novel *NTRK1* gene mutation and clinical report of HSAN-IV phenotype in a Mexican patient. L. Patron.

1007T Further clinical and molecular characterization of the novel autosomal recessive neurodegenerative disorder related to the *ATP8A2* gene. A. Telegrafi.

1008F Evaluating clinical and biochemical endpoints for therapy in Pex7 deficient mouse models. W. Fallatah.

1009W ‡ Mitochondrial accumulation and increased lipid metabolism in a *Dhtkd1*^{Tyr486*} knock-in mouse model of the CMT2Q neuropathy. M. Gu.

1010T Myotonia congenita with a novel missense mutation in *CLCN1* gene (*c.680T>A*, p.Ile227Asn). A. Kiraz.

1011F Is the association of heterozygous variations in *MORC2*, *MFN2* and *AARS* genes responsible for a severe axonal form of Charcot-Marie-Tooth disease? A. Lia.

1012W Development of new strategies for the treatment of hereditary cystatin C amyloid angiopathy (HCCAA). A. Gutierrez-Uzquiza.

1013T Search for target genes of transcriptional regulation by dentatorubral-pallidoluysian atrophy protein (DRPLAp) that acts as transcriptional co-regulator. K. Hatano.

1014F New *DNAJC5* mutation initially missed by Sanger sequencing and whole-exome sequencing identified in a familial case of adult-onset neuronal ceroid lipofuscinosis (ANCL). I. Jedlickova.

1015W ‡ Clinical features and the pathomechanism of early childhood-onset neurodegenerative encephalopathy arising from biallelic *TBCD* mutations. N. Miyake.

1016T Mutant human proteins linked to familial neurodegeneration cause secondary protein misfolding in the spinal cord. M.C. Pace.

1017F Unbalanced translocation causing unbalanced brain: A case of hemimegalencephaly. A.R. Barone.

1018W Expanding the natural history of *KIF1A* associated disorders (KAND). L. Boyle.

1019T An autopsy case of familial amyloid polyneuropathy (FAP) with novel transthyretin (TTR) mutation (TTR, Lys80Arg). H. Furuya.

1020F Identification of novel *SNORD118* mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. K. Iwama.

1021W New homozygous missense mutation in *NT5C2* underlying hereditary spastic paraplegia SPG45. A. Onoufriadis.

1022T Spastic paraplegia type 4: A novel *SPAST* splice site donor mutation and expansion of the phenotype variability. A. Orlacchio.

1023F A de novo *HNRNPU* gene mutation identified in a patient with symptomatic infection-associated acute encephalopathy and developmental delay. S. Shimada.

1024W ‡ Naturally occurring human genetic variation suggests *LRRK2* inhibition is a safe therapeutic strategy for Parkinson's disease. I.M. Armean.

1025T Phenotypical features and genetic findings in Lithuanian patients with CMTX1. B. Burnyte.

1026F Lack of CHCHD2 mutations in Parkinson's disease in a Southern Italy population. G. Iannello.

1027W DNAJC13 familial Parkinson's disease from South Italy. R. Procopio.

1028T A rare male patient with classic Rett Syndrome caused by MeCP2_e1 mutation. A. Goji.

1029F Epidemiology and genetics of Chiari 1 malformation. B. Sadler.

1030W Report of phenotypic variability of periventricular nodular heterotopia in a four-generation Caucasian family with a novel *FLNA* mutation. D. Khattar.

1031T Sensory, behavioral, and social phenotypes observed in individuals with Williams syndrome in Japan. T. Awaya.

1032F Combining Bionano and exome sequencing identifies a homozygous structural variation in the novel *AGBL3* gene underlying microcephaly. D. Belandres.

1033W Genetics of childhood-onset psychosis. M. Ameri.

1034T Expansion of the molecular and phenotypic spectrum of *CAMTA1*-related neurological disorders. L.B. Henderson.

1035F ‡ Delineation of a new neurobehavioral syndrome associated with mutations in RFX3. H. Hodges.

1036W Delineation of the phenotype associated with *de novo TBR1* variants in 15 unrelated patients and review of the literature. S. Nambot.

1037T ‡ *De novo TCF20* pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological deficits with similarities to Smith-Magenis syndrome: Seven new cases further delineate the phenotypic presentation of this new syndrome. F. Vetrini.

1038F Intellectual disability with severe self-injury behavior caused by *THOC2* splice site variant. N. Ishihara.

1039W Guidelines for phenylbutyrate drug levels in the management of urea cycle disorders. Y. Jiang.

1040T ‡ Novel *de novo TAOK1* variants associated with a neurodevelopmental phenotype, macrocephaly, and joint hypermobility. H.M. McLaughlin.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1041F Neurodevelopmental profile for boy with unique 6p deletion. A. Saba.

1042W ‡ *ARID4A* de novo variants identified by exome sequencing among individuals with neurodevelopmental disorders. K.G. Monaghan.

1043T De novo missense variant in *CSNK2A1* can cause intellectual disability, behavioral problem, and dysmorphic features: The first male child with Okur-Chung neurodevelopmental syndrome. M. Akahira-Azuma.

1044F Exome sequencing links cerebellar malformations to known neurodevelopmental disorders. K.A. Aldinger.

1045W *De novo* variants at residue 480 in *FAR1* are associated with an autosomal dominant early-onset neurological disorder. J. Juusola.

1046T Alpha-thalassemia X-linked intellectual disability (ATRX) syndrome in a Colombian patient. A. Paredes.

1047F Mutations in *DDX3X* are a common cause of syndromic intellectual disability. X. Wang.

1048W It does not have to be the whole exome: Mendeliome sequencing increases the diagnostic yield in patients with unexplained intellectual disability by 30%. A. Rump.

1049T Exploring the therapeutic potential of CRISPR/Cas9 technology for the treatment of MeCP2 duplication syndrome. E. Maino.

1050F A 10q23.31 microduplication is associated to autosomal dominant primary microcephaly. D. Oliveira.

1051W A missense mutation in the CRBN gene that segregates with intellectual disability and self-mutilating behaviour in a consanguineous Saudi family. A. Sheereen.

1052T Novel *AHDC1* mutations cause intellectual disability and developmental delay. Y. Tsurusaki.

1053F Mutation in *OASL* gene causing speech delay and intellectual disability. M. Alfadhel.

1054W Only genotype-first approach permits *BRWD3* mutations' diagnosis. J. Delanne.

1055T Attempts to elucidate role of *ZBTB11* gene as a novel candidate gene in intellectual disability. Z. Fattahi.

1056F Inositol monophosphatase 1 (IMPA1) deficiency and cognitive impairment. T. Figueiredo.

1057W A novel pathogenic variant of the *HECW2* gene in a Japanese boy with global developmental delay, hypotonia, and short stature. M. Minatogawa.

1058T *De novo* LoF mutations in *MED12* cause a syndromic form of X-linked ID in females. D.L. Polla.

1059F A novel missense variant in the POMK gene causes Walker-Warburg syndrome. E. Preiksaitiene.

1060W Clinical heterogeneity in Renpenning Syndrome patients due to c.459_462del mutation in PQBP1 gene: A case report. J. Rojas.

1061T Intellectual disability and macrocephaly in three sisters from a consanguineous marriage with a novel *SZT2* homozygous mutation. C. Trujillo.

1062F The phenotypic spectrum of Xia-Gibbs Syndrome. Y. Jiang.

1063W Updating penetrance estimates for deletion and duplication syndromes with variable phenotypic manifestation. J.W. Ahn.

1064T Exome Pool-Seq in neurodevelopmental disorders. C. Zweier.

1065F Novel mutation in *ARHGEF9* associated with developmental delay and seizures. E. Fattakhov.

1066W Is incontinentia pigmenti a genetic male disease? F. Fusco.

1067T A new X-linked form of syndromic intellectual disability on Xp11.22. D.A. Scott.

1068F Family-based whole exome sequencing for identifying novel variants in primary myopathies. J. Hwang.

1069W ‡ Engineering tissue specific delivery of enzymes for lysosomal disease treatment. K. Cygnar.

1070T Mutational spectrum of Duchene Muscular Dystrophy in Colombian patients. P. Ayala-Ramírez.

1071F A combination of capture-based high-coverage NGS and WES analysis uncovers potential deleterious variants in the *NARS2* gene expanding the phenotypical spectrum of combined oxidative phosphorylation deficiencies. Y. Wang.

1072W Adenosylcobalamin synthesis in cultured fibroblasts from patients with isolated methylmalonic aciduria. D. Watkins.

1073T A zebrafish *mut*^{0} model recapitulates key aspects of severe methylmalonic acidemia. K.T. Ellis.

1074F Novel biallelic mutations in the *PNPT1* gene encoding a mitochondrial-RNA-import proteinPNPase cause delayed myelination. A. Kikuchi.

1075W *RMND1*-related mitochondrial disease: Phenotypic delineation of four patients including renal manifestations. N.T. Le.

1076T Identification of the genetic causes of mitochondrial oxidative phosphorylation (OXPHOS) disease. S.C. Lim.

1077F The clinical spectrum of BCS1L mutations: Case report of a novel mutation and review of the literature. J. Priestley.

1078W Severe lactic acidosis, myopathy, and normal mental status in an infant with biallelic *GTPBP3* pathogenic variants. H. Vernon.

1079T Severe leukodystrophy with complete clinical recovery caused by recessive *BOLA3* mutations. C.A. Stutterd.

1080F A viable knockout murine model of *Mmaa* (*cblA*) deficiency provides a platform for microbiome manipulations. A.F. Lesser.

1081W ‡ Identification and validation of new hepatic biomarkers in methylmalonic acidemia (MMA). I. Manoli.

1082T Identification of large effect variants underlying non-syndromic MA in families segregating the disease. G.A. Arenas-Perez.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

1083F DFNA5 and the infamous skipping of exon 8. K.T. Booth.

1084W Genetics mutation: A novel frameshift mutation in the *USH1G* gene in an Iranian patient with USHER syndrome. F. Tabei.

1085T ‡ Leveraging consanguinity in inherited retinal diseases uncovers missing genetic variation: Rare novel disease genes and a multitude of novel variants in known disease genes. K. Van Schil.

1086F In silico analysis and identification of TYR mutations in a Cypriot family. R. Kalkan.

1087W Genetic therapeutic strategies for Bardet-Biedl syndrome type 1. M. Cring.

1088T Unique mutation spectrums in hearing-impaired Mongolian patients reveal possible migration events and founder effects of common deafness mutations. Y.H. Lin.

1089F Single base deletion in *ATOH7* gene causes abnormal eye development in a consanguineous family. M. Ansar.

1090W Molecular genetics of the Usher syndrome in Saudi Arabia: Identification of known and novel mutations by homozygosity mapping and next generation sequencing. K. Ramzan.

1091T *SLC6A6*: Nutritional therapeutic potential of a novel autosomal recessive gene for progressive retinal degeneration and cardiomyopathy. E. Ranza.

1092F Genetic causes of CHARGE syndrome identified by whole exome sequencing. D.M. Martin.

1093W Novel mutations underlying sensorineural hearing loss in Brazil. K. Lezirovitz.

1094T Unexpected difficulties in discovery of genes involved in hearing loss. S. Naz.

1095F Investigation of novel genetic bases for strabismus using linkage analysis and next-generation sequencing. J. Jurgens.

1096W Linkage analysis and whole genome sequencing analysis in familial isolated strabismus. X. Ye.

1097T A novel homozygous deletion in last exon of CYP1B1 gene cause primary congenital glaucoma in an Iranian female patient. M. Noruzinia.

1098F Identification of novel mutations of *POU3F4* in seven Chinese families with X-linked nonsyndromic hearing loss. H. Yuan.

1099W Next generation sequencing of three families with severe keratoconus identifies putative disease-causing variants. S.E.M. Lucas.

1100T Identification of novel candidate genes for recessive visual impairment by analyzing 132 consanguineous families. M. Ansar.

1101F ‡ Genomic analysis of inherited hearing loss in the Palestinian population. A.I. Abu-Rayyan.

1102W Siblings with Perralut syndrome and *LARS2* mutation who presented with neurologic abnormalities. R. Kosaki.

1103T A homozygous truncating variant of *KCNE1* (p.Tyr46*) associated with deafness in the absence of a long QT interval reveals a novel genotype-phenotype correlation. R. Faridi.

1104F Variation in *MERTK* in patients with retinal dystrophy. C. Jespersgaard.

1105W The NIH Oculocutaneous Albinism Natural History Study. D.R. Adams.

1106T Novel *PXDN* mutations cause microphthalmia and anterior segment dysgenesis. N. Chassaing.

1107F Comprehensive analysis of CNVs in patients with congenital eye malformations by targeted next generation sequencing. M. Corton.

1108W ‡ Genetic characteristics of an international large cohort with Stargardt disease: The progression of atrophy secondary to Stargardt disease (ProgStar) study. K. Fujinami.

1109T First systematic molecular analysis of 45 Greek patients with retinal dystrophies by next generation sequencing reveals 21 novel mutations in 30 genes and establishes a wide spectrum of distinct retinal degenerative diseases. S. Kamakari.

1110F Novel *RS1* gene mutations of X-linked retinoschysis Lithuanian patients. V. Kucinskas.

1111W Genetic analysis of children and families with heritable retinal dystrophies in Costa Rica: Identification of an X-linked mutation. D.J. Wolff.

1112T Unclassifiable brachydactyly (brachydactly E+A2) due to a novel missense mutation in *IHH*. H. Numabe.

1113F A novel missense variant in *IRF6* gene implicated in causing Van der Woude syndrome. N. AlDhaheri.

1114W Origin of *EIF4A3* pathogenic expansion, the causative mechanism of a craniofacial syndrome (RCPS). G.S.P. Hsia.

1115T Craniosynostosis: Expanding the phenotype of 3 rare syndromes. E.H. Zackai.

1116F Disorders of sexual development in genetics pediatrics. Three different ambiguous genitalia cases report from Hospital Para El Nino Poblano, México. J. Aparicio.

1117W Microdeletion of 17q21.31 causes a novel malformation syndrome. K. Kurosawa.

1118T Phenotypic and genotypic spectrum in Richieri-Costa-Pereira syndrome. D. Bertola.

1119F Combination of *UBR1* and *UBR5* mutations in a severe form of Johanson-Blizzard Syndrome with total agenesis of lateral nasal process and situs inversus. A. Cetinkaya.

1120W *SHOX* duplication in a Kabuki syndrome patient: A possible effect on clinical phenotype. A.P. Marques-de-Faria.

1121T A novel mutation in PDE3A gene in a 7-year-old female patient with dysmorphic features, developmental delay, short stature, and unilateral brachydactyly without high blood pressure. A. Alali.

1122F A unique contiguous gene syndrome encompassing Potocki-Shaeffer syndrome and aniridia. G. Delplancq.

1123W Novel pathogenic variants of *EP300* in two Japanese patients with Rubinstein-Taybi syndrome type 2. T. Kaname.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1124T Identification of *AFF4* missense mutation in a girl with Cornelia de Lange syndrome (CdLS) like phenotype and obesity. E. Nishi.

1125F A novel genetic syndrome with *RAB11B* mutation. N. Okamoto.

1126W A case report of novel mutation in NSD1 gene, which causes Sotos Syndrome. J. Prieto.

1127T *SATB2*-associated syndrome: Differential diagnosis and genotype-phenotype correlations by detailed facial dysmorphism analysis using facial recognition technology in 38 individuals. Y. Zarate.

1128F Noonan-like syndrome with loose anagen hair: Expanding the phenotype. A. Belonis.

1129W A Japanese sporadic case of Adams-Oliver syndrome with a novel missense variant in *DLL4*. M. Nagasaka.

1130T Redefining phenotypic spectrum of constitutional *CDK13* mutations: Three patients without cardiac defects. T. Uehara.

1131F *IRF6* mutational screening in a sample of Mexican patients with non syndromic cleft lip palate. J. Velazquez.

1132W ‡ Urine-derived podocytes-like cells: From a diagnostic to a CRISPR/Cas9 gene therapy perspective in Alport syndrome. A.M. Pinto.

1133T The expansion of heterotopic bone in fibrodysplasia ossificans progressiva is Activin A-dependent. A.N. Economides.

1134F Diamond Blackfan "anemia" due to *RPL26* mutation in a family with radial ray and congenital anomalies. L. Mehta.

1135W A novel genetic disorder characterized by severe developmental delay and dysmorphism, recurrent pancreatitis, and organomegaly. M. Morimoto.

1136T A growing need for reverse clinical genomics: Demonstrated by phenotypic characterization of CDK13-related disorders. B. Bostwick.

1137F Phenotypic variability in a cohort of patients with VACTER-L association. O.M. Moreno.

1138W ‡ Truncating *MAGEL2* mutations produce fetal lethality in mice and may recapitulate pathogenesis of Schaaf-Yang syndrome. Y. Negishi.

1139T Case report Kleefstra syndrome in a Colombian patient. A. Nova.

1140F Acid ceramidase deficiency (Farber disease): A qualitative research study documenting the clinical impact of symptoms in a diverse population of patients and caregivers. A. Solyom.

1141W Natural history study design including retrospective and prospective components to address the lack of clinical data on acid ceramidase deficiency presenting as Farber disease. D. Tetzl.

1142T Rare syndrome: First reported Egyptian sibs with 3MC(1) syndrome and detection of novel *MASP1* gene mutation in the family. A.N. Khalaf.

1143F Expanding the phenotype of *DST*-related disorder: A case report suggesting a genotype/phenotype correlation. M. Pinelli.

1144W Genomic approaches to investigate children born small for gestational age (SGA) without catch up-growth. B.L. Freire.

1145T Genetic diagnosis and clinical characteristic reviews in neonate patients of *KMT2D* gene mutation caused Kabuki syndrome. B. Wu.

1146F ‡ *De novo* truncating mutations in X-linked *MED12* are associated with Hardikar syndrome in females. D. Li.

1147W A novel homozygous mutation in MMP21 is associated with heterotaxia and cardiac defects. L. Aspit.

1148T Exome-first approach identified two novel indels and gene deletions in Mowat-Wilson Syndrome cases previously diagnosed as clinical Angelman syndrome. F. Gosso.

1149F Homozygous truncating variants in *TMCO1* contribute to a spectrum of craniofacial, skeletal and neurodevelopmental abnormalities. J. Ji.

1150W Robinow syndrome in an infant with multiple anomalies due to *DVL3* mutation: A lesson in detailed clues. B. Keena.

1151T Frontonasal dysplasia with facial skin polyps: From Pai syndrome to differential diagnosis through a series of 25 patients. D. Lehalle.

1152F Treacher Collins syndrome 1: Extreme manifestations. C. McDougall.

1153W Triple diagnosis by whole exome sequencing. Z. Yüksel.

1154T Ovotesticular DSD associated with a pathogenic mutation in the BMP15 gene. S. Albanyan.

1155F ‡ A genotype-first approach identifies genes contributing to variable phenotypic presentations in a Smith-Magenis Syndrome cohort. C. Zhang.

1156W A novel *FBXO28* frameshift in a patient with the predominant features of 1q41-q42 deletion syndrome: A case for haploinsufficiency and primary pathogenicity. C.D. Balak.

1157T Exome sequencing in a family with autosomal recessive amelogenesis imperfecta. G. Chavarria-Soley.

1158F ‡ *De novo* coding and noncoding variants in novel disease genes account for a significant fraction of isolated and complex congenital diaphragmatic hernia. H. Qi.

1159W Clinical and molecular phenotypes of Coffin-Siris syndrome among UAE population. F. Al Ali.

1160T Twelve novel *GUSB* mutations and genotype-phenotype correlation in mucopolysaccharidosis VII (MPS VII) patients. Q. Abu Ali.

1161F Clinical exome data analysis and novel variant identification for MPS VII disease in GUSB gene. A. Bhattacherjee.

1162W Mutations in the condensin II component NCAPG2 cause autosomal recessive neurodevelopmental syndrome. T.N. Khan.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

1163T ‡ Frequency of germline pathogenic variation in *NF1* and eight other RASopathy genes in the Exome Aggregation Consortium (ExAC) database: A pilot study. A. Pemov.

1164F Enhancement of hepatic autophagy increases ureagenesis and protects against hyperammonemia in a mouse model of ornithine transcarbamylase deficiency and other models of secondary hyperammonemia. L. Soria.

1165W Search for the mutation causing the ThoracoAbdominal Syndrome (TAS), an X-linked dominant disorder. P. Majdalani.

1166T Germline mutations associated with polycomb repressive complex 2 cause Weaver syndrome. E. Imagawa.

1167F Functional analysis of a novel *TUBA1A* gene mutation associated with cerebral dysgenesis and cleft palate. R. Peretz.

1168W A prospective study of natural history, physiology, and biochemistry of propionic acidemia. O.A. Shchelochkov.

1169T SOPH syndrome: Multisystem disorder with facial dysmorphism, skeletal dysplasia, episodic liver failure, immune dysfunction and intellectual disability. A. Yadav.

1170F ‡ The genetic architecture of Bardet Biedl Syndrome. M. Kousi.

1171W Clinical and genetic characteristics of seven patients with Floating-Harbor syndrome. P. Castro.

1172T ‡ An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease-gene discovery. A. Haghighi.

1173F ‡ Insights from applying ClinGen framework in evaluating the clinical validity of 30 published studies of gene-disease relationship of monogenic disorders. R. Attali.

1174W ‡ Evaluating the evidence available for associating genes of unknown significance (GUS) with disease phenotypes: Review of 100 studies. S. Tzur.

1175T Oral cavity findings in *A2ML1*-related otitis media. R.L.P. Santos-Cortez.

1176F ‡ Deep phenotyping for patients by patients: A lay-friendly version of the Human Phenotype Ontology. M.A. Haendel.

1177W Sporadic, isolated Fanconi syndrome due to a mutation of EHHADH. E.G. Seaby.

1178T A combinatorial approach for the selection of novel bioactive peptides. R.R. Handley.

1179F ‡ Mouse models of human disease: How mouse model data can provide mechanistic insight into human disease etiology and development of therapeutics. M. Tomczuk.

1180W Challenges in translating pharmacogenetic variants from Illumina's Multi-ethnic Genotyping array into clinical practice. N. Rafaels.

Bioinformatics and Computational Approaches

1181T The ET-HPN highlights a potential ALS-related disease cluster. B.E. Graham.

1182F The system biology concept applied to a secondary analysis of Body Mass Index Genome Wide Association Study (BMI GWAS) data. E. Cirillo.

1183W Causal gene prediction from type 2 diabetes susceptibility loci through integration of genetic association and functional annotation data. J. Fernandez-Tajes.

1184T ‡ LabWAS: A catalog of real-world associations between genetic variants and lab values. J.A. Goldstein.

1185F Genome-wide prediction of susceptibility loci for complex diseases based on regulatory data. T. Yang.

1186W The Type 2 Diabetes Knowledge Portal: Clearing a path from genetic associations to disease biology. B. Alexander.

1187T Retrospective electronic medical record analysis to identify patients at risk of hypophosphatasia. C. Peroutka.

1188F Evaluation of genetic sequencing to improve newborn screening for VLCAD disease. B. Cai.

1189W ‡ Using automatic adipose measures from electronic health record based imaging data for discovery. E.D.K. Cha.

1190T *In-silico* characterization of cell-type composition in adipose tissue: Implications for 'omic analyses and associations to adiposity measures. C.A. Glastonbury.

1191F Non-additive SNP effect discovery from GWAS with LAMP. J. Sese.

1192W ‡ The grid-interpolation algorithm: A novel approach for fast and efficient mixed model analysis of high-dimensional phenotype data. J.R. O'Connell.

1193T Identification of genes alternatively spliced in HIV-infected CD4+ T-cells. S. Han.

1194F Depression and mental illness affect pre and post multiple sclerosis diagnosis. C. Gardner.

1195W A haplotype assembly workflow for HLA and KIR typing from next-generation sequencing data. S. Tian.

1196T A high-throughput deep sequencing approach for CRISPR off-target assessment in therapeutic genome editing applications. A. Ajetunmobi.

1197F High throughput BCR sequencing of the thymus and blood B cell repertoire in myasthenia gravis patients pre and post thymectomy. R. Jiang.

1198W PheWAS and permutation analyses indicated involvement of the *CLEC16A* locus in immune-related phenotypes. M.E. March.

1199T Understanding chronic fatigue syndrome using immune cell specific RNA-seq in a time series after an exercise perturbation. P. Comella.

1200F ‡ Finding NEMO: *De novo* mutation detection in the IKBKG gene. Z. Deng.

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1201W Exome sequencing by NimbleGen kit is not suitable for *SOX2* and *SOX3* molecular screening due to bald spot. A.F.F. Benedetti.

1202T Hunting rare variants in chronic kidney disease. S.R. Cameron-Christie.

1203F BREATH: An open-access database and website of normal human and mouse lung development generated by the Molecular Atlas of Lung Development Program (LungMAP). R.F. Clark.

1204W An unbiased, genetic-data-driven benchmarking strategy for gene and variant prioritization algorithms. R. Fine.

1205T The gene expression signature associated with rheumatoid arthritis is altered during pregnancy. D. Jawaheer.

1206F The accuracy of whole genome sequencing and the Platypus variant caller in identifying genetic variation within the structurally challenging epidermal differentiation complex. C. Malley.

1207W Somatic mutation hunting: The search for the genetic architecture of linear localized scleroderma. R.G. Higgins.

1208T *In silico* screening for potentially aberrant novel essential splice sites (PANESS) created by single nucleotide variants (SNVs) in the human genome. C.C. Bruels.

1209F Computational prediction and molecular validation of novel therapeutic targets for potent splicing modulators. D. Gao.

1210W Alterations of transcriptome landscaping in head trauma-related human brain disorders. H. Cho.

1211T NIA Genetics of Alzheimer's Disease Data Storage Site (NIAGADS) Genomics Database. E. Greenfest-Allen.

1212F Integrated causal network analysis of genomic and epigenomic data. Z.X. Hu.

1213W FGWAS: Functional Genome Wide Association Analysis. C. Huang.

1214T NIA Genetics of Alzheimer's Disease Data Storage Site (NIAGADS): Update 2017. H. Lin.

1215F The Alzheimer's Disease Sequencing Project (ADSP) data update 2017. Y. Zhao.

1216W Personalized and cell-specific pathway score computations from risk alleles and regulatory information in 2370 subjects with multiple sclerosis. L.R. Madireddy.

1217T ‡ Omics-based machine learning modeling of monogenic neurological diseases. J.A. Botía.

1218F Identification of tolerated reading-frame changes induced by stop-lost and frame-shift variants in Alzheimer's disease. M. Butkiewicz.

1219W Missense variant interpretation based on mutational burden at analogous amino acid positions across gene family members. E. Perez-Palma.

1220T Evaluation of basic massive parallel sequencing parameters in relation to true/false positivity's findings of rare variants from an isolated population from South-Eastern Moravia in the Czech Republic with high incidence of Parkinsonism. R. Vodicka.

1221F A statistical inference framework to improve functional prediction of missense variants in neurodevelopmental disorders. J. Du.

1222W Assessment of lesion-associated gene and variant pathogenicity in focal human epilepsies. L.M. Neupert.

1223T Accurate identification of de novo structural variants in a trio using a reference agnostic, rapidly queryable format to reduce the proportion of unsolved cases. S.N. Shekar.

1224F A single-cell transcriptome atlas of mouse hypothalamus: Identification and characterization of high-fat diet and GLP-1 responding arcuate nucleus cell types. P.N. Timshel.

1225W The GCAD workflow for processing 5000 whole genomes and 11,000 whole exomes from the Alzheimer's Disease Sequencing Project using Amazon cloud. Y.-F. Chou.

1226T ‡ Neuron-squared (N^2): An industrial-scale iPSC disease-modeling project for neuropsychiatric disorders. R. Randhawa.

1227F ‡ Towards translating genetic findings of polygenic diseases to personalized drug development: Proof-of-concept study for drug combinations to target multiple genes. I.S. Vlachos.

1228W SV^2: Accurate structural variation genotyping and de novo mutation from whole genomes. D. Antaki.

1229T Novel pathway transcriptomics method greatly increases detection of molecular pathways associated with the trait. C. Chatzinakos.

1230F Gravity: A tool to analyze genetic variants in individuals using gene networks. F. Cliquet.

1231W GWAS-based machine learning approach to predict duloxetine response in major depressive disorder. M. Maciukiewicz.

1232T The Registry of Candidate Regulatory Elements: Integrating human and mouse epigenomic data to fine map and annotate genetic variants. J. Moore.

1233F Bayesian multivariate analysis of RNA sequencing data to identify brain-specific protein-protein interactions. S. Muller.

1234W Tissue-specific gene expression inference. K. Vervier.

1235T Augmenting multi-ethnic image signals to enhance schizophrenia prediction. H. Qin.

1236F A statistical framework of mapping risk genes from *de novo* mutations in whole-genome sequencing studies. Y. Liu.

1237W ‡ Comparison of different approaches to detect CNV from SNP genotyping array and whole-exome sequencing. B. Chaumette.

1238T Exomerate: A machine-learning approach to identify high-confidence CNVs from exome sequencing data. V. Pounraja.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

1239F Improving pathogenicity prediction of structural variation in neurodevelopmental disorders: A machine learning approach. P.T. Tandon.

1240W Biological pathways and drug gene-sets: Analysis and visualization. H.A. Gaspar.

1241T A paradigm for using human GWAS summary statistics to accurately test gene expression correlation networks derived from model organisms or post-mortem tissues. S. Bacanu.

1242F Unprogrammed presentation number

1243W Ultra-accurate complex disorder prediction: Case study of neurodevelopmental disorders. L. Huynh.

1244T Sample-specific background correction leveraging vast historical patient cohort maximizes sensitivity of noninvasive prenatal screening. D. Muzzey.

1245F Identification of homozygous deletions from exome sequencing data. M. Kohda.

1246W Comparison of pipelines and databases for detection and annotation of mitochondrial variants from whole-exome sequencing data. J.C. Tsai.

1247T Assessment of the performance of splicing predictors at non-canonical intronic sites and implications for variant classification. D.H. Tran.

1248F Chromosomal sequencing analysis: Assessing the performance of copy number variant analysis by next generation sequencing compared to traditional clinical microarray analysis. S.P. Strom.

1249W A Bayesian network approach for *de novo* variant calling and its application on rare Mendelian disorders. A. Toth-Petroczy.

1250T MiRMed: Therapeutic recommendation and drug repositioning framework for rare and common diseases using micro-RNA signatures. K. Shameer.

1251F ‡ Examining age, tissue, and genetic effects on RNA splicing with allele-specific resolution in a diverse mouse population. D.A. Skelly.

1252W Exomic variants of an elderly cohort of Brazilians: ABraOM database. M.S. Naslavsky.

1253T Age-related changes in abundance of extracellular RNA in human serum. D.F. Dluzen.

1254F Mixture models reveal multiple positional bias types in RNA-seq data and lead to accurate transcript concentration estimates. J. Aleksejeva.

1255W ‡ A novel clustering model for droplet-based single cell transcriptomic data. W. Chen.

1256T CookHLA: Accurate, efficient, and memory-efficient HLA imputation. S. Cook.

1257F A novel Word2vec based tool to estimate semantic similarity of genes by using gene ontology terms. D. Duong.

1258W Systematic assessment of joint genotyping. U. Evani.

1259T Improving sequence read mapping and allele calling for the polymorphic *PRDM9* gene using a reference graph approach. H. Gibling.

1260F Comprehensive benchmarking of error correction methods for next generation sequencing via unique molecular identifiers. B.L. Hill.

1261W ART_PacBio: A fast and accurate simulator for PacBio sequencing platforms. W. Huang.

1262T A high-throughput pipeline for genotyping within primary health care to enable precision prevention. S. Jai Kumar Ahuja.

1263F Reducing reference bias in whole-genome variant calling using reference graphs. M. Kallberg.

1264W Integrating networks and comparative genomics reveals retroelement proliferation dynamics. B.A. Knisbacher.

1265T Limits of indel detection using CLC alignment and variant calling. T. Koganti.

1266F Comparison of short tandem repeat estimation methods with various conditions. K. Kojima.

1267W A graph method for population genotyping of structural variants. P. Krusche.

1268T Accurate radiation biodosimetry through automation of metaphase cell image selection and chromosome segmentation. Y. Li.

1269F WGSA 07: Updated annotation pipeline for human genome sequencing studies. X. Liu.

1270W PoolHap2: Inferring within-host haplotype frequencies from pathogen next-generation sequencing data. Q. Long.

1271T LRSim: A linked reads simulator generating insights for better genome partitioning. R. Luo.

1272F Genetic correlations as a tool for finding new biomarkers for female reproductive health phenotypes. R. Magi.

1273W Privacy preserving Fisher's exact test for GWAS. K. Misawa.

1274T The predictive power of RNA-Seq: Modeling blood chemistry and hematology test results. M. Naymik.

1275F Quality comparison of whole exome sequencing data by different types of specimen. K. Park.

1276W ClinGen Pathogenicity Calculator 2.0: New features and lessons learned from the data mining of 2,400 variant interpretations according to ACMG/AMP guidelines. R. Patel.

1277T ‡ *Indexcov*: Whole-genome coverage in <1 second per BAM. B.S. Pedersen.

1278F Variants impact on splicing regulatory element determination pipeline. N. Prodduturi.

1279W Working through heterozygous variants in NGS screenings: The DOMINO algorithm allows recognizing potentially dominant genes by machine-learning. M. Quinodoz.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1280T Precision phenotyping of sample biopsies of kidney transplants by leveraging public microarray data and cell types enrichment analysis. D. Rychkov.

1281F Cloud-based quality measurement of whole-genome cohorts. W. Salerno.

1282W Ultra-rapid detection of identity-by-descent tracts for biobank-scale inference. R. Shemirani.

1283T Unprogrammed presentation number

1284F Collective feature selection to identify important variables for epistatic interactions. S. Verma.

1285W PipelineDog: A simple and flexible graphic pipeline construction and maintenance tool. J. Xing.

1286T ‡ Bayesian multiple eQTL detection with control for population structure and sample relatedness. B. Zeng.

1287F ‡ Robust and accurate estimation of DNA sample contamination agnostic to genetic ancestry. F. Zhang.

1288W Predicting exposure to ionizing radiation by biochemically-inspired genomic machine learning. J.Z.L. Zhao.

1289T Leveraging allele-specific expression to refine fine-mapping for eQTL studies. J. Zou.

1290F The role of CTCF and cohesin complex in chromatin looping and higher-order organization of human genome. D. Plewczynski.

1291W Weighted gene co-expression network analysis using peripheral blood of patients with 22q11.2 deletion syndrome. A.G. Dantas.

1292T Showing your work: Combining genetic variant interpretations with evidence to enable reanalysis and reuse. B.C. Powell.

1293F Supervised enhancer prediction with epigenetic pattern recognition and targeted validation across organisms. M. Gu.

1294W Novel high-resolution multi-ethnic HLA imputation reference panels constructed based on high-coverage whole-genome sequencing data. Y. Luo.

1295T Detecting copy number variants in 200,000 individuals: The Department of Veterans Affairs Million Veterans Program (MVP). M. Li.

1296F A graph-based Arab reference genome using whole read overlap assembly. Y. Mokrab.

1297W DUP-OE: A new tool to discovery the origin and expansion of duplication. X. Zhuang.

1298T Illuminating the Druggable Genome (IDG): An NIH Common Fund program. J. Baker.

1299F Enhancing discoverability and reusability of 1.2 million human genomic datasets. M. Corpas.

1300W Semantic web technology accelerates integration of genetic and phenotypic information in biomedical databases. T. Katayama.

1301T Controlled-access databases for genetic and phenotypic human data in Japan. Y. Kodama.

1302F DASHR 2.0: Database of small non-coding RNAs in normal human tissues and cell types. P.P. Kuksa.

1303W Predicting genetic ancestry for 805,482 patients using clinical data from electronic health records. N. Tatonetti.

1304T Improved phenotype-based computational methods to support diagnosis of genetic disease. J. Chen.

1305F ‡ Quantification of transplant-derived circulating cell-free DNA in absence of a donor genotype. E. Sharon.

1306W Copy number variation detection and variant curation improves interpretation of exomes for inborn errors of metabolism. S.E. Brenner.

1307T QRank: A novel quantile regression tool for eQTL discovery. X. Song.

1308F A novel set of 96 indices for accurate sample demultiplexing on Illumina platforms. B. Carrion.

1309W ‡ Gene-based tests using imputed genotype dosages showed increased statistical power than using best-guessed genotypes. M. Hwang.

1310T User-driven prioritisation of study addition to the NHGRI-EBI Genome-Wide Association Study (GWAS) Catalog. J.A.L. MacArthur.

1311F GRASP v3: An updated GWAS catalog and contrast to similar catalogs. B.A.T. Rodriguez.

1312W Reinterpreting genetic studies of kidney disease with integrated analysis of kidney-cell specific chromatin accessibility and transcriptomics data. K. Sieber.

1313T Genomic multilateration. K. Kim.

1314F Estimating incidence of inborn errors of metabolism from the frequency of variants in general population. I. Mihalek.

1315W A novel compuational strategy for DNA methylation prediction. F. Yu.

1316T Scaling workflows for growing microbiome applications. J. Lai.

1317F Deriving disease signatures using data driven appraches on microbiome. K. Poorey.

1318W ‡ Noninvasive reconstruction of fetal methylome by sequencing of maternal plasma DNA. K. Sun.

1319T Stargazer: A software tool for calling star alleles from next-generation sequencing data using *CYP2D6* as a model. S. Lee.

1320F Construction of Japanere reference genome and Japanese reference panel of thousands of individuals in Tohoku Medical Megabank Project. M. Nagasaki.

1321W SPACE, a tool for dynamic exploration of principal component analyses. N.D. Berkowitz.

1322T ‡ Quickly determining subject ancestries in large datasets using genotypes of dbGaP fingerprint SNPs. Y. Jin.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

1323F PubCases: A diagnosis assistant tool for rare diseases based on disease-phenotype associations extracted from published case reports. T. Fujiwara.

1324W ‡ MVP: A deep neural network method of predicting pathogenicity of missense variants enables novel genetic discoveries. Y. Shen.

1325T Search Candidate Regulatory Elements by ENCODE (SCREEN): A web-based tool for visualizing genomic annotations. H.E. Pratt.

1326F SinCCE (Single-Cell Cluster Ensemble): Cluster ensemble for single-cell RNA-seq data. Y. Yang.

1327W Improving quality of variant calling by integrating whole genome and whole exome sequencing from same samples. X. Li.

1328T The Mega2R suite of R packages: Tools for accessing and processing common genetic data formats in R. D.E. Weeks.

1329F An iterative strategy to improve the power of epistasis analysis. J. Wen.

1330W Telomere length estimation and analysis on large scale whole-genome sequencing data. M.A. Taub.

1331T 1000 Genomes Project data and additional openly consented data resources can be accessed via the International Genome Sample Resource (IGSR). S. Fairley.

1332F An alignment-based approach for sensitively detecting SVs using optical maps data. X. Fan.

1333W Leveraging unique molecular identifiers to improve low-frequency variant calling in QIASeq V3 panels. B. Vilhjalmsson.

1334T Multi-sample isoform quantification from RNA-seq for known and novel transcripts. A.E. Byrnes.

1335F PennSeq2: Efficient quantification of isoform-specific gene expression from RNA-seq data using weighted likelihood method. Y. Hu.

1336W VariantFX: An open-source framework for aggregation, visualisation and analysis of Mendelian disease cohort sequence data. M. Ahmad.

1337T NGS based CNV calling in a clinical diagnostic setting. D. Becker.

1338F A generalized non-parametric genotype caller using an EM-like algorithm. T.A. Benaglia.

1339W Phenotype-specific information improves prediction of functional impact for noncoding variants. C. Bodea.

1340T Faster genotype phasing and imputation for large-scale data. B.L. Browning.

1341F Fast and easy pipeline for quality control and assurance for GWAS. T.M. Brunetti.

1342W An empirical strategy to screen markers on case-control genomic studies. B.S. Carvalho.

1343T Accurate quantification of allele-specific methylation from genetically diverse population. K. Choi.

1344F Development of a framework for laboratory information tracking in support of research and clinical operations. B. Craig.

1345W A new method for analysis of sequencing reads without reference genomes. H. Dai.

1346T Gene identifiers: More than just a number. P. Denny.

1347F Cohort based precise interpretation algorithms of copy number variants detected by exome sequencing on individuals of severe disorders. X. Dong.

1348W Fabric Genomics' Opal variant interpretation platform enables rapid, whole genome analysis turnaround in under an hour. A.P. Fejes.

1349T Minerva & Me: Public participation in research: Crowdsourcing for computational phenotyping method development. M. Ferlaino.

1350F Prediction of gain-of-function and loss-of-function mutations using Combined Annotation Dependent Depletion (CADD). A. Ghosh.

1351W Efficient pipeline for whole genome simulation and summary statistic calculation with flexible demographic models. A.L. Gladstein.

1352T The European Variation Archive: A repository for short and structural genomic variation. C.Y. Gonzalez Garcia.

1353F The analysis of negative selection with heterogeneous Mendelian models in coding, as well as noncoding regions for cohorts of diverse undiagnosed diseases. F. Gu.

1354W Graphtyper: Population-scale genotyping using pangenome graphs. B. Halldorsson.

1355T PathwayMatcher: Direct mapping of omics data to the Reactome pathway knowledgebase. L.F. Hernández Sánchez.

1356F Development and organization of genomics metadata by ENCODE. B. Hitz.

1357W Compare HLA typing by next generation sequence methods: An example in Taiwan Biobank database. C. Hsiung.

1358T Target Gene Notebook: Connecting genetics and drug discovery through enabling computational and logistical tools. J. Hutz.

1359F ‡ A graph-based pipeline to evaluate common structural variations based on haplotypes and reassembly. S. Ji.

1360W Rapid whole-genome annotation and search in the cloud: SeqAnt enables easy identification of alleles for traits of interest. A. Kotlar.

1361T ‡ High-performance whole genome sequence variant analysis in the TOPMed project using cloud environments. R. Kuraisa.

1362F Improved access to variant-centric data in ClinVar. M.J. Landrum.

1363W Bioinformatics and Elasticsearch: The perfect combination to unify and visualize life sciences massive data. M. Leclercq.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1364T A graph remapping framework for in silico adjudication of SNVs, indels, and structural genetic variants from genetic sequencing data. D.H. Lee.

1365F SAV (Sparse Allele Vectors): Efficient variant file format that scales to analysis of millions of deep genomes. J. LeFaive.

1366W Pathview Web: User friendly pathway visualization and data integration. W. Luo.

1367T Optimal workflow for next generation sequencing data processing using existing technology. J.E. Martin.

1368F Harnessing the power of linked data: Using semantic annotations on pediatric and adult datasets to enable cross-search. M. Mattioni.

1369W Representing the human genome with synthetic spike-in controls. T. Mercer.

1370T Use of a metadata-driven architecture for tools and techniques to enhance methods for data sharing and facilitate meta-analysis for genetic association. M.C. O'Leary.

1371F Improving SNP array copy-number variant calling using site-specific variance models and windowed intensity normalization. T. Poterba.

1372W LUBA: A software toolbox for efficiently manipulating and analyzing NGS data. A. Ryutov.

1373T Hail: Scaling statistical genetics to tens of thousands of whole genomes. C. Seed.

1374F IMSindel: An accurate intermediate-size indel detection tool incorporating *de novo* assembly and gapped global-local alignment into split reads. D. Shigemizu.

1375W ENCODE Portal: A cohesive collection of genomic assays towards discovery of functional elements. C.A. Sloan.

1376T Cloud computing environment for hosting federated genomic databases. W. Souza.

1377F The ENCODE annotation pipeline: Repeatable software tools for ChIP-seq, RNA-seq, DNase-seq, and whole-genome bisulfite experiments. J.S. Strattan.

1378W Quantitative assessment of the feasibility of using whole-genome sequencing data at common single nucleotide polymorphism positions to reproduce high-confidence genotype calling and copy number variation detection results from SNP microarray data. N.S. Ten.

1379T GA4GH Genomic Beacon Network: Security challenges and mitigation. S. Ur-Rehman.

1380F UDPICS, a laboratory-medical informatics system for improving translational research in the NIH Undiagnosed Diseases Program. Z.M. Valivullah.

1381W Improving clinical exome interpretation using lab internal cohort data. C. Wu.

1382T Integrated breakpoint analysis and structural variation detection using sequencing reads from multiple sequencing technologies for an Ashkenazi trio. C. Xiao.

1383F ‡ Exploiting the co-localization of trait-associated SNPs and eQTLs to identify potential biological mechanisms underlying complex diseases. T. Xu.

1384W ALEC: Amplicon Long-read Error Correction for targeted long-read sequencing. Y. Yang.

1385T An alignment-based method to trim adapter and filtrate low-quality reads from pair-end FASTQ files. R. Ye.

1386F A statistical framework for longitudinal genomic data integration. Y. Zhang.

1387W SOAPgaea: A Hadoop/Spark based computing framework for bioinformatics in big data. Y. Zhang.

1388T Gene set comparison analysis using both first and second moment information. L. Zhang.

1389F A new diagnostic platform (Genomic Intelligence®) improves accuracy of whole exome and genome sequencing diagnosis in rare disease. A. Fisher.

1390W Uncovering the genetic architecture of complex traits: A Kalman filter approach. D. Palmer.

1391T Organize and share your bioinformatics analysis with the R package workflowr. J.D. Blischak.

1392F TeraPCA: A fast and scalable method to study genetic variation in tera-scale genotypes. A. Bose.

1393W Performance optimization of a genomic variant store for genotype-phenotype correlation in PhenoTips software. P. Buczkowicz.

1394T ‡ From quantity to quality: A population-based approach for building reference panel imputation. M. Cocca.

1395F A framework for using protein structure specific features to elucidate ambiguous non-synonymous single nucleotide variants. J.B. Jespersen.

1396W Genoppi: A web application for interactive integration of experimental proteomics results with genetic datasets. A. Kim.

1397T A unified web platform for network-based analyses of genomic data. T. Li.

1398F Quantitative transcription factor occupancy across cell types, conditions, and genetic variants. K. Luo.

1399W PHESANT: A tool for performing automated phenome scans in UK Biobank. L.A.C. Millard.

1400T Comparative analysis of methods for discovery of germline copy-number variants from exome data. B.D. O'Fallon.

1401F DNA sequence and quality value accuracy for a new Sanger sequencing instrument. S.J. Schneider.

1402W Genotype array missing variant imputation with 78 batches comprising ~84,000 individuals. I.B. Stanaway.

1403T Graph embedding and visualization of genetic data using paired factor analysis. G.T. Wang.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

1404F Polaris: A collection of 220 publicly available whole genomes for sharing validated structural variants. M.A. Bekritsky.

1405W ‡ CNVs in clinical WGS: Deployment and interpretation for rare and undiagnosed disease. A.M. Gross.

1406T The quantity of detected copy number variation (CNV) deletions substantially increases when coverage of whole-genome sequencing (WGS) data increases from 30x to 91x. Y.S. Huang.

1407F Full-spectrum copy number variation detection by high-throughput DNA sequencing. Y. Jiang.

1408W A clinically validated whole genome pipeline for structural variant detection and analysis. A. Kaplun.

1409T Robust identification of deletions in next generation trio sequence data based on clustering of Mendelian errors. K.B. Manheimer.

1410F Automated parameter tuning for more accurate CNV calling in WES/WGS data. M. Wiewiórka.

1411W Integrative DNA copy number detection and genotyping from sequencing and array-based platforms. Z. Zhou.

1412T CNVs from targeted NGS data: Building a cohort for validation and semiautomatic regression testing in a diagnostic setting. M. Ziegler.

1413F Towards "gold standard" sequence-resolved structural variants in benchmark human trio reference samples. J.M. Zook.

1414W A simple, flexible and EHR-agnostic platform for rapid cohort identification and characterization in a large and diverse biobank environment. F. Mentch.

1415T ClinGen Allele Registry: Linking information about human genetic variation across the web. P. Pawliczek.

1416F Finding possible missed variant candidates through coverage comparison among varying sequencing technologies. H. Fauni.

1417W Diagnostic variant prioritization using a statistical framework for patient genome interpretation. N. Stong.

1418T Insights into the performance of whole-exome sequencing technologies. Y. Yu.

1419F Hepatic eQTL meta-analysis leads to identification of novel *cis*-eQTL in pharmacogenes. A.S. Etheridge.

1420W Detecting sex specific mRNA and miRNA – eQTLs: Insight into sex biased gene regulation. J.J. Shen.

1421T Factorbook: ENCODE ChIP-seq and DNase-seq data visualizer. M. Purcaro.

1422F Prioritizing phenome-wide associations using epigenome fine-mapping. M.D. Ritchie.

1423W New statistical tools to simulate, analyze, and assess the performance of CRISPR regulatory screens. G. McVicker.

1424T CRISPinatoR: A web-based sgRNA design tool that accounts for post-transcriptional influences on protein translation. Y. Yeu.

1425F ‡ Pioneering an efficient migration of 10,000 whole genomes: Catching up with the latest human genome assembly. S. Graf.

1426W Finding associated variants in genome-wide associations studies on multiple traits. L. Gai.

1427T ‡ A map of highly constrained coding regions in the human genome. J. Havrilla.

1428F VIVA: A collaboration tool for processing, storing, exploring, and sharing of next generation sequencing (NGS) data. M. Kohram.

1429W How low can you go? Recommendations for ultra-low input RNA-sequencing. E.A. Tsai.

1430T ‡ Impact of polygenic risk on changes in biomarkers over time due to lifestyle intervention and aging. M. Conomos.

1431F Small RNA-Seq workflow in Strand NGS: Detection of novel small RNAs. H.K. Adil.

1432W ‡ Estimating the impact of allele specific expression on detecting genetic associations. J. Dannemiller.

1433T ‡ Unsupervised pattern discovery in noncoding variants enables identify their potential functional consequences. H. Yang.

1434F Ultra-sensitive profiling of eukaryotic and viral communities residing inside and outside the human body across 300 individuals and 1736 built environments. N. LaPierre.

1435W Investigations of unmapped reads from human exome sequencing. R. Sood.

1436T High-speed mosaicism detection for agnostic genome-wide detection, using the DGX-1 array processor on an NHGRI website for the genetic community. T. Markello.

1437F ‡ A survey of genetic variant frequency in 220,000 Han Chinese individuals. Z. Huang.

1438W PheWeb: Do-it-yourself PheWAS. P. VandeHaar.

1439T A novel method of phenotype similarities of rare-disease patients using gene-pathway-phenotype relationships. S. Ogishima.

1440F PQC: A phenotype checking and tracking tool. N.W. Rayner.

1441W Recombination rate estimation in large-scale genetic data. S. Choi.

1442T Automated abstraction of phenotypes from electronic health records facilitates genetic diagnosis. K. Wang.

1443F Dynamic aggregation of external evidence and standardized manual curation of genetic and experimental evidence in ClinGen's interfaces promote curation quality and consistency. M. Wright.

1444W Effects of filtration on imputation in clusterised variants. C.M. Charon.

1445T Modeling and analysis of RNA structuromes. Z. Ouyang.

1446F Development and validation of computational approaches for GWAS-informed target gene identification. S.K. McFarland.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1447W ‡ OASIS: Omics Analysis, Search and Information System for biological discovery in whole-genome sequence and trans-omics datasets. J.A. Perry.

1448T Leveraging genomic data for Bayesian analysis of high-throughput regulatory functionalization assays. A. Ghazi.

1449F Integrating functional genomics knowledge to construct comprehensive models for complex traits and translational studies. B. Li.

1450W Custom targeted design workflow for next generation sequencing. B. Marosy.

1451T RNASeqFPro, a full processing pipeline for RNA-Seq differential gene expression analysis. M. Pjanic.

1452F ‡ Drug side effects and adverse events are predicted by genetics of their intended targets. P.A. Nguyen.

1453W ‡ PALMER: A novel pre-masking method for detecting mobile element insertions using long-read sequencing technology. W. Zhou.

1454T General validation framework using semi-supervised learning on complex cfDNA clinical assays. K.R. Haas.

1455F Winston: Optimizing parallelized variant calling from large-scale whole genome sequencing data. B. Jew.

1456W Pipeline for DNA-seq analysis: Streamlining of data processing, quick and accurate variant calling, and annotating variants. A. Pal.

1457T A reference haplotype panel for genome-wide imputation of short tandem repeat variants. S. Saini.

3021W Computational pipeline for single nucleus RNA sequencing and its application to human skeletal myotubes. Q. Wang.

Omics Technologies

1458F Hypertriglyceridemia as a presenting sign of HMG-CoA synthase deficiency. E. Conboy.

1459W Characterizing NAFLD/NASH drug targets using metabolomics and genetics. D.M. Waterworth.

1460T Elucidation of the complex metabolic profile of cerebrospinal fluid using an untargeted biochemical profiling assay. L. Hubert.

1461F Infant weight gain trajectories linked to oral microbiome composition. K. Makova.

1462W Integrative analysis of genome, epigenome and transcriptome data from adipose tissue of obesity in Koreans. B.-J. Kim.

1463T RNA sequencing analysis identifies differentially expressed genes in Lymphoblastoid Cell Lines (LCLs) generated from diabetic retinopathy patients. Y. Cho.

1464F Optimizing simultaneous isolation of biomolecules and cells from cryopreserved adipose tissue for omics applications. E.C. Lawrence.

1465W Loss of function variant in *NFKB1* causes autoimmune lymphoproliferative syndrome-like disease. F. Vairo.

1466T Novel human T cell receptor variable gene alleles revealed by long-amplicon TCRβ repertoire sequencing with Ion Torrent. T. Looney.

1467F Transcriptome altered by lytic human cytomegalovirus infection on human foreskin fibroblast cells (HFF) using RNA-seq. H. Li.

1468W Simultaneous digital measurement of protein and mRNA content by massively parallel single cell sequencing to better identify T cell subsets. C. Chang.

1469T ‡ Modulation of Notch pathway in adult mouse airways by antisense oligonucleotides. T.R. Grossman.

1470F The translational utility of metabolomics in the integrative omics era: A case study in asthma. J. Lasky-Su.

1471W Global gene expression patterns in X-autosome balanced translocation patients. A. Di-Battista.

1472T Tau phosphorylation is impacted by rare AD-associated AKAP9 mutations specific to African Americans. T. Ikezu.

1473F Somatic mutations are abundant in focal cortical dysplasia. V.S. Almeida.

1474W Proteomics analysis of the dentate gyrus isolated from two different animal models of mesial temporal lobe epilepsy. A. Morato do Canto.

1475T RNA sequencing and proteomics approaches reveal novel multi-cellular deficits in the cortex of Rett syndrome mice. N.L. Pacheco.

1476F Metabolomic studies of a brain-specific mouse model of tuberous sclerosis complex: Major changes in the methylation pathway. M.J. Gambello.

1477W Kinome profiling of neural stem cells (NSC): Kinome profiling of NSC derived from induced pluripotent stem cells (iPSC) of Huntington's disease patient. A. Baharani.

1478T Bridging the therapy gap for rare genetic disorders: Comprehensive high-throughput drug repurposing screening to identify potential new treatment opportunities. C.M. Maher.

1479F ‡ Reproducibility in iPSC omics: An international multicentre study. V. Volpato.

1480W ‡ Targeted enrichment without amplification and SMRT Sequencing of repeat-expansion disease causative genomic regions. T. Clark.

1481T Novel treatment approaches for autism spectrum disorder: An in vitro model. L. Boccuto.

1482F LCM-Seq: Single cell-type whole genome bisulfite sequencing and transcriptomic profiling in post-mortem brain. D.M. Almeida.

1483W Single-nuclei transcriptomics in the brains of individuals with depression who died by suicide. C. Nagy.

1484T Personalized dosing of dichloroacetate by clinical genotyping assay. T. Langaee.

1485F Sequencing a baby for an optimal outcome: A clinical genomic application of newborn hearing screening. A.B.S. Giersch.

1486W Building and scaling the world's fastest clinical-grade whole genome based pipeline. Y. Ding.

1487T Disease relevant structural variation analysis by next-generation mapping. A. Hastie.

1488F A systems biology approach to the understanding of asthma severity through the integration of metabolomic, transcriptomic and epigenetic networks. R.S. Kelly.

1489W An integrative view of genetic and transcriptional regulatory interactions of the human placenta. F. Delahaye.

1490T Sequence read length effects on differential gene expression analyses using RNA-sequencing technologies. A.C. Shetty.

1491F Optimization of CRISPR/Cas9-mediated gene editing in monkey embryos. X. Luo.

1492W Development of a dual platform strategy for targeted DNA sequencing in genetic screening. C. Schumacher.

1493T Enrichment of long reads for Mendelian disease using the Oxford Nanopore MinION. E. Farrow.

1494F NGS pretesting and QC using Illumina Infinium Arrays. J. Romm.

1495W 900 exomes for rare disease research: Outcomes of the 2016 BBMRI-LPC WES call in collaboration with EuroBioBank and RD-Connect. S. Beltran.

1496T Performance comparison of two exome enrichment systems for enhanced coverage of disease-associated regions. L. Tian.

1497F Improvement in automated NGS sample preparation workflows: Combining the Agilent Bravo Liquid Handling Platform with Covaris Focused-ultrasonicators. C. Whitman.

1498W Spike-in controls designed for detecting sample bleeding and misidentification in sequencing workflows. J. Kinman.

1499T NEBNext Ultra II FS DNA: An enzyme-based, single tube fragmentation system for library construction. V. Panchapakesa.

1500F The Personal Genome Project Canada: Whole genome sequences and comprehensive medical annotation of 56 Canadians. M.S. Reuter.

1501W Comprehensive discovery of genomic variation from the integration of multiple sequencing and discovering technologies. X. Zhao.

1502T Detection of small exonic CNVs, SNPs and INDELs in a single assay. S. Melo.

1503F An optimized single tube workflow for robust, low input NGS library preparation. G. Durin.

1504W Microbiome technology comparison: Axiom microbiome array, 16S rRNA sequencing and metagenomics. J. Foster.

1505T A spin column and magnetic bead-based approach for the isolation of host and bacterial DNA from human feces. D. Wieczorek.

1506F Preparing small RNA libraries from low input and single cell total RNA. S. Shore.

1507W TaqMan Advanced miRNA assays to simultaneously study expression of miRNA and mRNA from serum samples. H. Veereshlingam.

1508T Complete, streamlined, reduced-bias workflow for RNA extraction and small RNA library preparation from serum and plasma samples. R. Wong.

1509F Study of touch DNA in simulated situations for forensic purposes. F.T. Goncalves.

1510W Design of Axiom Asia Precision Medicine Genotyping Array optimized for East and South Asian populations using improved SNP selection algorithms. A. Mittal.

1511T ‡ Pathogenic variants that alter protein code often disrupt splicing. R. Soemedi.

1512F Low-input transcript profiling with enhanced sensitivity using a highly efficient, low-bias and strand-specific RNA-seq library preparation method. M. Arn.

1513W High-throughput single-cell genomic profiling with droplet microfluidics. D. Eastburn.

1514T Highly efficient transcriptome profiling method for single-cell or low input RNA. K. Krishnan.

1515F Tissue-of-origin identification of forensic material by targeted sequencing of stable regions of RNA using RNase-H2. L. Parker-Katiraee.

1516W Quality deep-sequencing miRNA data from matched fresh and FFPE cells for expression analysis profiling. K. Tokarz.

1517T Evaluation of NXType™ NGS high-resolution HLA typing kit. S. Khor.

1518F European Genome-phenome Archive: Finding, applying for, and accessing controlled-access data. J. Spalding.

1519W Machine learning applied to single-molecule electronic DNA mapping for structural variant verification in human genomes. B. Bready.

1520T Automation of ultra-high molecular weight DNA isolation and labeling for genome mapping. P. Lynch.

1521F Application of electronic mapping to metagenomic characterization of a complex microbial sample. J.M. Sage.

1522W A comprehensive workflow for copy number variation identification from whole-genome sequencing data. B. Trost.

1523T Implementation of an automated sample quality control tool in a whole exome sequencing workflow. A. Regala.

1524F Capillary electrophoresis using the SeqStudio™ Genetic Analyzer as part of genome editing workflows. S. Jackson.

1525W ‡ Chromosomal integration of libraries of full-length mutant genes with associated barcode tags. X. Jia.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1526T Monitoring guide RNA synthesis for CRISPR/Cas9 genome editing workflow. M. Liu.

1527F Copy number variants can be detected using next generation sequencing. R. Drouin.

1528W HiSeq X and NovaSeq optimization for clinical applications. K. Walker.

1529T An optimal long-read workflow for human genome sequencing. J. Lenhart.

1530F Digital gene expression of up to 96 targets in 96 samples for cell line screening with nCounter® PlexSet™. G.T. Ong.

1531W NCBI resources for visualization and analysis of genome assemblies. V.A. Schneider.

1532T Genomic DNA analysis using automated pulsed-field capillary electrophoresis. J. Uthe.

1533F Targeted sequencing using a long-read sequencing technology. I. McLaughlin.

1534W Irreversible inactivation of ribonuclease A on a surface by UV LED. T.L. Thompson.

1535T iMETHYL: An integrative database of whole genome DNA methylation analysis combined with whole genome and whole transcriptome analyses of over 100 Japanese individuals. S. Komaki.

1536F CNVs from whole exome sequencing and array: First comparative study in a Brazilian clinical cohort. E.A. Zanardo.

1537W Hitting the target: An analysis of noncoding alterations as captured by panels and diagnostic exome sequencing at a commercial lab. B. Schoenfeld.

1538T Evaluation of PCR followed by high-resolution melting analysis and synthetic constructs on four commercial thermocyclers. K. Dawkins.

1539F Merck-custom Affymetrix Axiom™ genotyping array performance evaluation. J. Xu.

1540W Streamlined, efficient, and uniform molecular inversion probe capture for targeted sequencing. E. Boyden.

1541T Use of a molecular inversion probe (MIP) system for the detection of copy number variants. K. Jefferson.

Epigenetics and Gene Regulation

1542F ELOVL5, an epigenetic biomarker, predisposes for the risk of type 2 diabetes mellitus with inflammation. H. Lee.

1543W Star strand miR-192 (miR-192*) as an overlooked metabolic regulator in pre-diabetic liver. K.K. Miu.

1544T ‡ Physical interaction in human beta-cells between islet eQTL sites and target gene promoters at loci associated with type 2 diabetes risk and glycaemic traits. J. Torres.

1545F The molecular basis of increased diabetes susceptibility in carriers of the PGC1α (Ser482) risk allele. R. Vandnebeek.

1546W Cytosine methylation predicts renal function decline in American Indians. R. Hanson.

1547T ‡ Dissecting features of epigenetic variants underlying cardiometabolic risk using full-resolution epigenome maps. F. Allum.

1548F An epigenome-wide association study in whole blood of measures of adiposity among Ghanaians: The RODAM Study. K.A.C. Meeks.

1549W Serum bilirubin levels, UGT1A1 gene expression, and risk for ulcerative colitis. C.J. Gallagher.

1550T Methylation-wide association study of sex-specific methylation effects on central adiposity. A. Justice.

1551F ‡ Body mass index variant protects Mexicans from obesity through long intergenic non-coding RNA on chromosome *20q13.33*. Y.V. Bhagat.

1552W Transcriptome study of metabolic healthy obesity in African Americans. A. Gaye.

1553T A multi-tissue transcriptome association analysis of BMI provides a whole-body view into the impacts of adiposity. T. Tukiainen.

1554F CLEC16A dysfunction compromises lipophagy and mitophagy and facilitates risk to autoimmunity. R. Pandey.

1555W ‡ Promoter capture Hi-C in primary human white adipocytes identifies an interaction hub at a Mexican lipid locus. K.M. Garske.

1556T Transcriptomic profiles of duodenal biopsies in cholesterol gallstone diseases. E. Riveras.

1557F Allele specific chromatin signals uncover regulatory mechanisms in autoimmune and B cell related diseases. M. Cavalli.

1558W Comprehensive identification of differentially methylated regions associated with systemic sclerosis in dermal fibroblasts from African-American patients. W.A. da Silveira.

1559T Identification of hypermethylated T- and B-cell receptor signaling molecule clusters in systemic lupus erythematosus (SLE) patients by integrative analysis. M.B. Guo.

1560F A functional SNP in 2p14 associated with rheumatoid arthritis by modulating *ACTR2* gene expression via long-range interaction. B. Lu.

1561W Whole blood DNA methylation signatures of Crohn's disease susceptibility and progression. H.K. Somineni.

1562T ‡ An epigenome-guided approach to causal variant discovery in autoimmune disease. R.C. Pelikan.

1563F Treatment-naïve multiple sclerosis cases and controls exhibit differentially methylated regions in CD4+ and CD8+ T cells. B. Rhead.

1564W Functional annotation of chronic lymphocytic leukemia (CLL) risk loci. H. Yan.

1565T ‡ Integrative analysis identified pervasive long-range regulation mediated by super-enhancers shared in multiple autoimmune diseases. X.F. Chen.

1566F ‡ *Cis* regulatory variation determines dynamic HLA-DQB1 allelic expression in response to T cell activation. M. Gutierrez-Arcelus.

1567W Investigation of the effect of an autoimmunity associated SNP in the 6q23 locus on enhancer function using CRISPR/Cas9. S. Singh.

1568T An atlas of immune chromatin accessibility and gene expression. D. Calderon.

1569F Integrative fine-mapping of genetic loci affecting risk for multiple sclerosis using stimulated primary immune cells. R. Hinch.

1570W The role of T cell stimulation intensity in the expression of immune disease genes. D.A. Glinos.

1571T Blood cell type-specific genome-wide DNA methylation analysis of Chinese patients with early-onset systemic lupus erythematosus identifies loss of DNA methylation in genes related to the Type I Interferon pathway. H.Y.B. Chung.

1572F Integrative methylation/mRNA analyses identified an interferon-inducible-gene interaction network with a key gene PARP9 in rheumatoid arthritis. S. Lei.

1573W Epigenome-wide association study of autoimmune thyroid disease by next-generation capture sequencing. T.C. Martin.

1574T Using clustering analysis and meQTLs to probe differential methylation in females with multiple sclerosis. B. Reinstadler.

1575F A proteomic approach to identify transcription factors that selectively bind to causal polymorphisms in inflammatory bowel disease (IBD). C.J. Cardinale.

1576W Functional characterization of *TNIP1* causal variants associated with Systemic Lupus Erythematosus. S. Pasula.

1577T Long-range regulation of *IRF5* expression mediated by a functional SNP associated with systemic lupus erythematosus and systemic sclerosis. H.N. Thynn.

1578F Annotating the regulatory genome of CD4+ T cells: Predicting active in vivo transcription factor binding sites. T. Amariuta.

1579W Annotations that capture tissue-specific transcription factor binding explain a large fraction of disease heritability. B. van de Geijn.

1580T Integrative analysis of transcriptional regulation unveils regulatory modules that stratify SLE transcriptome. T. Wang.

1581F Altered methylation marks and compromised spermatogenesis in human male infertility. S.Kumar. Mohanty.

1582W Comparison of X chromosome inactivation in peripheral tissues and visceral organs in females with X-linked diseases. M. Reboun.

1583T Transcriptome analysis of cystic fibrosis molecular signatures. J.E. Ideozu.

1584F DNA hypermethylation and other epigenetic regulatory signaling pathway genes asociated with hidradenitis suppurativa (acne inversa). D. Jhala.

1585W Dissecting regulatory mechanisms altering skin pigmentation in Africans using genetic and functional genomic data. D. Kelly.

1586T Hippocampus and blood *APOE* locus DNA methylation in Alzheimer's disease. L. Bekris.

1587F Transcriptomic analysis of whole blood reveals potential biomarkers in African American Alzheimer disease. S.K. Sivasankaran.

1588W Understanding the endogenous regulation of Ataxin-1 in SCA-1. R. Manek.

1589T Haploinsufficiency models of *CHD8* in neuronal cells display alterations in chromatin landscape and regulatory consequences in Wnt signaling. E. Kerschbamer.

1590F Correlation of methylomic profiles between blood and cerebral spinal fluid in aneurysmal subarachnoid hemorrhage patients. A. Arockiaraj.

1591W Epigenomic signature of adrenoleukodystrophy predicts compromised oligodendrocyte differentiation. A. Pujol.

1592T Epigenetic silencing in Friedreich ataxia is caused by hypermethylation of the *FXN* promoter CpG island shore. L.N. Rodden.

1593F The study of Vitamin D *effect on VDR* gene expression in multiple sclerosis patients. Z. Shirvani-Farsani.

1594W ‡ Supplemental treatment for Huntington disease (HD) with miR-132 that is deficient in HD brain. M. Fukuoka.

1595T Decreased expression of *Beclin2* and *LC3* genes in PGRN deficiency: A CRISPR-Cas9 neuronal cell model. S. Napoletano.

1596F An integrated genetic-epigenetic approach for assessing risk for stroke in the Framingham Heart Study. R. Philibert.

1597W Isogenic iPSC-derived neurons for modeling the differential regulation of *SNCA* expression: Implication to the heterogeneity of synucleinopathies. O. Chiba-Falek.

1598T Interpreting regulatory effects of disease-associated variants: A lesson from *SNCA* rs356168. O. Glenn.

1599F Translation regulation in Alzheimer's disease. A. Shieh.

1600W 5-hydroxymethylcytosine alterations in the human postmortem brains of autism spectrum disorder. Y. Cheng.

1601T Genome-wide analyses of DNA methylation in autism brains suggest epigenetic-mediated dysfunction in GABA signaling. J.I. Young.

1602F A massively parallel reporter assay for variants associated with schizophrenia and Alzheimer's disease. D. Avramopoulos.

1603W Gene body methylation of tyrosine hydroxylase (*TH*) in the striatum is associated with cocaine dependence in humans. K. Vaillancourt.

1604T DNA methylation profiles in a cohort of Brazilian children with ADHD. T.V.M.M. Costa.

1605F A reference map for open chromatin-associated histone methylation and acetylation landscapes in the human frontal lobe. K.G. Girdhar.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1606W Convergence analysis on risks for schizophrenia by integrating genomics, DNA methylation and gene expression. D. Lin.

1607T The role of DNA methylation and the 5-HTTLPR long/short variant of the serotonin transporter gene (*SLC6A4*) in antidepressant treatment response. A.J. Lisoway.

1608F Dynamic DNA N6-methyladenine modification in mammalian brain and implications in neuropsychiatric disorders. B. Yao.

1609W A multi-dimensional characterization of anxiety in monozygotic twin pairs reveals susceptibility loci in humans. R.S. Alisch.

1610T Methylomic profiling and replication implicates deregulation of PCSK9 in alcohol use disorder. F.W. Lohoff.

1611F Epigenome-wide association study of opioid dependence in European American women. J.L. Montalvo-Ortiz.

1612W EGR family genes; new potential markers for etiology and symptoms' severity of schizophrenia. M. Amini faskhodi.

1613T Transcriptional profiling of long noncoding RNA in PTSD patients reveals a potential early biomarker of trauma-induced alterations in the acute phase after exposure. G. Guffanti.

1614F DNA methylation as a candidate biomarker for predicting antidepressant response. C. Ju.

1615W DNA methylation markers associated with injection drug use status and HIV infection among chronic injection drug users in the ALIVE study. C. Shu.

1616T Small non-coding RNAs in major depression and antidepressant response. R. Lin.

1617F Transcriptional and genetic changes underlying psychiatric disorders converge on a network of transcription factors and their target genes in the human brain. S.A. Ament.

1618W A direct regulatory link between microRNA miR-137 and *SHANK2* with implications for neurodevelopmental disorders. S. Berkel.

1619T G-quadruplex binding chemicals may ameliorate the cognitive function of ATR-X syndrome. T. Wada.

1620F An epigenome-wide association study of Williams syndrome. R. Kimura.

1621W Epigenetic suppression of VEGF in retinal pigment epithelial cells by ascorbate. D. Sant.

1622T C-to-U RNA editing of osteopontin in mouse retina with laser-induced choroidal neovascularization. J. Chen.

1623F DNA hypermethylation is associated nonsyndromic cleft lip and palate. B. Gorijala.

1624W Developmental *cis*-regulatory elements revealed by open chromatin landscapes in mouse fetal tissues. Y. Zhao.

1625T Disruption of a remote putative novel enhancer in the *cis*-regulatory domain of *FOXL2* in a multigenerational Polynesian family with BPES. H. Verdin.

1626F Differential expression of immunoglobulin genes in blood and lesion burden in familial cerebral cavernous malformation type 1 (CCM1) patients. H. Kim.

1627W *NSD1* haploinsufficiency evokes DNA hypomethylation at imprinted DMRs and the increased expression of imprinted genes. H. Watanabe.

1628T Genome-wide miRNA profiling in plasma of pregnant women with Down syndrome fetuses. I. Svobodova.

1629F The NIA Aging Cell Repository: Facilitating aging research on cells in culture. D. Requesens.

1630W Transcriptional profiling of aging effects in human trabecular meshwork. S. Ramdas.

1631T Aging and subregion specific transcriptional changes in the rat hippocampus. I.S. Piras.

1632F A longitudinal study of DNA methylation as a mediator of age-related diabetes risk. C.D. Grant.

1633W An evolutionary perspective of DNA methylation associated with age within the primate lineage. G. Housman.

1634T Testing a stochastic model of epigenetic drift in longitudinal DNA methylation data. C. Robins.

1635F Identification, replication and characterization of epigenetic remodelling in the aging genome. S. Li.

1636W Accelerated epigenetic aging in middle-aged African Americans and Whites. S. Tajuddin.

1637T Better statistical methods to predict age from DNA methylation. Q. Zhang.

1638F Fasting and solar time independently regulate expression of hundreds of genes in skin and fat tissue in population-level transcriptomes. A. Couto Alves.

1639W Identifying causal mutations with RNA-seq in mice with Mendelian disorders. N. Raghupathy.

1640T Discover regulatory grammar across 127 human cell types using tree-based recurrent neural network. Z. Zhang.

1641F ‡ Single cell methylomes: A method to assess mammalian neuron diversity. L. Kurihara.

1642W DNA methylation of *TNF* decreases after an intense bout of eccentric exercise. B. Hussey.

1643T Common DNA sequence variation leads to variation in 3D genome organization. Y. Qiu.

1644F Chromatin plasticity during hematopoietic cell differentiation and stimulation. J.V. Ribado.

1645W Pleiotropic effects of trait-associated genetic variation on DNA methylation: Utility for refining GWAS loci. E. Hannon.

1646T Fine mapping of interacting functional elements in Hi-C peaks. A.T. Jaroszewicz.

1647F ‡ Comprehensive functional annotation of the zebrafish genome. T. Liu.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

1648W DNA methylation and its impact on inter-population differences in disease risk and prognosis. M. Loh.

1649T X-chromosome epigenetic markers for age-prediction. I.C.T. Mello.

1650F Genomic DNA methylation changes in myalgic encephalomyelitis. L. Sarria.

1651W Wnt signaling in neural crest development: A possible mechanism for nonsyndromic cleft lip and palate. A. Vedenko.

1652T Shared household environment makes an important contribution to variation in the human methylome. Y. Zeng.

1653F Rare variants and parent-of-origin effects on whole blood gene expression assessed in large family pedigrees. A. Brown.

1654W Stratified comparison and network analysis of large eQTL-studies reveals factors affecting validity of cis- and trans eQTLs. H. Kirsten.

1655T Glomerular and tubulointerstitial eQTLs of patients with nephrotic syndrome. R. Putler.

1656F Epigenetic marks at major histocompatibility complex affect male fertility. S. Sarkar.

1657W Low correlation observed between DNA methylation in blood measured between a majority of CpG sites measured on both Illumina 450K and EPIC BeadChips. M.W. Logue.

1658T DNA methylation of *PPARGC1A* is associated with cycling performance. D.J. Hunter.

1659F Exploratory transcriptome and methylome analysis in Gilbert's syndrome. A. Tosevska.

1660W Various relationships between DNA methylation and gene expression in different tissues and ages. K. Wang.

1661T Nanopore full length mRNA sequencing resolves transcript structure in single auditory hair cells. P. Ranum.

1662F Local and systemic alterations in extracellular RNA following traumatic knee injury implicate catabolic and inflammatory biomarkers. A.J. Griswold.

1663W An epigenome correlation map using Infinium 450 DNA Methylation Array. W. Guan.

1664T The 5-HTTLPR polymorphism does not moderate the effect of sleep loss on neural responses to implicit threat and fear learning and memory. V.C. Kodavali.

1665F Powerful and robust method for XCI-escape inference from bulk RNA-seq. R. Sauteraud.

1666W Prevalence, tissue-specificity and age-dependent heritability of skewed X-inactivation. A. Zito.

1667T RIPK3-dependent regulation of cell death switch (live or dye) as major determinant in incontinentia pigmenti. A. Pescatore.

1668F ‡ Identification of genetically associated changes in 3D-chromatin architecture by leveraging haplotype information across a three-generation family. W.W. Greenwald.

1669W HyCCAPP uncovers CALR as a novel DNA-binding protein. H. Guillen.

1670T Furthering the GTEx project legacy through the GTEx biospecimen resource. E. Gelfand.

1671F The components of the human epigenetic machinery are highly co-expressed and very intolerant to variation. L. Boukas.

1672W An integrated -omics analysis of the epigenetic landscape of gene expression in human blood cells. E.M. Kennedy.

1673T DNA methylation changes as an exposure signature of cigarette smoking. E. Kim.

1674F Targeted DNA methylation *in vivo* using an engineered dCas9-MQ1 fusion protein. Y. Lei.

1675W Looking for an epigenetic footprint of music: Behavioral effects of auditory stimulation and its relation to the methylation level of BDNF exon IV within the hippocampus of Wistar rats. M.M. Velásquez Toledo.

1676T ‡ Novel deep learning approaches reveal sophisticated epigenetic regulation in eukaryotes. Z. Wei.

1677F The impact of arsenic exposure on whole blood DNA methylation: An epigenome-wide study of Bangladeshi adults. B. Pierce.

1678W Characterizing causal *cis*-regulatory variants using computational approaches and CRISPR/Cas9 genome editing. M. Brandt.

1679T Regulatory role of conserved non-coding elements. B. Ambroise.

1680F What we talk about when we talk about enhancers. M.L. Benton.

1681W GGmend: A Mendelian randomization method for finding gene-on-gene regulatory effects in the presence of unobserved confounders. R. Brown.

1682T A novel computational and experimental approach for allele-specific expression analysis in high-throughput reporter assays. C. Kalita.

1683F ‡ Identifying imprinted genes using parent of origin effects on gene expression in the Hutterites. S. Mozaffari.

1684W ‡ Efficient detection of trans-gene regulation via association with predicted gene expression. J.A. Mefford.

1685T Determining blood cell-type composition using DNA methylation sequencing. W.A. Cheung.

1686F ‡ Inter-individual variation in microbiome composition controls human gene expression. A.L. Richards.

1687W Characterizing tissue-specific lincRNA transcription and regulatory roles. A.D.H. Gewirtz.

1688T ‡ The landscape of short RNAs in human cell types and tissues. Y.Y. Leung.

1689F Silencing of transposable elements may not be a major driver of regulatory evolution in primate induced pluripotent stem cells. M.C. Ward.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1690W Map of the functional domains of the long non-coding RNA XIST generated using CRISPR mediated deletions. T. Dixon-McDougall.

1691T Recognition of human elements regulating escape from X-chromosome inactivation in mouse. S.B. Peeters.

Developmental Genetics and Gene Function

1692F Interaction of exocrine and endocrine pancreas in obesity ciliopathies. T. Hostelley.

1693W Down-regulation of *SRSF3* mRNA expression in mice with *KIF23* c.2747C>G mutation known as a cause of congenital dyserythropoietic anemia type III (CDA III): Expression study. A.-L. Vikberg.

1694T Haploinsufficiency of a histone modifier, Kmt2d, in a mouse model of Kabuki syndrome leads to widespread defects in the B cell lineage. G. Pilarowski.

1695F Models of human disease available from The Jackson Laboratory. S. Rockwood.

1696W Circulating cells protect against radiation-induced intestinal injury in a murine parabiosis system. J. Sung.

1697T *Xenopus* as a model of precision medicine: Application of CRISPR to mimic the mutations of human CSBS syndrome patients. S. Cha.

1698F Gene expression in the developing mouse pituitary gland. A.S. George.

1699W The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. V. Jordan.

1700T Identification of novel candidate genes for 46,XY disorders of sex development (DSD) using a C57BL/6J-Y^{POS} mouse model. E. Vilain.

1701F Kisspeptin and Kisspeptin receptor may be involved in the regulation of adrenocortical development and steroid hormone secretion. N. Settas.

1702W Genome-wide association study of infantile hypertrophic pyloric stenosis identifies four new loci and highlights the importance of embryonic *NKX2-5/BARX1* pathways. L. Skotte.

1703T Novel pathogenic variant in OFD1 results in male lethal oral facial digital syndrome type 1 with pituitary aplasia. D. Aljeaid.

1704F Human knockout of a cell cycle gene *CDC25B:* a novel disease-causing gene for an Alström-like syndrome with cataract, dilated cardiomyopathy and multiple endocrinopathies. C. Lam.

1705W Gene expression profiling of single oocytes reveals pathways and regulators involved in follicle activation. Y. Lyu.

1706T Investigating phosphatidylserine metabolism in Lenz-Majewski syndrome. C. Demetriou.

1707F Skeletal dysplasias in Saudi Arabian population. M. Faden.

1708W Structural optimization of TransCon CNP: Development of a sustained-release prodrug of CNP for achondroplasia. K. Sprogøe.

1709T Missense and splicing mutations in the retinoic acid catabolizing enzyme *CYP26C1* in idiopathic short stature. G. Rappold.

1710F The role of p63 isoforms in the epidermal development as replicated in cellular models for normal human skin and genetic skin diseases. I. Barragán Vázquez.

1711W *TP63* is expressed in adult epidermal and iPSC-derived melanocytes supporting the role ofΔNp63 in ectodermal gatekeeping and cell migration to the epidermis. D. Cunha.

1712T Familial dysautonomia: The regulation of *IKBKAP* in the nervous system and therapeutic approaches. S. Yannai.

1713F iPSC-derived neurons from patients with idiopathic ASD show deficits in neuronal differentiation and synaptic function. C. Garcia-Serje.

1714W Multi-system contributions to *Gabrb3*-related neurodevelopmental risk *in utero*. H. Moon.

1715T A *Drosophila* model of essential tremor. L.N. Clark.

1716F ‡ A mutation in *MAL* is associated with a neurodevelopmental condition characterized by central hypomyelination, cerebellar atrophy and developmental delay. M. Elpidorou.

1717W Defective myoblast differentiation in human muscle dysferlin-deficient cells. R. Ishiba.

1718T ‡ Evolutionary conserved ARX-regulatory pathway in mammals and nematode to find a convergent druggable pathway damaged in neurodevelopmental disorders. L. Poeta.

1719F Emerging role and clinical spectrum of DNM1 in intellectual disability and epilepsy. F. Bolduc.

1720W *c-fos* transcript profile in adult zebrafish brain after prolonged pentylenetetrazole exposure. K. Brito.

1721T Functional evaluation of rare variants in glutamate receptor *GRIN*, *GRIA*, *GRIK*, and *GRID* genes reveals a diversity of effects on receptor activity. S.J. Myers.

1722F *De novo* NMDAR *GRIN* mutations in M2 channel pore-forming domain associated with neurological diseases. H. Yuan.

1723W Intracellular mislocalization of mutant proteins as a screen for therapeutic agents to treat genetic diseases. T. Kouga.

1724T Genetics of congenital megacolon in East Asians. C. Tang.

1725F Pathogenic U2-type 5' GC donor splice site in the *FOLR1* gene causes cerebral folate deficiency with autism and attention deficient hyperactive disorder in three affected from a large consanguineous family. M. Alaamery.

1726W Impact of defective protein N-glycosylation on the developing mouse cerebellum. V. Cantagrel.

1727T PEA15 deficiency is associated with striking neurologic and motor abnormalities in *Felis catus*. J.N. Cochran.

1728F Pathologic characterization of a neurodevelopmental abnormality in the cerebral cortex of domestic cats with loss of PEA15. E.C. Graff.

1729W ‡ Investigation of synergistic interactions among genes in the 15q11.2-q13.1 region using *Drosophila melanogaster*. K.A. Hope.

1730T IBGC mouse model with *SLC20A2* mutation and potential prevention and therapeutics. J.Y. Liu.

1731F CRISPR/Cas9 overcomes the challenges of microsatellite knockin development. R. Oliveira.

1732W *Clec16a* knockdown mice develop a neuronal phenotype with ataxia. M. Bakay.

1733T The binding of RNA regulates the formation of nuclear membraneless structures by Matrin 3 and TDP43 in myocytes. M.C. Gallego Iradi.

1734F Neuronal inflammation and dysregulated mitophagy features in ubiquitous *Clec16a* knockdown mice. H.S. Hain.

1735W Impact of rare variants in genes that encode components of the endocannabinoid system. D. Smith.

1736T Genetic analysis of Japanese patients with neurofibromatosis type 1 and the neuronal complications. K. Fujita.

1737F Disruptions to the miRNA regulatory pathway may cause an increased rate of schizophrenia in individuals with 22q11.2 DS. W. Manley.

1738W Modeling limb-girdle muscular dystrophy with in vitro human skeletal myotubes. J.L. Marshall.

1739T The chr14.232.a pseudogene in the 14q21.2 region regulates the expression of the contiguous *LRFN5* gene and is deleted in a patient with autism. G. Cappucio.

1740F Modeling the effects of autism-related *TBR1 de novo* mutations on human cortical development. B.A. DeRosa.

1741W Does lack of X-inactivation for *SLC6A14* explain the very high male/female ratio in nonsyndromic autism? F.R. Jimenez-Rondan.

1742T Further evidence supporting the involvement of *ERC1* gene variation in ASD. S. Raskin.

1743F Evaluation and co-expression of marker genes of cell types in brain. R. Dai.

1744W Stem cell models for studying the role of epigenetic machinery in abnormal neurogenesis. N. Kommu.

1745T ‡ Changes of open chromatin regions reveal stage-specific transcriptional network dynamics in human iPSC-derived neurons. W. Moy.

1746F Identifying pathogenic genes associated with autism and other developmental disorders by in-depth analysis of chromosome microarray studies. V. Goitia.

1747W Rare family with partial duplication in 7q11.23 link four genes associated with intellectual delay and autistic phenotypes. J.R. Korenberg.

1748T Behavioral characterization in a mouse model of Bohring–Opitz syndrome. K. Walz.

1749F Multimodal MRI and DTI reveal common systems mechanism underlying Downs syndrome and Alzheimer's disease. L. Dai.

1750W ‡ Comprehensive catalog of cell types in the developing brain using single-cell transcriptional profiling. J.M. Simon.

1751T Cytoplasmic FMRP-Interacting Protein 2 (*CYFIP2*) causes syndromic intellectual disability. A. Begtrup.

1752F Truncating *de novo* mutations in *DLG4* responsible for intellectual disability with Marfanoid habitus. S. Moutton.

1753W Effect of an intronic mutation in the CLIP1 Gene (CLIP-170) in a patient with autosomal recessive intellectual disability. A. Rincon.

1754T MeCP2 AT-hook1 mutations disrupt DNA binding and chromatin compaction in patients with intellectual disability and schizophrenia. T.I. Sheikh.

1755F *De novo IRF2BPL* pathogenic variants cause severe precocious neurodegenerative disease. F. Tran Mau-Them.

1756W DDX3X: Robust phenotype-genotype correlations from recurrent de novo mutations in DDX3X in patients with global developmental delay and intellectual disability. R. Jiang.

1757T ‡ Regulating transcriptional activity by phosphorylation of the intellectual disability and seizure associated ARX homeodomain transcription factor. C. Shoubridge.

1758F ‡ Molecular and biochemical analyses to understand the genotype-phenotype correlation in patients with the maternally inherited MELAS disorder. A. Gropman.

1759W A recessive variant in forkhead box domain of *FOXF2* is associated with profound hearing loss and inner ear anomaly. G. Bademci.

1760T Custom capture high-throughput sequencing for mutation detection: Results from 217 coloboma subjects across 196 genes identifies novel mutations in genes associated with ocular coloboma. V.K. Kalaskar.

1761F Characterizations of *NMNAT1* mutants and mouse model of *NMNAT1*-LCA. X. Feng.

1762W Novel genes associated with optic nerve hypoplasia in 6 family trios: A clinical and exome study. P. Bitoun.

1763T Integration of whole exome sequencing, expression profiling, and pathway analysis for the identification of novel genes in familial exudative vitreoretinopathy. M.-Y. Chung.

1764F Evaluation and treatment of nystagmus in a Brazilian boy with septo-optic dysplasia. L. Gabriel.

1765W LCA9-associated NMNAT1 mutant protein study in Drosophila. J. Sun.

1766T Bardet-Biedl syndrome, postaxial polydactyly, Shh signalling pathway and a founder effect in a Libyan extended consanguineous family from Tarhunah Berber tribe. N.Bouayed. Abdelmoula.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1767F Acquired ventriculamegaly in a case with *SOX 9* mutation. A. Matsumoto.

1768W The role of WNT regulatory variants in nonsyndromic cleft lip and palate. L. Maili.

1769T A dog model of non-syndromic cleft palate. B. Schutte.

1770F Syndromic and nonsyndromic congenitally missing teeth: Prevalence, clinical manifestations and patterns. E. Severin.

1771W A PITX1 variant in a large pedigree with dominant lower extremity anomalies. Y. Guo.

1772T Multidisciplinary assessment of 49, XXXYY, a rare X and Y chromosomal variation (XYV). P. Lasutschinkow.

1773F ‡ Mutations in *NAA10* and *NAA15* are associated with a range of cardiac and neurodevelopmental phenotypes. G.J. Lyon.

1774W Protective mechanisms in Cornelia de Lange patients with early truncating variants in *NIPBL* generate an N-terminal truncated protein that is able to mediate cohesin loading in the absence of MAU2. I. Parenti.

1775T 11q terminal deletion associated with mild phenotype of Jacobsen syndrome. C. da Silva-Camargo.

1776F Deciphering the mechanisms of developmental disorders (DMDD): Shedding light on human genetic disease using embryonic lethal knockout mice. A. Galli.

1777W On the significance of craniosynostosis in a case of Kabuki syndrome with a concomitant *KMT2D* mutation and 3.2 Mbp de novo 10q22.3q23.1 deletion. A. Topa.

1778T Expanding the phenotypic spectrum of *de novo KAT6A* mutations and their impact on biological pathways through functional genomics. V. Arboleda.

1779F The IMPC: A global research infrastructure for understanding the role of genes in human development and disease. V. Munoz Fuentes.

1780W Associated anomalies in cases with esophageal atresia. C. Stoll.

1781T Defining requirements for cleavage of prelamin A by the zinc metalloprotease ZMPSTE24. T. Babatz.

1782F ‡ A mouse model of Proteus syndrome. M.J. Lindhurst.

1783W Sex differences in mtDNA content and its relationship to mitochondrial enzyme activities in the context of human skeletal muscle aging. M. Tesarova.

1784T Mechanistic insight of inactivation of mouse chitinase-like protein Ym1. F. Oyama.

1785F Gene expression profiling of puberty-associated genes reveals abundant tissue and sex-specific changes across postnatal development. H. Hou.

1786W SRY potentially regulates early dopaminergic differentiation from male hiPSCs. D.D. Cao.

1787T An iPSC approach to examine the molecular mechanisms underlying *SRCAP* mutations in Floating-Harbor syndrome. R.L. Hood.

1788F ROS induced oxidative stress up-regulates DNA repair gene uracil DNA glycosylase, a potential anti-leishmania drug target. A. Mishra.

1789W Generation of humanized CD4 knock-in mice using CRISPR/Cas9. K.C. Chen.

1790T The association of haploinsufficiency of ARID2 with Ras-MAPK signaling pathway. M. Kang.

1791F 30 kDa fragment of beta-dystroglycan co-immunoprecipitates with Dp71 isoforms in PC12 cells. C.O. Azotla Vilchis.

1792W Analysis of copy number variation and association with facial shape in a large cohort of Bantu African children. F. Yilmaz.

1793T *ARMC5* and PMAH: From human genetic defects to the *Armc5*$^{+/-}$ mouse. F.R. Faucz.

1794F Establishment of primary cell lines from normal and abnormal human products of conception. D. O'Day.

1795W Genomic characterization of human induced pluripotent stem cells after CRISPR/Cas9 fluorescent tagging. T.S. Grancharova.

Complex Traits and Polygenic Disorders

1796T Assessment of the impact of variants in constrained non-essential splice sites in fifty-two thousand type 2 diabetes cases and controls. J.M. Mercader.

1797F Replication of newly identified type 2 diabetes candidate gene variants in Northwest Indian population groups. V. Sharma.

1798W HLA imputation and allelic associations with type 1 diabetes in African Americans. C.C. Robertson.

1799T Genetic variability in energy expenditure and the risk of severe obesity. A.C.P. Fonseca.

1800F Genome-wide meta-analysis of macronutrient intake identifies two novel loci: Cohorts for heart and aging research in genomic epidemiology. J. Merino.

1801W Diabetes in cystic fibrosis and type 2 diabetes (T2D) have overlapping genetic risk architecture. M. Atalar.

1802T Impact of genetic variants identified in genome-wide association studies of diabetic retinopathy in Chinese patients with type 2 diabetes. C.Y.Y. Cheung.

1803F ‡ Chromatin accessibility landscapes in adipose tissue and preadipocytes at cardiometabolic trait loci. K.W. Currin.

1804W ‡ Multivariate genome wide association study uncouples "favourable" from "unfavourable" adiposity alleles. Y. Ji.

1805T Whole exome sequencing and exome array genotyping in 3,943 Korean type 2 diabetes cases and controls. S. Kwak.

1806F Identifying novel genetic variants associated with EHR-derived serum zinc in DiscovEHR study cohort. R.P. Metpally.

1807W Evaluating tyrosine hydroxylase (*TH*) as a type 2 diabetes candidate gene in American Indians. A. Nair.

1808T ‡ Glucose challenge metabolomics identifies C10- and C12-carnitines as possible contributors to insulin resistance. C. Nowak.

1809F A rare frameshift mutation in exon 9 of glucokinase regulator (GCKR) is associated with a severe progressive histological form of Nonalcoholic Fatty Liver Disease (NAFLD). C.J. Pirola.

1810W ‡ Novel genetic determinants of diabetic kidney disease. R.M. Salem.

1811T Diabetome: A comprehensive collection of diabetes phenotype and genotype data. S. Shah.

1812F Measures of body composition and muscle fitness associate with nonalcoholic fatty liver disease (NAFLD). E.K. Speliotes.

1813W A functional locus at 8q21.13 to FABP4 levels is modulated by BMI and kidney function: Meta-analysis of five GWAS. S.W. van der Laan.

1814T Systemic approach to understand human non-alcoholic fatty liver disease. T. Yoo.

1815F ‡ Expanding the spectrum of type 2 diabetes risk alleles through a genome-wide association study imputed to the Haplotype Reference Consortium Panel. D. Taliun.

1816W Profiling of the long non-coding RNA (lncRNA) *MALAT1* in the liver of patients with nonalcoholic fatty liver disease (NAFLD) shows association with an aggressive histological phenotype. S. Sookoian.

1817T Multifactor dimensionality reduction (MDR) method to study association of type 2 diabetes mellitus with ENPP1 (K121Q), TCF7L2 (G>T) and GYS1 (A1>A2) gene variants in Punjabi population, India. B. Doza.

1818F A type 1 diabetes genetic risk score developed in Europeans discriminates between type 1 and type 2 diabetes in South Asian Indians in India. J. Harrison.

1819W ‡ Genetic factors influencing glycated hemoglobin, fasting glucose, and fasting insulin levels in the Population Architecture using Genomics and Epidemiology Study. H.M. Highland.

1820T Association study of ENPP1 (K121Q), TCF7L2 (G>T), GYS1 (A1/A2) variants with type 2 diabetes mellitus (T2DM) in north Indian Punjabi population. M. Kaur.

1821F Pathway-informed genetic testing and analysis for type 2 diabetes. C. Ma.

1822W ‡ Fine-mapping fasting glucose and fasting insulin loci with whole genome sequence data from the Trans-Omics for Precision Medicine (TOPMed) Program. A. Manning.

1823T Glucose metabolism in pregnancy: Analysis of diabetes related genetic risk scores against glucose curve trajectories. G.H. Moen.

1824F Tissue specific isoform annotations in rare variant analysis. A. Ndungu.

1825W Meta-genome-wide association study identifies multiple loci in the MHC region and a locus on chromosome 1 for serum C-peptide in type 1 diabetes. D. Roshandel.

1826T Replication of 93 T2D associated SNPs in Jat Sikhs, population of Punjab, India. G. Singh.

1827F Type 1 diabetes genetic risk score identifies neonatal diabetes patients with highest probability of mutations in Iranian population. H. Yaghootkar.

1828W ‡ Trans-ethnic discovery of the genetic architecture of glycaemic control. C. Langenberg.

1829T Characterization of potential regulatory variants at the *SH2B1* body-mass index GWAS locus. M.E. Cannon.

1830F Analysis of whole exome and whole genome sequencing using family-based linkage suggests rare variants with large effects are relatively common in extended families. N.D. Palmer.

1831W Meta-analysis in continental Africans and African Americans identifies PLCB3 as a novel locus of serum uric acid. G. Chen.

1832T Transient genetic effects important for early growth programming. O. Helgeland.

1833F Genome-wide scan using Korea Biobank Array discovered that two rare missense variants on GPT gene were associated with liver enzyme level. Y.J. Kim.

1834W Genome-wide association study of clinically-defined gout and subtypes identifies multiple susceptibility loci including transporter genes. H. Matsuo.

1835T Genome-wide study suggests a parent-of-origin effect on birth weight at *ANK1-NKX6-3* type 2 diabetes locus. R.N. Beaumont.

1836F ‡ Meta-analysis in 93,701 East Asians identifies new loci associated with type 2 diabetes. X. Sim.

1837W Using genetics to understand the relationship between inflammation and cardiometabolic traits. N.R. van Zuydam.

1838T Phenome-wide association study of exome data from childhood obesity cohort reveals pleiotropic loci for 13 obesity-related traits. S.B. Cho.

1839F Molecular assessment of variants in inherited lipodystrophy genes: Prevalence and clinical impact in a large clinical care cohort. C. Gonzaga-Jauregui.

1840W Genetic association of irisin with obesity and metabolic syndrome. H.B. Jang.

1841T Genotype determination: Analysis of *PNPLA3, GC*, and *LCP1* genes in nonalcoholic fatty liver disease in south of Iran. S.S. Tabei.

1842F Metabolomics screen in five metabolic tissues from healthy, prediabetic and type 2 diabetic subjects suggests new defects and points of gene environment interaction. C. Wadelius.

1843W Identification of I287S homozygous mutation in the MLX gene in an infant with non-alcoholic steatohepatitis: A case report. Y. Watanabe.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1844T ‡ Causality links between gut microbiome and glucose/insulin metabolism and type 2 diabetes. S. Sanna.

1845F Zebrafish larvae as a model system for high-throughput, image-based screens in insulin resistance and diabetes. A. Emmanouilidou.

1846W New insights into the role of genetic variation within *FGF21* in the pathogenesis of obesity. E. Aerts.

1847T Genetic evidence that early carbohydrate-stimulated insulin secretion affects accumulation and distribution of adiposity. C.M. Astley.

1848F Exome sequencing in African American children with early-onset obesity reveals new insights. A. Chesi.

1849W Identifying subject-specific regulatory networks of diet-induced weight loss. D.C. Croteau-Chonka.

1850T Copy number variation and mutation analysis indicate a possible interesting role of *POU3F2* in the Prader Willi *like* phenotype. E. Geets.

1851F A dinucleotide deletion in a putative miRNA target site in long-chain fatty acid elongase Elovl6 associates with higher thermogenesis and lower body mass index in Pima Indians. P. Kumar.

1852W Low serum insulin-like growth factor-II levels correlate with high body mass index in older American Indian adults. Y. Muller.

1853T ‡ Contrasting the genetic architecture of human thinness and severe obesity. F. Riveros Mckay Aguilera.

1854F The role of genetic and self-identified ancestry in determining obesity among African and Hispanic Americans. A. Vishnu.

1855W Genetic diversity and functional genomic mapping in an Emirati population with type 2 diabetes. K.S. Elliott.

1856T Effects of long noncoding RNA regulation of gene expression on type 2 diabetes. A.J. Payne.

1857F Identification of eQTLs affecting expression levels in both adipose and skeletal muscle tissues. W.-C. Hsueh.

1858W Evaluating the contribution of alternative splicing in the liver to variation in lipid levels. K.A.B. Gawronski.

1859T Meta-analysis of >150 genome-wide studies for association with blood lipid levels. S.E. Graham.

1860F Population and medical genetics of the Kibbutzim Family Study. S. Carmi.

1861W ‡ Regulatory activity and deletion of rs3780181 suggests a molecular mechanism at the *VLDLR* lipid GWAS locus. J. Davis.

1862T Heritability and genetic correlation of 25 complex traits in Taiwanese population. C. Lin.

1863F Genome-wide association study of HDL efflux phenotypes in 5,143 French Canadians. K.S. Lo.

1864W Genetic effect assessment of functional variants on blood lipid traits by exome-wide association study. S. Moon.

1865T A multi-trait genetic association approach to identify genetic loci not identified before in single-trait GWAS of lipid traits. M. Preuss.

1866F Low LDL cholesterol concentrations are associated with increased risk of type 2 diabetes. Q. Feng.

1867W Whole exome sequencing identifies coding variants associated with NMR-based lipid phenotypes in a large cardiovascular cohort. S. Giamberardino.

1868T ‡ Novel genetic variants associated with lipid levels in a multi-ethnic population from the Population Architecture using Genomics and Epidemiology (PAGE) Study. Y. Hu.

1869F ‡ Genetic analysis of lipids in >300,000 participants in the Million Veteran Program. D. Klarin.

1870W Genome-wide association study of anthropometric, cardiovascular, and lipid biomarkers in an ethnically diverse cohort of sub-Saharan Africans. M. Hansen.

1871T *TM6SF2* rs58542926 impacts lipid processing in liver and small intestine. N.A. Zaghloul.

1872F ‡ Gender differences in genetics of body composition and obesity traits after an intensive exercise intervention. A.I. Vazquez.

1873W ‡ Evaluation of loss-of-function mutation in *PCSK9* gene in large nationwide health registry based PheWas study in Finland. M. Alanne-Kinnunen.

1874T Genetic effects of familial hypercholesterolemia variants on LDL cholesterol levels among multi-ethnic veterans: The Million Veteran Program Study. Y.V. Sun.

1875F Lipid loading in human liver cells induces differential expression of 88 genes. M. Alvarez.

1876W Human liver transcriptomes reveal potential new cholesterol genes under tight co-regulation with statin-targeted cholesterol synthesis pathway genes. A. Ko.

1877T Genetic regulation of adipose tissue transcript expression is involved in modulating serum triglyceride and HDL-cholesterol. S.K. Das.

1878F The X-factor of complex disease: Development of methods and software for analysis of the X chromosome in GWAS and RVAS reveals X-autosomal gene-gene interactions and X-linked associations underlying lipid levels and their sexual dimorphism. A. Keinan.

1879W ‡ Common and rare genetic variants for asthma, hay fever and eczema. W. Ek.

1880T Multi-omic approaches to identifying clinical biomarkers of asthma exacerbations in African Americans. H. Gui.

1881F Shared genetic etiology and ancestry variations between asthma and major complex diseases. T. Mersha.

1882W ‡ Mapping human airway smooth muscle cell transcriptional and epigenetic responses to asthma-promoting cytokines reveals enrichments for asthma-associated SNPs. E.E. Thompson.

1883T Unraveling the genetic architecture of generalized vitiligo in a homogeneous, isolated Romanian village. G. Andersen.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

1884F Polygenic risk score predicts development of HCV-associated mixed cryoglobulinemia and response to interferon-free therapy. M. Artemova.

1885W Identification of several genes modifying multiple sclerosis risk conferred by tobacco smoke: A case-only analysis. F.B.S. Briggs.

1886T Admixture mapping of 13,569 individuals provides evidence for increased European origin of the major histocompatibility complex class i region in multiple sclerosis. C. Chi.

1887F Immune-phenotypes among patients with systemic lupus erythematosus and their association with *HLA-DRB1* alleles. L.M. Diaz-Gallo.

1888W GWAS polygenic model approach applied to primary biliary cholangitis (PBC) in a Japanese population. O. Gervais.

1889T Lupus-associated functional polymorphism in *PNP* causes cell cycle abnormalities and interferon pathway activation in human immune cells. Y. Ghodke-Puranik.

1890F Association of protein tyrosine phosphatase non-receptor N22 gene functional variant R620W with systemic lupus erythematosus patients from Kuwait. M.Z. Haider.

1891W ‡ Transcription factors are associated with disease risk loci: Epstein-Barr virus nuclear antigen 2 (EBNA2) is an environmental factor associated with multiple autoimmune diseases. J.B. Harley.

1892T Identification of the primary functional variants in primary biliary cholangitis susceptibility gene loci *NFKB1/MANBA*. Y. Hitomi.

1893F *TYK2* correlates with multiple sclerosis symptomatology at onset. J. Jiangyang.

1894W Genetic screening of Galectin-3 CRD variants in RA: A case-control association study. T. Kaur.

1895T Genetic variation in the estrogen receptor alpha gene (*ESR1*) and susceptibility to rheumatoid arthritis. S.E. Lofgren.

1896F Identification of rare variants in Italian multiplex families with multiple sclerosis using a next generation sequencing approach. E. Mascia.

1897W Genetic association between not related to HLA immune gene polymorphisms and development of specific autoantibody is limited to few genetic loci in patients with rheumatoid arthritis. L. Padyukov.

1898T Variants near *HLA-DQA1* contribute to the development of antibodies to anti-TNF in Crohn's disease. A. Sazonovs.

1899F Longitudinal clinical, molecular, and immunohistochemistry changes during mycophenolate mofetil therapy in patients with systemic sclerosis. D.M. Toledo.

1900W Integrative analysis of genetic, gene expression and DNA methylation data on systemic lupus erythematosus (SLE). W. Yang.

1901T Identification of one novel IBD susceptibility locus through a genome-wide association study in Korean populations. B.D. Ye.

1902F The influence of human genetic variation on HIV related non-Hodgkin lymphoma. C.W. Thorball.

1903W *IL1RN* variants influence systemic juvenile idiopathic arthritis susceptibility and are a biomarker of non-response to treatment with anakinra. E.G. Shuldiner.

1904T First report of the mutational and phenotypic spectrum of hereditary spherocytosis in Indian patients. A. Aggarwal.

1905F Autoimmunity genes are associated with acquired hypothyroidism in a large clinical cohort. J. Freudenberg.

1906W Trans-ethnic meta-analysis of fetal hemoglobin genome-wide association results identifies common variants at the *KLF1* locus. Y. Ilboudo.

1907T An intergenic variant between HLA-DRA and HLA-DRB contributes to the clinical course and long-term outcome of ulcerative colitis in Asians. H.S. Lee.

1908F Genome-wide association study of otitis media in children. J. Li.

1909W Multiple HLA B*57 alleles, sharing the amino acid residue valine^{97}, are associated with drug-induced liver injury due to flucloxacillin in a European population. P. Nicoletti.

1910T ‡ Transcriptome analysis of systemic lupus erythematosus reveals distinct susceptibility, activity and severity signatures. N. Panousis.

1911F *NKG2D* variation and viral bronchiolitis. A. Pasanen.

1912W Human genetic variation impacts total IgA levels and pathogen-specific IgG levels. P. Scepanovic.

1913T Characterising copy number variation at the Crohn disease-associated gene intelectin 1 (*ITLN1*). F. Almalki.

1914F Genetic footprints and functional analysis of polymorphisms in the *PKLR* gene. O.C.L. Bezerra.

1915W Exome sequencing identifies variants of the alkylglycerol monooxygenase gene (*AGMO)* as a cause of relapses in visceral leishmaniasis in Sudan. S. Marquet.

1916T *De novo* mutations implicate novel genes with burden of rare variants in systemic lupus erythematosus. A. Roberts.

1917F A long non-coding RNA in the rheumatoid arthritis risk locus at chromosome 18 is involved in T cell activities. M. Houtman.

1918W WGS identifies rare variants influencing variation in blood cell traits in Mexican American families. N.B. Blackburn.

1919T Trans-ethnic meta-analysis of the Korean, East Asian and European Immunochip data identifies three novel IBD susceptibility loci. S. Jung.

1920F Associations between a polymorphism of the gene encoding the Toll like receptor and response to infliximab in Japanese patients with Crohn's disease. S. Ma.

1921W GWA and MHC-fine mapping analyses of multiple sclerosis (MS) age at diagnosis (AAD) identify novel associations with an HLA-DQ heterodimer and *GZMA*. P.G. Bronson.

1922T Pleiotropy analysis of penicillin and sulfa drug allergy in the Kaiser GERA cohort. A. Majumdar.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

1923F Genetic markers associated with leprosy susceptibility in a group of incident household contacts from Brazil. L. Arnez.

1924W Genome-wide association study identifies *HLA-DR/DQ* region for childhood nephrotic syndrome in Japanese. X. Jia.

1925T A genome-wide association analysis identifies *NMNAT2* and *HCP5* as susceptibility loci for Kawasaki disease. J. Kim.

1926F Immunochip v2.0 meta-analysis identifies shared genetic loci for inflammatory bowel disease in Korean population. S.B. Lee.

1927W GWAS identified associations of *HLA-DRB1-DQB1* haplotypes and *BTNL2* gene with response to a hepatitis B vaccine. N. Nishida.

1928T Reduced severity of collagen-induced arthritis in peptidylarginine deiminase type 4 knockout mice. A. Suzuki.

1929F A locus on chromosome 5 shows African-ancestry-limited association with alloimmunization in sickle cell disease. L.M. Williams.

1930W GWAS meta-analysis in Chinese and European populations identified a novel locus associated with systemic lupus erythematosus on Xp11. H. Zhang.

1931T Targeted sequencing in 1000 SLE patients discovers regulatory alleles that downregulate DAP expression and promote autoimmunity. P. Raj.

1932F Cross-genetic heritability of maternal and neonatal immune mediators during pregnancy. M. Traglia.

1933W Association analysis of rheumatoid arthritis through whole-exome sequencing in a Singapore Chinese cohort. V. Kumar.

1934T Assessing the mechanisms of thymic involution in an animal model of multiple sclerosis. S.G. Gregory.

1935F ‡ Sex-specific transcriptional responses to lipopolysaccharide (LPS) in peripheral blood leukocytes (PBLs) in the Hutterite founder population. M. Stein.

1936W Role of rare variants in progression form latent to active tuberculosis in Peruvian population. S. Asgari.

1937T Deciphering genetic susceptibility to tuberculous meningitis: Exome sequencing and a GWAS in a South African population. M. Möller.

1938F Genome wide association in Peru demonstrates that progression to active tuberculosis is a polygenic and highly heritable trait. S. Raychaudhuri.

1939W Genetic variation in *GLS2* is associated with development of complicated *Staphylococcus aureus* bacteremia. W.K. Scott.

1940T High density imputation genome wide association study of spontaneous resolution of hepatitis C virus. C.I. Vergara.

1941F Whole genome sequencing of pharmacogenetic drug response in racially and ethnically diverse children with asthma. A.C.Y. Mak.

1942W *ITPKC* and *SLC11A1* gene variations are associated with Kawasaki disease patients. Y. Bae.

1943T ‡ Location, location, location: Single cell gene expression of mucosal T cells vs peripheral blood T cells in Crohn's disease. E.A.M. Festen.

1944F ‡ Comprehensive bioinformatic characterization around RASGRP1 gene identifies multiple potential functional variants for lupus susceptibility. J.E. Molineros.

1945W Male-specific association of the *FCGR2A* His167Arg polymorphism with Kawasaki disease. Y. Kwon.

1946T Local ancestry interaction models reveal a novel asthma association with asthma on chromosome 1q23.1 specific to people of African ancestry. M.P. Boorgula.

1947F A genome-wide association analysis of Hashimoto's thyroiditis. V. Boraska Perica.

1948W HLA-DPB1 and Graves disease in Han Chinese. Y. Lee.

1949T Genome-wide association study identifies candidate loci associated with intraoperative remifentanil requirements during laparoscopic-assisted colectomy. D. Nishizawa.

1950F NELFCD and CTSZ loci are associated with jaundice-stage progression in primary biliary cholangitis in the Japanese population. M. Kawashima.

1951W Exome-wide association study of kidney function in 55,041 participants of the DiscovEHR cohort. C. Schurmann.

1952T Alternative splicing of *ICAM3* in Crohn's disease. I. Arijs.

1953F ‡ Chronic obstructive pulmonary disease subtyping through multiple -omics data integration. B.D. Hobbs.

1954W Identifying genetic determinants of age at menarche and age at menopause in the Japanese population. M. Horikoshi.

1955T Markers of the adaptive immune response are associated with progressively worse chronic kidney disease status. D.C. Crawford.

1956F Effect of CAG repeat length in the *androgen receptor* gene on hirsutism among healthy Israeli women of different ethnicities. S. Ben-Shachar.

1957W Rare variation associated with immunosuppressant drug concentrations: Moving beyond common SNPs in predicting drug metabolism. A.A. Seyerle.

1958T *NUDT15* variants contribute to thiopurine-induced myelosuppression in European populations. M.D. Voskuil.

1959F Exome sequencing highlights novel DNA variants with a potential role in polycystic ovary syndrome. S.G. Wilson.

1960W Search for genetic factor associated with right-sided colonic diverticula in Korean population: Genome-wide association study. E. Choe.

1961T Dysregulated gene and miRNA expression in different stages of Crohn's disease. I. Cleynen.

1962F Comparative bacteria communities between gallbladder bile and gallstone in gallstone disease patients in Taiwan. H. Yang.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

1963W ‡ 101 novel loci and novel associations with gene expression detected in transethnic genome-wide study of estimated glomerular filtration rate in over 270,000 participants: The Million Veteran Program. A.M. Hung.

1964T An analysis of Crohn's disease genes in the French-Canadian population. B.E. Avila.

1965F Evaluation of candidate genes for Hirschsprung disease using target sequencing. W.Y. Lam.

1966W Identification of blood UMOD and HER2 as causal mediators of chronic kidney disease using Mendelian randomization in the ORIGIN trial. J. Sjaarda.

1967T Mendelian randomization shows non-linear causality between vitamin D levels and kidney function. A. Teumer.

1968F Differential metagenomic analysis associated between alcoholic and non-alcoholic fatty liver disease using 16S rRNA gene sequencing. Y. Yun.

1969W Discovery of health disparities among African American patients at Vanderbilt University Medical Center. K. Actkins.

1970T Fetal but not maternal APOL1 genotype is associated with increased risk for preeclampsia among African-Americans. C.L. Simpson.

1971F Patterns of *APOL1* G1 association with kidney function in young adults in five African diaspora populations. B. Tayo.

1972W Integrated linkage and rare variant association tests reveal rare variants associated with elevated androgen levels in polycystic ovary syndrome. M. Dapas.

1973T Genome-wide CNV analysis identifies *TGFBR3* as a candidate causal gene for endometriosis and infertility. E. Hatchwell.

1974F Identification of lung cell populations from single-cell RNA-seq profiling of murine emphysema model. J.H. Yun.

1975W The multi-phenotype derived Nephrotic Syndrome Severity (NS2) score empowers genomic discovery. C.E. Gillies.

1976T Transcriptomic analysis of the ratio of serum aspartate transaminase to serum alanine transaminase (Ast/Alt ratio) using a genotype-by-diet interaction model identifies a number of potentially important genes for liver disease in the San Antonio Family Heart Study. V.P. Diego.

1977F Chromatin interactions reveal novel gene targets for drug repositioning in rheumatic diseases. P. Martin.

1978W Genetic burden contributing to extremely low or high bone mineral density in a senior male population from MrOS study. S. Chen.

1979T An integrative analysis of gene expression profiling and genome-wide DNA methylation datasets shows a different underlying molecular mechanism between Kashin-Beck disease and osteoarthritis. Y. Wen.

1980F Identification of genetic variants associated with knee OA in patients with knee replacement surgery. Y. Zhang.

1981W Evaluation of shared genetic aetiology between osteoarthritis and bone mineral density identifies *SMAD3* as a novel osteoarthritis risk locus. S. Hackinger.

1982T Identification of *IRF4*, *NOTCH4* and *RPS12* genes for non-syndromic sagittal craniosynostosis in a genome-wide association study using logistic regression. H. Sung.

1983F Copy number variant causes the mis-calculation of single nucleotide polymorphism in association analysis. Z. Yan.

1984W ‡ Osteoporosis-associated risk variant in distal enhancer at 1p36.12 regulates expression of long noncoding RNA through long-range loop formation. Y. Guo.

1985T Broad mutation spectrum of *FBN1* gene implicated in a cohort of idiopathic scoliosis and its related genotype-phenotype correlation study. M. Lin.

1986F ‡ PheWAS meta-analyses on bone microarchitecture phenotypes assessed by HR-pQCT and CRISPR/Cas9 gene-editing in zebrafish identify novel genetic risks of osteoporosis and fractures: The Bone Microarchitecture International Consortium (BoMIC). Y.H. Hsu.

1987W Novel genetic risk factors identified from a genome-wide association study for lumbar disc degeneration in Southern Chinese. Y. Li.

1988T ‡ The genetic architecture of osteoarthritis: Insights from UK Biobank. E. Zeggini.

1989F Longitudinal genome-wide association analyses and heritability estimates of pediatric bone mineral density. D. Cousminer.

1990W Exome sequencing in idiopathic scoliosis families implicates cilia genes in disease etiology. E.E. Baschal.

1991T Association of TGF β1 gene polymorphisms with primary knee osteoarthritis in Asian Indians. Q. Hasan.

1992F The genetic architecture and phenotypic spectrum in the skeletal ciliopathies. W. Zhang.

1993W Targeted sequencing of sagittal nonsyndromic craniosynostosis in regions on chromosomes 3, 7, and 20. C.M. Justice.

1994T Shared and subtype-specific genetic variation define the genetic susceptibility of juvenile idiopathic arthritis. Y. Li.

1995F Characterizing the molecular biology of systemic sclerosis with RNA-Seq of both skin and PBMCs. E. Roberson.

1996W Developement of the scleroderma genotype-phenotype map for assessing phenotypic relationships and drug repurposing. K. Chesmore.

1997T Polymorphisms of genes involved in extracellular matrix homeostasis may play a role in the risk to develop anterior cruciate ligament and medial meniscus tears. L. Casilla.

1998F Filaggrin variations associated to atopic dermatitis in Ecuadorian pediatric population. V. Romero.

1999W Differential alternative splicing of *MAPT* in brains supports its role in the pathogenesis of Parkinson disease. L. Wang.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2000T Mapping functional regulatory variants at Alzheimer's disease risk loci. M. Allen.

2001F Whole-exome sequencing analyses for late-onset Alzheimer's disease in Japanese. Y. Asanomi.

2002W Identification and validation of novel key drivers of Alzheimer's disease from multiscale causal networks that integrate large-scale DNA, RNA, and proteomic data. N.D. Beckmann.

2003T Whole-genome sequencing in non-Hispanic white familial late-onset Alzheimer's disease identifies rare variation in AD candidate genes. G.W. Beecham.

2004F Novel candidate AD-risk loci identified through whole exome sequencing in African Americans. M.M. Carrasquillo.

2005W A patient-derived iPSC model of a rare *TTC3* mutation segregating with Alzheimer's disease. H.N. Cukier.

2006T Genome-wide association study of brain amyloid deposition as measured by PiB-PET imaging and assessment of the genetic variance of amyloid deposition. F.Y. Demirci.

2007F Complement receptor 1 (*CR1*) intragenic duplication and Alzheimer's disease. E.J. Hollox.

2008W Genome-wide association study for Alzheimer's disease in a Puerto Rican dataset. J. Jaworski.

2009T A whole exome study of Alzheimer's disease which is augmented by population data found the noble AD risk genes. J. Kim.

2010F Alzheimer's disease exome sequencing study in the Finnish population isolate. M.I. Kurki.

2011W ‡ Performance of a genetically-based biomarker risk algorithm for an Alzheimer's disease prevention screening study. M.W. Lutz.

2012T ‡ Novel Alzheimer disease loci identified in subsets of whole exome sequencing data stratified by APOE genotype. Y. Ma.

2013F Ambidexterity and Alzheimer's disease risk. E.E. Mlynarski.

2014W Variant prioritization by pedigree-based haplotyping in an Alzheimer's disease pedigree. R.A. Nafikov.

2015T Contribution to Alzheimer's disease risk of rare variants in *TREM2, SORL1* and *ABCA7* in 1,779 cases and 1,273 controls. G. Nicolas.

2016F Single variant and polygenic score analysis of whole exome data from the Alzheimer's Disease Sequencing Project (ADSP). J.S. Reddy.

2017W The roles of *CD33* and *TREM2* in neurodegeneration associated with Alzheimer's disease (AD) and frontotemporal dementia (FTD). A. Rendina.

2018T Targeted sequencing of deep-phenotyped individuals for Alzheimer's disease susceptibility prediction. J. Seo.

2019F Genetic markers in LUZP2 and FXBO40 genes are associated with the normal variability in cognitive performance in the elderly. V.A. Stepanov.

2020W Whole genome sequence analysis of Caribbean Hispanic families with late onset Alzheimer's disease. B. Vardarajan.

2021T A new CAG repeat disease responsible for X linked cerebellar ataxia? L. Parodi.

2022F Neurobehavioral traits in family members inform gene discovery in ASD. S. Luzi.

2023W Genome-wide analysis in pediatric-onset multiple sclerosis (MS) confirms a role for adult MS risk variants and reveals new candidates. L.F. Barcellos.

2024T Multiple sclerosis in Orkney: The contribution of common variants to excess prevalence. C.L.K. Barnes.

2025F Elucidating the role that genetic ancestry plays on the impact of variation in the major histocompatibility complex on risk of multiple sclerosis. A. Beecham.

2026W Genetics of vaccination related narcolepsy. H.M. Ollila.

2027T Assessment of genomic variations in multiple sclerosis patients identifies mutations in *ADAMTS14*, *IL22RA2*, *HNRNPA1* and *TNPO1* genes indicating the existence of molecular mimicry and cytokine/interferon receptor pathway disruption inducing autoimmunity. A.M. Veerappa.

2028F ‡ Genomic and functional evaluation of the role of the TNFSF14-TNFRSF14 pathway in susceptibility to multiple sclerosis. M. Zuccala'.

2029W Predicted expression of *TMEM163* is associated with traumatic brain injury risk in a biobank population. J. Dennis.

2030T A genome-wide screen to identify suppressors of neurodegeneration in Gaucher disease. S.U. McKinstry.

2031F Genome editing by CRISPR-Cas9 followed by RNA sequencing to identify the transcriptional regulatory role of MEIS1 in restless legs syndrome. F. Sarayloo.

2032W Investigation of rare variations in four SLI candidate genes in Pakistani SLI population. E.M. Andres.

2033T Defining the critical region for brain malformations in 6q27 microdeletions. M.D. Dias Hanna.

2034F Genomic variants related to verapamil response in the treatment of migraine. F.M. Cutrer.

2035W Targeted sequencing of migraine-epilepsy susceptibility locus on chromosome 12q. M.E. Hiekkala.

2036T RNA-seq analysis after moderate blast exposure in peripheral blood samples. H. Kim.

2037F Very rare homozygous variants: A flashlight to possible involvement in ALS? A. Orr-Urtreger.

2038W Whole-genome sequencing points to *SV2A, DENND4B, MIB2, SPTBN2,* and *APP* as new genes of interest in two individuals sporadically affected with childhood apraxia of speech. B. Peter.

2039T Mutation identification for epilepsy in the U.S. Latino population using whole exome sequencing. C. Xu.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

2040F Identification of somatic mutations in malformations of cortical development. N.G. Griffin.

2041W Whole exome sequencing reveals known and novel genomic variants in a cohort of intracranial vertebra-basilar artery dissecting aneurysm (IVAD). S. Zhao.

2042T Polygenic analysis of persistent cisplatin-induced peripheral neuropathy implicates immune-mediated processes. O. El Charif.

2043F Hirschsprung's disease and the related genes in Taiwan. W. Yang.

2044W Leveraging large-scale exome sequencing data from >5,000 individuals to elucidate the genetic influences of amyotrophic lateral sclerosis. S.M. Farhan.

2045T Whole genome sequencing and rare variant analysis in essential tremor families. Z. Odgerel.

2046F Novel loss-of-function mutation of α-tocopherol transfer protein leads to vitamin E deficiency in a family. E. Smith.

2047W Exome sequence analysis identifies novel loci associated with carpal tunnel syndrome in DiscovEHR study cohort. S. Krishna Murthy.

2048T Genome-wide association study reveals candidate susceptibility loci for idiopathic hypersomnia. K. Tanida.

2049F Genome- and phenome-wide association analyses uncover MET as a susceptibility gene of cerebral palsy. J.J. Connolly.

2050W RNAseq gene expression profiling of CD4+ and CD8+ T cells from multiple sclerosis patients and healthy controls. S.D. Bos.

2051T *Gba1* haploinsufficiency in a Parkinson mouse impacts longevity and symptom severity independent of SNCA aggregate. N. Tayebi.

2052F Effect of genetic variants associated with uric acid on multiple sclerosis: A Mendelian randomization study. M.D. Montierth.

2053W A longitudinal metabolome-wide association study on beta amyloid in adults with increased risk for Alzheimer's disease. B.F. Darst.

2054T Interplay of genetic risk at *SNCA* locus and dysbiosis of gut microbiome in Parkinson's disease. Z.D. Wallen.

2055F Somatic copy number gains of alpha-synuclein (*SNCA*) in synucleinopathies (Parkinson's disease and multiple system atrophy). C. Proukakis.

2056W Protective effect of smoking and caffeine on Parkinson's disease: A gene-environment study. V. Altmann.

2057T ONDRISeq: Genetic diagnosis of neurodegenerative disease patients using targeted next-generation sequencing. A.A. Dilliott.

2058F Genome-wide CNV analysis identifies candidate causal genes for Parkinson's disease in the lysosomal pathway. P.S. Eis.

2059W Genetic modifiers modulating the age of onset of amyotrophic lateral sclerosis caused by expanded GGGGCC repeats. H. Kim.

2060T CGG interruptions alter protein properties and increase disease penetrance in SCA8. B.A. Perez.

2061F The NINDS human genetics DNA and cell line repository: A publicly available biospecimen collection for neurological disease research. A. Resch.

2062W Burden analysis of ALS-gene variants in patients with and without *C9orf72* expansion. J.P. Ross.

2063T Identification of candidate amyotrophic lateral sclerosis risk loci using pedigree based analyses of next-generation sequencing data. K.L. Russell.

2064F Common variants at 5q33.1 predispose to migraine in African-American children. X. Chang.

2065W A novel mutation in *INF2* gene: Expanding the genetic spectrum of Charcot-Marie-Tooth disease and glomerulopathy. P. Gupta.

2066T No rare deleterious variants from *STK32B*, *PPARGC1A*, *CTNNA3* are associated with essential tremor. G. Houle.

2067F Whole-genome sequencing in primary progressive multiple sclerosis uncovers mutations in genes for inherited leukodystrophies and other MS phenocopies. X. Jia.

2068W Prioritizing Parkinson's disease genes using population-scale transcriptomic data. G.T. Wong.

2069T Genetic analysis of SNCA gene polymorphisms in Parkinson's disease in an Iranian population. M. Rahimi.

2070F RNA-sequencing reveals novel immunological pathways in neuromyelitis optica. I. Adrianto.

2071W Decoding GWAS discoveries of neurodegenerative diseases: Gene expression changes in single neurons. L. Tagliafierro.

2072T The generation of iPSC-derived astrocytes from patients with Gaucher disease with and without Parkinsonism provide a model to study Parkinson pathogenesis. B. McMahon.

2073F Selective activation of caspase family of genes in multiple sclerosis patients inducing neuronal apoptosis. Y. Kattimani.

2074W Common genetic variation contributes to cognitive performance in Russian elderly population. O. Makeeva.

2075T Identifying the genetic underpinnings of social withdrawal. N. Roth Mota.

2076F Genetic investigation of restricted and repetitive traits in autism. M.L. Cuccaro.

2077W ‡ MAPK3 identified as candidate gene influencing schizophrenia and BMI in the 16p11.2 CNV region. L. Davis.

2078T Inherited mutations in Human Accelerated Regions (HARs) are associated abnormal social and cognitive behavior. R.N. Doan.

2079F CNV meta-analysis of major neurodevelopmental and neuropsychiatric disorders. J.T. Glessner.

2080W Identification of novel variants in autism spectrum disorder using whole-exome trio sequencing. R.S. Harripaul.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2081T ‡ Discovery of multifaceted genomic features as a promising approach to novel autism risk gene identification. Y. Ji.

2082F Gene expression profiling predicts clinical outcome in autism spectrum disorder: Confirmation of potential biomarkers and initial characterization of clinically homogeneous subgroups. F. Macciardi.

2083W Using the DGRP to identify gene networks associated with autism-like behaviors. L.T. Reiter.

2084T Epigenetic dysregulation of *DYRK1A* may have a role in ASD development in a discordant monozygotic twin pair. C. Sjaarda.

2085F Epigenetic factors and gene-environment interactions in autism: Prenatal maternal stress and the SERT gene. Z. Talebizadeh.

2086W Assembling the effects of genetic and environment risk factors in autism spectrum disorder using patient-derived neurons. K. Tammimies.

2087T Complete gene knockouts in autism spectrum disorder. T.W. Yu.

2088F Chronic psychosocial stress in mice alters brain myelination in a genetic background-dependent manner. I. Hovatta.

2089W Analysis of the genetics and heritability of a shared endophenotype in ADHD and FASD. J. Kapalanga.

2090T Genetic polymorphism and gene-environment interactions of dopamine receptor genes and nicotine dependence in the population of the Northwest Indian region. J. Kaur.

2091F Whole-genome sequencing to identify risk loci for nighttime eating in American Indians. C. Koroglu.

2092W RNAseq transcriptome study of schizophrenia in the MGS African American sample. A. Sanders.

2093T Minds, genes, and machines: Performance on online cognitive assessments is correlated with individual characteristics in 23andMe customers. O.V. Sazonova.

2094F Network analysis of gene polymorphisms in GABA, dopamine, cannabinoid, mu-opioid and alcohol metabolism pathways with alcohol dependence in scheduled class (SC) population of Punjab (Northwest India). R. Sharma.

2095W Understanding remission on venlafaxine in late-life depression: A genome-wide approach. V.S. Marshe.

2096T Abnormal expression of sonic hedgehog as a biomarker and therapeutic target for depression and suicide in bipolar disorder. M. Galdzicka.

2097F A multi-omics analysis towards understanding of the polygenicity in schizophrenia. P. Jia.

2098W Genetics of schizophrenia in Cooperative Studies Program #572. N. Sun.

2099T ‡ A genome-wide association study reveals a novel locus significantly associated with ADHD in African Americans and replicating in ADHD cases of European ancestry. B. Almoguera.

2100F ‡ Using genetic diversity from East Asia to improve the biological insight into schizophrenia. H. Huang.

2101W A missense variant in *PER2* is associated with delayed sleep phase disorder. T. Miyagawa.

2102T Schizophrenia and adult height show an inverse polygenic correlation within specific functional domains of the genome. A.P.S. Ori.

2103F Copy number variation analysis of psychiatric traits in a large population-based sample of youth. M. Zarrei.

2104W Genome-wide association study of cognitive flexibility assessed by Wisconsin Card Sorting Tests. H. Zhang.

2105T Integrating multi-omics data to boost the translation of GWAS to biology and therapeutics for schizophrenia. Q. Wang.

2106F Neurodevelopmental copy number variants and clinical risk: A pediatric record population study. K. Ahn.

2107W Potential role of rare variants in the genetics of tardive dyskinesia. A. Alkelai.

2108T ‡ Exome sequencing study of bipolar disorder in a genetically isolated population. L. Hou.

2109F Maternal antenatal depression and child socio-emotional outcomes: Investigating intervening child genetic risk for ADHD and biological pathways. L.M. Chen.

2110W ‡ Genome-editing of the *RERE* super-enhancer alters expression of genes in independent schizophrenia GWAS regions. C. Barr.

2111T Identification of ADHD risk genes in extended pedigrees by combining linkage analysis and whole-exome sequencing. M. Klein.

2112F Limited contribution of rare, noncoding variation to autism spectrum disorder from sequencing of 2,076 genomes in quartet families. D.M. Werling.

2113W Genetics of bipolar disorder in Cooperative Studies Program #572. M. Aslan.

2114T Genome-wide associations with generalised anxiety disorder symptoms and self-reported anxiety disorder: An investigation of unique and shared genomic contributions. K.L. Purves.

2115F Parent-of-origin and maternal effects in attention deficit hyperactivity disorder. D. Smajlagic.

2116W GWAS to drug: PTPRD as a drug target for addictions, RLS and neurofibrillary neurodegenerations. G.R. Uhl.

2117T Genome-wide association study of comorbid alcohol dependence and major depression. H. Zhou.

2118F Expanding the neurological and skeletal phenotypes of individuals with *de novo KMT2A* mutations. A.J.S. Chan.

2119W Polygenic burden analysis of longitudinal clusters of psychopathological features in a cross-diagnostic group of individuals with severe mental illness. E.C. Schulte.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

2120T The regulatory landscape of genetic variants associated with psychiatric disorders and neurodegenerative diseases. A. Amlie-Wolf.

2121F ‡ Common genetic variation contributes to risk of severe developmental disorders. M. Niemi.

2122W Link genetic variation to schizophrenia through cognitive and brain anatomical phenotypes. Z. Liu.

2123T DNA banking and genetic analysis of adverse drug reactions in the New Zealand healthcare setting. M.A. Kennedy.

2124F Association of *HTR3C*, *HTR3D*, *HTR5A*, and *HTR6* gene polymorphisms with treatment response to risperidone in Chinese schizophrenia patients. S. Qin.

2125W Risperidone-induced multi-dimensional phenotypic alteration in first-episode drug-naive schizophrenia patients: A longitudinal study of DNA methylation and neurophysiological phenotyping. C. Chen.

2126T Genetic and functional analysis of the GRIN2C gene as a candidate gene of schizophrenia. M. Cheng.

2127F ‡ Major depressive disorder and nausea and vomiting during pregnancy. Shared genetic factors? L. Colodro Conde.

2128W Whole genome sequence association analysis of tobacco use in the Trans-Omics for Precision Medicine Whole Genome Sequencing Program (TOPMed). G. Datta.

2129T Genetics of cognitive function in schizophrenia and bipolar disorder. P.D. Harvey.

2130F Transcriptional signatures of childhood onset schizophrenia in hiPSC-derived NPCs and neurons are concordant with signatures from post mortem adult brains. G.E. Hoffman.

2131W Copy number variation in Thai individuals with schizophrenia and schizoaffective disorder. N. Jinawath.

2132T Sex-stratified analysis of obsessive-compulsive disorder reveals minor differences in genetic architecture. E.A. Khramtsova.

2133F The transcriptional consequences of bipolar disorder polygenic risk and medication use. C.E. Krebs.

2134W Rare heterozygous mutation in glutamate receptor gene segregating in a schizophrenia family. P. Kukshal.

2135T Large meta-analysis of Scandinavian exome sequencing studies of schizophrenia. F. Lescai.

2136F Increased predicted *C4A* expression is associated with cognitive deficit in both schizophrenia and Alzheimer's disease. N.S. McCarthy.

2137W BBS1 M390R/M390R mice have impaired anxiety-like behavior. T. Pak.

2138T Initial results from the meta-analysis of the whole-exomes of 20,000 schizophrenia cases and 45,000 controls. T. Singh.

2139F Enrichment of treatable metabolic disease gene variants in a large cohort of schizophrenia, bipolar and major depressive disorder patients. V. Sriretnakumar.

2140W Integrated analysis supports *ATXN1* as a schizophrenia risk gene. B. Su.

2141T The role of miRNAs in 22q11.2 deletion syndrome. A.K. Victor.

2142F Identifying a shared regulatory background for neurodevelopmental disorders through meta-analysis of genomewide association studies. Z. Yang.

2143W ‡ A study of subthreshold hallucinatory experiences and thier relationship to genetic liability for schizophrenia. H. Young.

2144T Genetics of attention deficity hyperactivity disorder dimensions. T. Zayats.

2145F Gene-based meta-analysis of GWAS in African American and European ancestry populations implicates novel genes for PTSD. H. Zhang.

2146W Centrality pattern of susceptibility genes to complex disorders in functional specific protein-protein interaction sub-networks. T. Zhang.

2147T Rare human knockouts in consanguineous pedigrees aggregated with schizophrenia and bipolar disorder compared to matched healthy population controls. Q. He.

2148F Catalogue of 1 billion candidate ultra rare variants across 11670 Han Chinese individuals. S. Mangul.

2149W Association between TNF-α G-308A polymorphism and depression: A meta-analysis. T. Kim.

2150T Antidepressant effectiveness study in major depressive disorder in STAR*D patients. W. Guo.

2151F Analysis of leukocyte telomere length in children and adolescents at risk of developing mental disorders. G. Xavier.

2152W Preliminary analysis of whole genome sequences of simplex autism spectrum disorder. M.B. Neu.

2153T Autism spectrum disorder in the Amish: Exome sequencing in a founder population unveils novel coding variants. C.G. Tise.

2154F Mid-childhood adaptive function in individuals with 22q11.2 deletion syndrome is associated with immune-deficiency, but not oral/palatal or cardiac phenotypes. J.G. Baskin.

2155W PYROXD1 is responsible for cellular functions in myoblasts and homozygous missense mutation in PYROXD1 causes limb-girdle muscular dystrophy among patients from Saudi Arabian cohort. M. Saha.

2156T ‡ Using electronic health records for the identification of novel genes associated with adverse drug reactions. L. Milani.

2157F The intersection of type 2 diabetes and cognitive impairment in Mexican Americans: Insights from the mitochondria. T. Silzer.

2158W Expanding the *RTN4IP1/OPA10* genotype-phenotype correlation: From isolated optic neuropathy to severe mitochondrial encephalopathy. I. Barbosa.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2159T Novel mutation in the *MT-ND4* gene at low heteroplasmy level likely associated with a mild phenotype: A case report. A. Gonzalez Garcia.

2160F A hypertension-associated mtDNA mutation alters the tertiary interaction and function of tRNALeu(UUR). M. Guan.

2161W Identification of genetic causes for age-related hearing loss. S.H. Blanton.

2162T Cochlear nerve deficiency presenting as auditory neuropathy spectrum disorder. A. Pandya.

2163F Association of PMEL missense variants with ocular pigment dispersion and pigmentary glaucoma. M.A. Walter.

2164W Gene-set enrichment analysis identifies pathways involved in tinnitus. E. Fransen.

2165T New risk loci for primary open-angle glaucoma. P. Gharahkhani.

2166F Identifying genes that underlie eye disorders and vision loss using predicted gene expression. J.B. Hirbo.

2167W Exome sequencing identifies susceptibility genes for chronic central serous chorioretinopathy. R.L. Schellevis.

2168T Optimizing accurate classification of electronic health record case control status for age-related macular degeneration in the Million Veteran Program. S.K. Iyengar.

2169F Identification of rare sequence variants in genes involved in focal adhesion and Wnt signaling pathways in keratoconus human corneas. J.A. Karolak.

2170W Updated carrier rates for deafness-inducing mutation c.35delG (*GJB2*) in Russia and common haplotypes associated with c.35delG in Siberia. O. Posukh.

2171T ‡ Forty novel genetic loci associated with intraocular pressure in a large multi-ethnic genome-wide association study. H. Choquet.

2172F A transethnic genome-wide association study identifies five novel genetic loci associated with primary open angle glaucoma. E. Jorgenson.

2173W Trial to make the combined genotyping to detect high-risk individuals for cold medicine related Stevens-Johnson syndrome (CM-SJS) with severe ocular complications (SOC). M. Ueta.

2174T Loss of ELOVL6, a fatty acid elongase, rescues ER stress-induced apoptosis in model of retinitis pigmentosa. R.A.S. Palu.

2175F Evaluation of genetic polymorphisms in the determination of optic disc parameters and circumpapillary retinal nerve fiber layer thickness in normal individuals in a sample of the Brazilian population. M. Atique-Tacla.

2176W Lineage-specific linkage analysis localizes novel rare variant-driven genomic loci for the glaucoma endophenotype of cup to disc ratio in a large extended pedigree from Nepal: The Jiri Eye Study. M.P. Johnson.

2177T *ANGPT1* association with adult-onset primary open angle glaucoma. J.N. Cooke Bailey.

2178F Common variants in *KLHL2* and *C4orf50* are associated with poorer anti-VEGF treatment response in age-related macular degeneration. O. Garcia Rodriguez.

2179W Additive effects of genetic variants associated with primary open-angle glaucoma. F. Mabuchi.

2180T Genome-wide gene expression profiling in the retina of common marmosets exposed to hyperopic or myopic defocus reveals large-scale sign-of-defocus-specific changes in gene expression, pathway switching, and strong left-right eye yoking. A. Tkatchenko.

2181F Using GWAS data to identify copy number variants associated with orofacial clefts. A.L. Petrin.

2182W Fluctuating dermatoglyphic asymmetry and familial recurrence of cleft lip/palate in a high-prevalence cluster of South America. J. Ratowiecki.

2183T Robinow, Ter Haar, Teebi or a new syndrome? Complex genotype with distinctive craniofacial features. F. Uysal.

2184F Clinical aspects associated with orofacial clefts in patients of smile operation in a Colombian population. J. Martinez.

2185W Exploring the impact of sex-specific genetic effects on orofacial clefting. J. Carlson.

2186T ‡ Identification of 16q21 as a modifier locus for orofacial cleft phenotypes. E.J. Leslie.

2187F Ballooning of redundant myelin sheaths in DS may resolve with Ca^{2+} as seen in the mouse. A.N. van Hoek.

2188W Variants in the degron motif of AFF3 cause a multi-system disorder with skeletal dysplasia and severe neurologic involvement. N. Voisin.

2189T Microdeletion of Xp22 encompassing *SHOX* and *ARSE*, showing incomplete penetrance and variable expressivity. B. Simpson.

2190F ‡ CRISPR/Cas9 engineering to generate an isogenic model of the 3-Mb 22q11.2 syndromic deletion. Y.T. Lin.

2191W Exome sequencing-based pipeline identifies functional variants within chromosome 1 associated with the risk of non-syndromic cleft palate. S. Beiraghi.

2192T Genotype-phenotype correlation in WAGR syndrome: Large deletion of chromosome 11p in a patient with classical and non-classical symptoms of the syndrome. P.N. Moretti.

2193F Syndromic cleft genes implicated in non syndromic forms: Towards translational phenotypes? B. Demeer.

2194W Genetic variants in a patient with pancreatitis after propofol administration. J.B. Cordero.

2195T ‡ Whole-exome analysis of late-onset Alzheimer's disease reveals novel candidate genes involved in cognitive function. C. Preuss.

2196F African haplotypic background mitigates the effect of *APOE* ε4 risk allele in Alzheimer disease. F. Rajabli.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

2197W Dissecting the sex-specific basis of APOE ε4 allele effect on longevity. P.R.H.J. Timmers.

2198T Age-related changes in white blood cell gene expression associated with skeletal fragility. E. Quillen.

2199F The genomic basis of human lifespan. P. Joshi.

2200W Genomics of the aging hematopoietic system. E. Bader.

2201T Identifying genetic variants associated with leukocyte telomere length in African Americans. A. Little.

2202F Meta-analysis of GWAS elucidates genetic architecture of dental caries. D. Shungin.

2203W Understanding relationships between longevity and physical senescence using Mendelian randomization approach. S. Ukraintseva.

2204T Allele-specific expression in healthy centenarians. L.C. Tindale.

2205F The PhenX Toolkit: Adding a resource for geriatric research. M. Phillips.

2206W Does parent-to-offspring transmission of telomeres contribute to telomere length heritability in humans? D.A. Delgado.

2207T ‡ Telomere length across many human tissues. K. Demanelis.

2208F GWAS replicates known asthma variants validating self-reported childhood asthma diagnosis in the COPDGene Study. L.P. Hayden.

2209W Newborn metabolomics and risk of episodic wheezing in childhood: Findings from the INSPIRE study. K.K. Ryckman.

2210T Influence of guideline adherence and ADRB2 SNPs inpredicting exacerbation frequency in asthma patients. A. Santani.

2211F A simulated evaluation of data-driven algorithms for addressing clinical heterogeneity in complex traits. A.O. Basile.

2212W ‡ Disease associations of the zinc receptor GPR39 in the DiscovEHR Study cohort. G.E. Breitwieser.

2213T Low frequency genetic variation in *TP53* is associated with final head circumference. B. St Pourcain.

2214F Pathogenic and likely pathogenic mutations identified in apparently normal individuals of Arab descent. A. Alkhateeb.

2215W Widespread prevalence of a CREBRF variant amongst Māori and Pacific children is associated with weight and height in early childhood. S.D. Berry.

2216T The impact of MUC5B and KLK4 genes on dental caries. T. Cavallari.

2217F Leveraging tissue specific omics data to estimate the disease/traits-related tissues. R. Chen.

2218W Will big data close the missing heritability gap? G.A. de los Campos.

2219T ‡ Performance of polygenic scores across ancestrally diverse populations. L. Duncan.

2220F ‡ Two novel loci detected and 10 known loci confirmed for estimated glomerular filtration rate in over 56,000 African Americans: The Million Veteran Program. T.L. Edwards.

2221W Slit2-Robo1 signaling may play a role in spontaneous preterm birth. M. Karjalainen.

2222T Adult height and risk of cardiometabolic disease. E. Marouli.

2223F GWAS of early childhood caries in an Appalachian population. E. Orlova.

2224W Novel genes identified by integrating genome-wide association analysis with transcriptomics in severe chronic obstructive pulmonary disease and quantitative emphysema. P. Sakornsakolpat.

2225T Ayurveda based deep phenotyping, a likely game changer for gene hunt in complex traits. B.K. Thelma.

2226F Genomic features of loci associated with multiple complex traits in genome-wide association studies. Y.-F. Wang.

2227W A novel relationship between GWAS-identified sleep traits loci in sleep duration variation in healthy adults. X. Xu.

2228T Structural variation influencing complex traits and metabolomic measurements. A. Sabo.

2229F Genetic polymorphisms of LIN28B and MKRN3 in association with precocious puberty. K. Lee.

2230W Finland, combining the population isolate structure with nationwide health care data for gene discovery. A. Palotie.

2231T Whole exome sequencing reveals a novel candidate gene, *HSPA1L*, for spontaneous preterm birth. J.M. Huusko.

2232F Whole exome sequencing analysis in severe chronic obstructive pulmonary disease. D. Qiao.

2233W Enhanced methods to investigate the role of *Trans*-eQTL to complex traits. C. Giambartolomei.

2234T Connection of RAB8A and MED16 with implications on 5'-deoxy-5-fluorouridine response. C.M. Murtagh.

2235F Cross-altitude analysis suggests a turning point at the elevation of 4,500m for polycythemia prevalence in Tibetans. C. Cui.

2236W Polymorphisms associated with skin, hair and eyes color for forensic phenotyping purposes in Brazilian population. C. Fridman.

2237T Whole genome sequencing in severe chronic obstructive pulmonary disease. D. Prokopenko.

2238F GWAS of fingerprint patterns. E. Feingold.

2239W Replication of HLA association with podoconiosis in diverse Ethiopian ethnic groups. T.T. Gebresilase.

2240T Fetal genome-wide meta-analysis of gestational age and preterm delivery. X. Liu.

2241F Genome-wide association meta-analysis of usual sleep duration in 31,703 Japanese population. T. Nishiyama.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2242W Genome-wide association study of complement activity pathways: The Cooperative Health Research in South Tyrol (CHRIS) study. D. Noce.

2243T GWAS reveals loci associated with velopharyngeal insufficiency. J. Roosenboom.

2244F A genome-wide association study of anorexia nervosa suggests a risk locus implicated in dysregulated leptin signaling. X. Wang.

2245W Genome-wide association studies of eye color in Han Chinese and Uyghur populations. L. Wang.

2246T Pinpointing GWAS signals: Indels vs. SNPs. S.A. Gagliano.

2247F Mutual allelic constraint in a human taste receptor gene cluster shows evidence of gene-gene interaction and may influence human reproductive partner compatibility. R. Subaran.

2248W High-risk genetic variants in genes involved in endothelial cell glycocalyx function in thrombotic storm. K. Nuytemans.

2249T Genetic typing of DC-SIGN in recurrent vulvovaginal candidiasis. N. Kalia.

2250F The Million Veteran cohort: A high quality and diverse resource of genetic data for large-scale analysis to enable personalized medicine. Y. Shi.

2251W Gut microbiota composition in children and adults: *Bacteroides* vs *Blautia*. D. Radjabzadeh.

2252T Genome-wide association study of cranial vault shape reveals novel loci at 15q26.3 and 17q11.2. M. Lee.

2253F Pharmacogenomic study of antithyroid drug-induced cutaneous reaction. P. Chen.

2254W Characterization of *CYP2D6* by whole genome sequencing. A.L. Halpern.

2255T Translating pharmacogenetics: An electronic phenotyping algorithm and survey study of diverse BioMe biobank patients treated with ACE inhibitors. H. Naik.

2256F Defining the opportunity: The IGNITE CPIC Prescribing Study. L. Wiley.

2257W ‡ Phenotypic consequences of the genetic regulation of expression of the Mendelian disease gene *CFTR*. X. Zhong.

2258T Designing of an efficient genotyping chip for discovery and pan-disease screening in the VA's Million Veteran Program. S. Pyarajan.

2259F Integration of a tool for patient self-assessment within primary care to enable precision prevention. K. Rageth.

2260W High-density imputation identifies rare and low-frequency variants associated with human height in Japanese population. M. Akiyama.

2261T Novel genes and mutations in patients affected by recurrent pregnancy loss. P. Quintero-Ronderos.

2262F Predictive modeling of gene expression in ethnic minorities. K. Keys.

2263W Genome-wide association study of asthma in individuals of mixed African ancestry reveals a novel association with markers on chromosome 2q14. S. Chavan.

2264T Association study of placebo-treated patients from 35 clinical trials suggests genetic contribution to the placebo response. A. Wuster.

2265F The Precision Medicine Initiative All of Us Research Program: Innovative access to unprecedented data. A. Ramirez.

2266W Data-driven approach to dietary phenotypes for nutrigenomics in UK Biobank. J.B. Cole.

2267T ‡ A broad survey of the relationship between autozygosity and fitness-related and sociodemographic traits in the UK Biobank. E.C. Johnson.

2268F Fine-mapping of tobacco and alcohol associated loci in around 900K participants. M. Liu.

2269W Genetic predictors of biomarker levels derived from prospective epidemiologic cohorts applied to electronic health records to identify new biomarker-disease associations. J.D. Mosley.

2270T New insights into the genetic architecture of complex human traits from Bayesian mixture model analyses in a large dataset. J. Sidorenko.

2271F ‡ Rare coding variant association analysis for anthropometric traits using more than 25,000 exome-sequenced samples from ExAC. S. Vedantam.

2272W Copy number variation associated with white blood cell phenotypes in the eMERGE Network. M.R. Palmer.

2273T Utilizing protein quantitative trait loci to identify functional candidates from genome-wide association studies. S. Sivertson.

2274F Integrative fine-mapping of 34 complex phenotypes. R. Johnson.

2275W Probabilistic assignment of causal genes at transcriptome-wide significant risk loci. N. Mancuso.

2276T The Macaque Genotype and Phenotype (mGAP) database: a novel resource to support genetic disease model development and translational research in nonhuman primates. B. Ferguson.

2277F ‡ Examining sex differences in genetic effects on subcortical brain structures. K. Grasby.

2278W *In silico* evaluation of a more comprehensive pharmacogenetic profile for predicting opiate metabolizer phenotype. F.R. Wendt.

2279T ‡ Drug target genes associated with clinical phenotypes in the genetically isolated population of Finland are more likely to succeed in pharmaceutical development. R. March.

2280F ‡ Transferability of polygenic risk prediction across diverse and admixed populations. A.R. Martin.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Evolution and Population Genetics

2281W Genomic characterization of the immunoglobulin heavy chain variable gene locus in individuals of African, Asian, and European descent reveals signatures of elevated haplotype diversity. O. Rodriguez.

2282T Isolated population from Runta, Boyacá: Findings related to founder effect of the nonsense variant of the HGSNAT gene. L.J. Torres.

2283F Local adaptation shaped the genetics of psychiatric disorders and behavioral traits in European populations. R. Polimanti.

2284W Detecting variation maintained by balance between recurrent mutation and selection in human populations. N. Koelling.

2285T New favored haplotype alleles in human adaptation to high altitude in Andes. T. Stobdan.

2286F Genetic structure of pre-Columbian remains of the Malambo culture in the Lower Magdalena, Colombia. M. Noguera.

2287W Comparative whole genome variation and architecture in a pediatric African American and Caucasian dataset. P. Sleiman.

2288T *GCH1* plays a role in high altitude adaptation of Tibetans. Y. He.

2289F Improving genotype imputation in population isolates using identity by descent. M. Abney.

2290W Consanguinity, IBD, and ROH: Detangling their complex relationship. A. Severson.

2291T Evolutionary rate and the human transcriptome: Function, mechanism, and disease genes. P. Evans.

2292F EP300 contributes to high altitude adaptation of Tibetans by regulating nitric oxide production. X.B. Qi.

2293W De novo genomic assembly and assessment of inbreeding in a severe bottleneck population. A.F. Scott.

2294T Dietary adaptation of *FADS* genes varied geographically in Indian populations. D. Wang.

2295F ‡ Detect co-evolution of genes in admixed populations with genome-wide data. M. Zhou.

2296W Where is Brazil? Placing admixed Brazilian populations in a global genetic map. A. Arcanjo Silva.

2297T *HLA-G* and *HLA-A* extended haplotypes in a Brazilian population sample: The close relationship between *HLA-G* promoters and *HLA-A* coding alleles. E.C. Castelli.

2298F Rewriting the genetic history of Austro-Asiatic communities in South and Southeast Asia. M.E. Phipps.

2299W Genetic differentiation of Hispanics using ancestry informative markers. C.H. Setser.

2300T One step for study of transposable element-associated structural variations (TASVs) using de novo assembled Korean genome. S. Mun.

2301F Haplotype map of Russian population. I.V. Evsyukov.

2302W Ancestry Hub: For whole-genome local ancestry analysis. S. Jiang.

2303T Relationships between the regulation of gene expression, mutational burden, and recombination in a large population cohort. H. Edgington.

2304F Tracing maternal lineage of Austronesian-speaking Melanesians and Micronesians in the Solomon Islands. M. Isshiki.

2305W Exploration of the ancestral genetic landscape of the Arabian Peninsula. D. Platt.

2306T Evolutionary patterns of long non-coding RNAs with coding capacity of oligopeptides. W. Lin.

2307F Ohana: Detecting selection in multiple populations by modelling ancestral admixture components. J.Y. Cheng.

2308W Peruvian Genome Project: A new reference of Andean haplotypes to study genome populations. H. Guio.

2309T MixFit: Methodology for computing ancestry-related genetic scores at the individual level and its application to the Estonian and Finnish population studies. T. Haller.

2310F ‡ Discovering rare variants and deciphering a population structure of 386 Mongolian individuals by whole-genome sequencing. C. Kim.

2311W Large-scale whole genome sequencing of the Estonian population reveals new insights into population history and recent natural selection. M. Metspalu.

2312T Association of *VASP* polymorphisms and infectious disease burden in global populations and identification of candidate resistance haplotypes in sub-Saharan Africa. M. Phun.

2313F Mapping the genetic diversity in indigenous Malays populations and cosmopolitan Malay. W. Saw.

2314W Mitogenomes sequencing: Reveals huge diversity in human maternal lineages of Jammu and Kashmir, India. I. Sharma.

2315T Recent changes in contemporary effective population size from identical by descent segments. A. Urnikyte.

2316F Selection analysis in Chileans identify adaptation signals in Native Americans, highlighting regulatory processes. L. Vicuña.

2317W Analysis and findings in high-depth target sequencing of over 20000 individuals in China. H. Xu.

2318T Standardized visualization of demographic history. Y. Zhou.

2319F Principal components analysis with sensible weighting of sequencing variants: Improved inference of fine scale population structure with whole genome sequencing data. T.A. Thornton.

2320W Assessing human diversity patterns using in-silico discovered Alu and LINE-1 mobile insertion elements in the Simons Genome Diversity Project. S. Watkins.

2321T Archaic-genome-agnostic detection of introgressed segments. S.R. Browning.

2322F ‡ Using ancient DNA from Sardinia to assess population stability from the Neolithic to present. J. Marcus.

2323W Impacts of European colonization on an indigenous community in British Columbia. A.C. Owings.

2324T Tracing the origin of ancient polynesian human genomes across the Pacific. P. Salazar-Fernandez.

2325F MHC-dependent mate selection within the Health and Retirement Study (HRS). Z. Qiao.

2326W Pseudogenes in the mouse lineage: Transcriptional activity and strain-specific history. P.M. Muir.

2327T Inference of allele-frequency trajectory histories from present genomes. Y. Field.

2328F SG10K: Insights into the genetic architecture of Singaporeans. C. Bellis.

2329W SeleDiff: A scalable tool for testing and estimating selection differences between populations. X. Huang.

2330T Distribution of common and rare variants in an underrepresented population in public genomic databases and the possible impact in precision medicine. C.S. Rocha.

2331F Deep learning for reference-free inference of archaic local ancestry. A. Durvasula.

2332W Variation and genetic control of mutation rates in house mice. B.L. Dumont.

2333T Patterns of shared signatures of recent positive selection across human populations. K.E. Johnson.

2334F The landscape of genetic variation in Estonians. M. Kals.

2335W Gene expression predictive performance varies across diverse populations. L.S. Mogil.

2336T ‡ Adaptive eQTLs in human populations. M. Quiver.

2337F ‡ A comparative study of endoderm differentiation in humans and chimpanzees. L.E. Blake.

2338W Determining the distribution of deleterious variation in population isolates using local ancestry and pedigree data. J. Mooney.

2339T The composition and intensity of *de novo* mutations in the Lithuanian exome. L. Pranckėnienė.

2340F Testing for local adaptation in populations of *Drosophila melanogaster* for olfactory receptor genes. V. Ramesh.

2341W Genome-wide population analysis of 2,543 microsatellites and STR-SNP haplotypes reveals a novel class of highly diverse polymorphisms. G. Shin.

2342T Inverse correlation between mutational and selective forces in human coding regions with distance from gene ends leads to opposite patterns of synonymous and non-synonymous variant prevalence. Y. Waldman.

2343F Patterns of genetic variation within the Genome Russia Project. S. OBrien.

2344W 1000 high coverage whole-genome sequences representative of the Taiwanese population from Taiwan Biobank. M. Su.

2345T NHGRI Sample Repository for Human Genetic Research: Cell lines and DNA from the 1000 Genomes and HapMap collections. E.M. Kelly.

2346F Increasing signal and refining population annotations using a common haplotypes co-association network. L. Doroud.

2347W ‡ Partitioning heritability of low-frequency variants reveals relative strength of negative selection across functional annotations. S. Gazal.

2348T *HLA-A* extended promoter and coding variability in a Brazilian population sample by using massivelly parallel sequencing. T.H.A. Lima.

2349F *MC1R* regulatory and coding polymorphisms and pigmentation in an admixed population from Brazil. C.T. Mendes-Junior.

2350W Allele frequencies of pathogenic single nucleotide variants in a Japanese population based on a whole-genome reference panel of 2,049 individuals. Y. Yamaguchi-Kabata.

2351T ‡ Substantial fraction of genes under recessive selection illuminates a missing component of human variation in population genetics and model organism studies of human disease. D.J. Balick.

2352F Rapid detection of identity-by-descent from whole genome sequence data. J.E. Hicks.

2353W Mutation rate estimation from population data. X. Tian.

2354T ‡ Extremely rare variants reveal patterns of germline mutation rate heterogeneity in humans. S. Zöllner.

2355F Detecting long-term balancing selection using allele frequency correlation. K. Siewert.

2356W Recurrent adaptation of different haplotypes in *FADS* genes to plant-based and animal-based diets in a diverse worldwide set of extant and extinct human populations. K. Ye.

2357T Genetic origins of Easter Island and remote Oceania. A. Ioannidis.

2358F A web-based initiative to accelerate research on African ancestry in the Americas. M.E. Moreno.

2359W Origin and affinities of Lakshadweep Islanders. M.S. Mustak.

2360T The genetic substructure of the Japanese population: Results from the Japan Multi-Institutional Collaborative Cohort Study. M. Nakatochi.

2361F IBD detection at biobank-scale. A. Naseri.

2362W Exploring the demographic and admixture history of Central Mexico. A.W. Reynolds.

2363T Signatures of multiple-mergers coalescence in genomic diversity data. D.P. Rice.

2364F Studying global variation of gene flow using geo-referenced genetic data. S. Song.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

2365W Genetic structure in Brittany highlights physical and cultural limits. J. Giemza.

2366T The Irish DNA Atlas: Revealing fine scale population structure and history within Ireland. E.H. Gilbert.

2367F Characterizing heterogeneity within fine-scale population structure. A. Hippen Anderson.

2368W The genomic health of ancient hominins. J. Lachance.

2369T FastNGSadmix: Admixture proportions and principal component analysis of a single low-depth sequencing sample. E. Jørsboe.

Molecular and Cytogenetic Diagnostics

2370F Assessment of BAP1 germline and somatic alterations in uveal melanoma. M.H. Abdel-Rahman.

2371W ‡ Paperwork matters! The importance of clinical phenotype information in variant interpretation. M. Anderson.

2372T Mutational spectrum in BRCA1 and BRCA2 genes in Colombia. I. Briceno.

2373F Retrospective analysis of 36 fusion genes in 6170 patients of *de novo* acute leukemia and myeloid neoplasms. X. Chen.

2374W Paternal inheritance of *BRCA* mutations in Algerian hereditary breast/ovarian cancer families. F. Cherbal.

2375T Novel acute myeloid leukemia amplicon panel for uniform detection of all target genes, Including *CEBPA*. C. Clear.

2376F Comparison of variant classification algorithms incorporating clinical and family history for breast and ovarian cancer. J. Clifford.

2377W A t(18;22)(q21;q11) involving *IGL/BCL2*, a rare event in in chronic lymphocytic leukemia. A. Dowiak.

2378T Differential expression of kallikreins as prognostic markers in recurrent prostate cancer. E. Guzel.

2379F A 34-gene, NGS assay for common hereditary cancers reveals a high percentage of variants of unknown significance in the *POLD1* and *POLE* genes. D. Hodko.

2380W Hereditary cancer panel results identify gaps in knowledge of cancer risks and limitations in current guidelines. H. LaDuca.

2381T TaqMan dPCR liquid biopsy assays targeting the TERT promoter region. M. Laig.

2382F Clearance of plasma *EGFR* mutations as a predictor of outcome following osimertinib treatment. A. Markovets.

2383W Digital spatial profiling platform allows for spatially-resolved, multiplexed measurement of solid tumor protein distribution and abundance in FFPE tissue sections. C. Merritt.

2384T Clinical and molecular findings correlation in patients with hereditary cancer syndromes: Experience of a Colombian reference laboratory. T. Pineda.

2385F Polymorphic SNPs in breast cancer using molecular studies in Indian population. M. Rao.

2386W Frequency of pathogenic and rare variants of uncertain significance in cancer patients and control cohort. K.M. Rocha.

2387T Hereditary cancer risk testing within a Colombian cohort reveals high incidence of Lynch syndrome. J.A. Rugeles.

2388F ‡ Use of functionally classified 207 BRCA2 missense variants to calibrate sequence-based computational prediction models. H. Shimelis.

2389W Concurrent multiple molecular alterations involving *ALK*, *RET*, *ROS1* and *MET* in non-small cell lung cancer. Z. Tang.

2390T Towards prediction of platinum treatment response in ovarian cancer using machine learning approaches. A. Vladimirova.

2391F Functional analysis of BRCAness in female cancers. M.P.G. Vreeswijk.

2392W Detection and quantitation of M-BCR and m-BCR fusion transcripts by pico-liter digital PCR. J. Woolworth-Hirschhorn.

2393T Frequent detection of chromothripsis in acute myeloid leukemia with complex karyotype and marker chromosomes. J. Lee.

2394F Tetraploidy and near tetraploidy in acute myelocytic leukemia and myelodysplastic syndrome: A report of seven new cases. J. Liu.

2395W A t(3;8)(q26.2;q24) involving the *EVI1 (MECOM)* gene on 3q26 in a case of acute myeloid leukemia preceded by polycythemia vera. K. Liu.

2396T Identification of cancer-associated copy number variations through integrated genomic analysis. M. Luo.

2397F Characterization of interstitial deletions of 9q in ten AML cases. Z. Qi.

2398W Cytogenetic characterization of Richter transformation in chronic lymphocytic leukemia. G. Tang.

2399T A plasma cell myeloma case with an abnormal clone showing a t(8;22)(q24.1;q11.2) within the context of a hyperdiploid complex karyotype. C.A. Tirado.

2400F A pediatric B-ALL with doubled near-haploidy by chromosomal microarray and negative MRD at day 29 of induction: Case report. J. Xu.

2401W A novel *XPA* gene mutation (c.773delG, p.R258Lfs*11) in two siblings with Xeroderma Pigmentosum. B. Balta.

2402T Implementation of a multi-gene panel in selected Greek breast and ovarian cancer patients reveals loss-of-function mutations in multiple genes. F. Fostira.

2403F Screening for rearrangements in *RB1* gene/ 13q14 through real-time PCR. R.M. Freitas.

2404W Molecular insights into the missing heritability of familial ovarian cancer. J. Stafford.

2405T Current laboratory practices in *BRCA1* and *BRCA2* testing: Results from an international survey of 86 laboratories. A. Toland.

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2406F Hyb & Seq™: The next generation of simultaneous RNA and DNA detection in liquid biopsy without the library and sample preparation bottlenecks. J. Beechem.

2407W Optimizations in target enrichment and bioinformatics enable sensitive detection of copy number variations in targeted NGS. J. van den Akker.

2408T Hereditary cancer risk testing of 11,570 individuals with a multi-gene panel. J. Ji.

2409F What's in a VUS rate? Simulated VUS calculations for hereditary cancer genes in a general population using population frequency data and ClinVar submissions. K.E. Kaseniit.

2410W Integration of calibrated functional assay data into *BRCA1* VUS evaluation. B.A. Thompson.

2411T ‡ Mate-pair sequencing provides advanced molecular characterization of genomic rearrangements in B-Chronic Lymphocytic Leukemia (CLL) and non-Hodgkins lymphoma. S.S. Smoley.

2412F Loss-of-function *POLE* and *POLD1* variants may not be associated with early-onset colon cancer/polyposis. E.K. Flynn.

2413W Library-free, targeted sequencing of native genomic DNA and RNA from FFPE samples using Hyb & Seq™ technology – the Hybridization-based Single Molecule Sequencing System. D. Kim.

2414T The prevalence of mosaicism in common cancer susceptibility genes from 232,328 individuals undergoing sequential testing. T. Slavin.

2415F Somatic driver mutations enhance survival prediction in familial chronic lymphoid leukemia. W. Zhou.

2416W Combined mutation and CNV detection by targeted next-generation sequencing in uveal melanoma. A. de Klein.

2417T Applying synthetic long reads to a custom Lynch Syndrome NGS panel to overcome pseudogene interference in *PMS2*, detect structural variations, and enable allelic phasing. C. Kao.

2418F ‡ Tumor characteristics provide evidence for mismatch repair (MMR) variant pathogenicity. S. Li.

2419W A case report of a rare germline SVA transposition event in *TP53*. W. Cheng.

2420T Diagnostic yield and mutation spectrum of multigene panel testing for hypertrophic cardiomyopathy. L. Qin.

2421F Prevalence of RASopathy gene mutations in patients who have had multi-gene panel testing for cardiomyopathy. W. Zhang.

2422W Molecular aproach of targeted next generation sequencing of 68 genes involved in cardiac arrhythmias of 148 unrelated patients. B. Turkgenc.

2423T ‡ Involvement of mtDNA variants in patients with cardiac manifestation. H. Cui.

2424F A genetic backround in Czech patients with inherited cardiomyopathies. P. Cibulkova.

2425W Post-mortem cytogenomic study of Brazilian patients reveals the CNVs connection to complex congenital heart defects. F.A.R. Madia.

2426T Molecular review of Polish patients with Smith-Lemli-Opitz syndrome. P. Halat.

2427F Reconciling newborn screening and genomic results to detect partial biotinidase deficiency: A BabySeq Project case report. J.B. Murry.

2428W One novel 2.43Kb deletion and one single nucleotide mutation of INSR gene in a Chinese neonate with Rabson-Mendenhall syndrome. L. Yang.

2429T Concurrent determination of *ABO RhD* blood types and the HIV-1 resistance marker *CCR5* deletion via rapid multiplex PCR and capillary electrophoresis-based genotyping. E. Schreiber.

2430F Chromosome microarray analysis as a first-tier test for cytogenetic abnormalities in patients with myelofibrosis. J. Mazzeu.

2431W Mutation profiling of 16 candidate genes in de novo acute myeloid leukemia patients. H.X. Liu.

2432T Xq22.1 contiguous deletion syndrome as a diagnostic challenge: Detection of a 17 kb deletion ends 30-year diagnostic odyssey. G. Raca.

2433F HLA typing using capture based next generation sequencing. S.K. Lai.

2434W A patient with hereditary pyropoikilocytosis caused by a combination of a novel in-frame deletion and a common functional but non-pathogenic allele, α^{LELY}, in *SPTA1*. T. Goto.

2435T Analysis of germ line predisposition in Chinese children with bone marrow failure. N. Dai-jing.

2436F The study of the applicability of three statistical methods in IGH immune repertoire analysis. F. Wang.

2437W 47,XY,+21/46, XX chimera identified in an infant with ambiguous genitalia without Down syndrome features. C. Charalsawadi.

2438T Novel approach using NGS assay for detection of mutations of the *CYP21A2* gene. G. Bennett.

2439F NEBNext Direct CFTR enrichment panel: Interrogation of genetic variants by Illumina sequencing. S. Adams.

2440W ‡ Novel pathogenic variants are routinely detected even in extensively-sequenced genes, such as CFTR. N. Faulkner.

2441T Mutations in the *DHX37* gene identified by whole-exome sequencing (WES) are a novel cause of the embryonic testicular regression syndrome (ETRS). T.E. da Silva.

2442F Diagnostic utility of whole-exome sequencing for chronic kidney disease. E. Groopman.

2443W De novo unbalanced insertional translocation, der(X)ins(X;5)(q?13;q12.3q13.1) in an adult female patient with developmental delay and ovarian insufficiency identified by DNA MicrorrayCGH and FISH. M. Pitch.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

2444T Pathogenic variants and variants of uncertain significance in autosomal dominant polycystic kidney disease (ADPKD) causative genes are commonly found in early-onset PKD patients: 2.5-year experience in a CAP/CLIA diagnostic laboratory. W. Chen.

2445F Diagnostic strategy in segmentation defect of the vertebrae: A retrospective study of 73 patients. M. Lefebvre.

2446W Small 17p13.3 duplication including *BHLHA9* in a Brazilian family with incomplete penetrance of split-hand/foot malformation. W.A.R. Baratela.

2447T Novel pathogenic variants in craniosynostosis genes identified by NGS. E. König.

2448F ‡ Detection of mosaic copy-number variation from whole-exome sequencing using XHMM and custom SNP approach. A. Sorlin.

2449W ‡ Diagnosing connective tissue disorders by clinical exome sequencing. H. Cheng.

2450T Mendelian disorders of cornification in Iran: Spectrum of clinical and genetic characteristics in a cohort of 189 consanguineous families including a new entity of ichthyosis follicularis with profound bilateral deafness and leukonychia totalis. L. Youssefian.

2451F Multigene next generation sequencing panel identifies pathogenic variants in patients with unknown subtype of epidermolysis bullosa: Subclassification with prognostic implications. A. Saeidian.

2452W Exome sequencing in 170 patients with diverse ataxia-related phenotypes identifies the genetic basis of disease in over 50%. A. Knight Johnson.

2453T Incidentaloma in neurogenetics: Pathogenic variant in NSD1 in a patient with spinocerebellar ataxia. H.M. Velasco.

2454F Intracellular FmRPpolyG-HSP70 complex: Possible use as biochemical marker of FXTAS. G. Bonapace.

2455W The AAGAAAG duplication at nucleotides 2023-2029 of SCN8A gene of EIEE13 (early infantile epileptic encephalopathy-13) presenting with no epilepsy, but variable expression of intellectual disability, ADD/ADHD and autism in the same family. M. Hajianpour.

2456T Further investigation of variants discovered in an early onset dementia cohort: Additional family member sequencing. S.A. Bucks.

2457F Comparison of the diagnostic yield of multi-gene panels for neuromuscular disorders. A. Gruber.

2458W Identification of a novel *de novo* nonsense mutation of the *NSD1* gene in monozygotic twins discordant for Sotos syndrome. J. Han.

2459T Novel myopathic phenotype due to a newly detected stop-loss mutation in *MYH7* gene. K. Sumegi.

2460F Whole exome sequencing: An effective and comprehensive genetic testing approach for leukodystrophy. F. Zou.

2461W Copy number analysis using next-generation sequencing: Comprehensive genetic testing and its application to neuromuscular and epilepsy panels. A. Entezam.

2462T Combining repeat expansion testing with phenotype based NGS panels provides significant diagnostic benefit. H.A. Marton.

2463F ‡ Diagnostic yield for neurological and neuromuscular disorder testing via high-depth multi-gene panel analysis with integrated sequence and copy number detection. T. Winder.

2464W Millder-Dieker syndrome: Clinical, radiological, and molecular characterization. A. Cortes.

2465T Clinical application of whole exome sequencing in patients with uncertain neurological disorders. Y. Lee.

2466F Molecular diagnosis of Colombian patients with myopathies through next generation sequencing panel. R. Garcia-Robles.

2467W ‡ A novel approach distinguishing the *SMN1* and *SMN2* genes in spinal muscular atrophy (SMA) using a linked-read NGS custom panel. R. Pellegrino.

2468T Detection rate of chromosomal microarray in individuals with ADD/ADHD. A.L. Baxter.

2469F Repeat expansions that cause central nervous system disorders present challenges for long-read sequencing technologies. M.T.W. Ebbert.

2470W ‡ Attacking a VUS from multiple angles: An integrated and functional approach for reclassifying variants of uncertain significance. R.N.T. Lassiter.

2471T Parental variant study is informative for variant classification in significant number of neurodevelopment genes. E.C. Weltmer.

2472F Neurogenetic disease study in Mali reveals novel mutations and suggests new disease-causing genes. G. Landoure.

2473W Characterization of copy number variations of genomic regions containing long noncoding RNA in children with neurological phenotypes. S.C. Smith.

2474T Analysis of total RNA in the cerebrospinal fluid environment. S.L. Farrugia.

2475F Optimized, modular, target-enrichment gene panels for the detection of genetic variants associated with neurological disorders. A.B. Emerman.

2476W Autism spectrum disorder: A study of series of cases with genetic alterations. J.L. Mussolini.

2477T An inherited distal 16p11.2 deletion demonstrates association with rhizomelic shortening, variable expressivity, and incomplete penetrance for psychiatric illness: A case report. E. Morris.

2478F ‡ Diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. C. Lowther.

2479W Clinical utility of exome sequencing in individuals with large homozygous regions. A. Prasad.

2480T A framework to identify contributing genes in patients with Phelan-McDermid syndrome. A.C. Tabet.

2481F Post-zygotic copy number variations in autism spectrum disorder discordant monozygotic twins. D.L. Nuñez.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2482W Chromosomal microarray analysis of pediatric patients with autism spectrum disorders and intellectual disabilities. A.R. Patel.

2483T ‡ Whole genome sequencing of neurodevelopmental disorders in Japanese. C. Abe-Hatano.

2484F Diagnostic yield of chromosomal microarray analysis in patients with intellectual disability and developmental delay. A.C. Ceylan.

2485W ‡ Unravelling structural chromosomal rearrangements by whole genome sequencing: Results of the ANI project, a French collaborative study including 55 patients with intellectual disability and/or congenital malformations. D. Sanlaville.

2486T Using parental report questionnaires to identify developmental delay in a 22q11.2 deletion specialty clinic. K. Coleman.

2487F Increased yield of clinically relevant candidates in the UK 100,000 Genomes Project using the Fabric Genomics platform. M. Babcock.

2488W Next generation sequencing based on long range PCR: A reliable, expeditious, cost effective genetic testing strategy for lysosomal storage diseases. M.C. Vanaja.

2489T StarSeq, an innovative method based on NGS for accurate detection of punctual mutations and copy number variants in children with intellectual disability and obesity. A. Bonnefond.

2490F Genetic causes of intellectual disability in 102 consanguineous families from Jordan. T. Froukh.

2491W Identification of copy number variations from whole-exome sequencing using eXome Hidden Markov Model (XHMM): A French experience. E. Tisserant.

2492T MIDAS Project status report: Trio whole exome sequencing in patients with intellectual disability. Y. Dinçer.

2493F Copy number reanalysis: The hidden contribution of *MED13L* to intellectual disability. L.K. Conlin.

2494W Trio whole genome sequencing for undiagnosed patients with moderate or severe intellectual disability. B. Cogné.

2495T ‡ Reanalysis of whole exome and genome data leads to new diagnoses in children with intellectual disability and developmental delay. C.R. Finnila.

2496F The clinical application of chromosomal microarray in in the diagnosis of children with developmental delay/intellectual disability in Korea: A single tertiary center experience. Y. Kim.

2497W Case study: Identification of a pathogenic microdeletion using exome data. M.N. Luong.

2498T Genetic evaluation of patients with intellectual disability (ID) using chromosomal microarray and next-generation sequencing at the "ID clinic". K. Takano.

2499F Targeted next-generation sequencing of 75 genes in Japanese patients with intellectual disability and multiple congenital anomalies of unknown etiology. D.T. Uehara.

2500W ‡ Diagnostic testing using capture-based NGS reveals a high rate of mosaicism in genes associated with neurodevelopmental disorders. D. McKnight.

2501T Recurrent telomere captures as the mechanism producing uniquely complex 1p UPD mosaicism. P.L. Pearson.

2502F Genetic spectrum of limb-girdle muscular dystrophy in Taiwan. Y.L. Lin.

2503W A novel intronic mutation in MTM1 detected by RNA analysis in a case of X-linked myotubular myopathy. A.H. AlHashim.

2504T Development of a unified *DMPK* and *CNBP* PCR workflow for determining repeat expansions relevant to myotonic dystrophies. J. Wisotsky.

2505F Clinical correlations of a streamlined molecular assay based on AmplideX® PCR/CE technology that determines repeat size for both normal and expanded alleles in *DMPK* for myotonic dystrophy 1. B. Hall.

2506W NGS-based diagnostics at Newborn Screening Ontario. L. Racacho.

2507T Wide range of maternal heteroplasmy for inherited pathogenic mtATP6 variants. J. Thompson.

2508F Development of a strategy for the genetic diagnosis of mitochondrial diseases in paediatric population. G. Barcia.

2509W Complexities of mitochondrial gene testing. C. Kaiwar.

2510T Mitochondrial genome sequencing in phenotype-based panels and exome sequencing increases test sensitivity. M.A. Reott.

2511F ‡ Expert specification of the ACMG/AMP variant interpretation guidelines and application of the ClinGen gene curation framework to genetic hearing loss. A. Abou Tayoun.

2512W ‡ A comprehensive resource and guideline for the development and validation of exome-based panels for clinical laboratories. R. Niazi.

2513T Exome sequencing leads to the identification of two novel MYO15A mutations in a German family with autosomal recessive non-syndromic hearing loss. R. Birkenhager.

2514F Genome and exome sequencing application in clinical diagnostics for deafness: Not quite there yet. H. Azaiez.

2515W Targeted exome sequencing as a molecular diagnostic tool for syndromic hearing loss. Y.S. Lima.

2516T Kagami Ogata Syndrome caused by a 14q32 microdeletion that did not encompass MEG3 DMR. W.T. Keng.

2517F *De novo* variant in *SOS2* with a concurrent *SHOX* deletion: Report of a patient with short stature, dysmorphic features and heart defect. D. Lyalin.

2518W Defects in cohesin components STAG1 and STAG2 expand the locus heterogeneity of "cohesinopathies". B. Yuan.

2519T ‡ Constitutive supernumerary marker chromosomes are the chromothripsis remnant of the supernumerary chromosome present in trisomic embryos. N. Kurtas.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

Photographing and recording posters are strictly prohibited if the author has displayed the no photography symbol. You agreed to adhere to this policy when you registered.

2520F Association of 22q11.2 duplication with two variants in *FREM2* gene in a patient with an unusual phenotype. M.I. Melaragno.

2521W Case report of an unusual situation in which mother and daughter have two different chromosomal abnormalities. J. Neri.

2522T Breakpoint mapping in five Brazilian cases of distal 5p deletion: Influence of copy number variable regions and haploinsufficiency to clinical phenotype. S.N. Chehimi.

2523F Mosaicism and chimerism: Mechanistic answers for chromosomally distinct cell populations. R. Rowsey.

2524W Comparison of diagnostic yield between clinical exome sequencing and whole exome sequencing. Y. Niu.

2525T Familial interstitial deletion 1(q43q44) due to maternal complex balanced insertional translocation (IT) and inversion in 3p. Y. Hadid.

2526F Investigating complex structural variants using third generation genome sequencing and mapping technologies. R. Rajagopalan.

2527W Bohring-Optiz syndrome caused by an ASXL1 mutation inherited from a germline mosaic mother. D. Copenheaver.

2528T Concurrent exome and copy number variation (CNV) analyses enable more precise diagnoses and shorten diagnostic testing time in patients with pediatric disorders. A.V. Dharmadhikari.

2529F Diagnostic exome sequencing (DES) coupled with rules-based candidate gene analysis identifies a causative *RRAS* lesion in a patient with a novel RASopathy. C.B. Mroske.

2530W *Clinical evaluation with Holm´s criteria for Prader-Willi syndrome in a cohort of 15 Mexican pediatric patients.* R. Lara-Enríquez.

2531T Diversity of *JAG1* mutations in Japanese patients with Alagille syndrome. T. Togawa.

2532F Profile and characterization of genetic variants identified in 208 patients with Kabuki syndrome assists in pathogenicity determination. C.F. Li.

2533W Molecular diagnosis of tuberous sclerosis complex by next generation sequencing in pediatric patients from Mexico. M.E. Reyna-Fabián.

2534T ‡ Application of next generation sequencing in NICU experiences from a 1239-patient pilot study. W. Zhou.

2535F Rapid Paediatric Sequencing (RaPS): Implementation of a framework for rapid genetic diagnosis in critically ill children using whole-genome sequencing. H. Williams.

2536W Complete STK11 deletion and atypical symptoms in Peutz-Jeghers Syndrome. Y.H. Hong.

2537T Whole *ROR2* gene deletion uncovering a pathogenic mutation in a patient with autosomal recessive Robinow syndrome. B.M. Ferreira.

2538F Expanding the spectrum of *TBL1XR* deletion: Report of a patient with brain and cardiac malformations. S. Oliveira.

2539W Study of genetic defects in patients with limb malformations. A. Rai.

2540T ‡ AMELIE accelerates Mendelian patient diagnosis directly from the primary literature. J. Birgmeier.

2541F Two children with copy number variants in the critical regions of both Wolf-Hirschhorn and Cri du Chat syndrome. L. Andoni.

2542W Recurrent unbalanced constitutional chromosomal translocation between chromosomes 8 and 12, der(8)t(8;12)(p23.1;p13.31), detected in three patients with similar phenotype. D. Huang.

2543T The first Japanese patient of Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL) diagnosed via *POLD1* mutation detection. I. Imoto.

2544F Enhanced detection of uniparental disomy (UPD) and copy number variations (CNVs) with delineation of parental origin for clinical trio exome cases. H. Dai.

2545W A rapid and reliable chromosome analysis method for products of conception using interphase nuclei. R. Babu.

2546T Clinical validation of whole-genome sequencing assay for constitutional disorders. A. Abhyankar.

2547F Analysis of 1374 comparative genomic hybridization (CGH) results: Indirect evaluation of carrier rate in a Colombian population. M. Garcia-Acero.

2548W 10x Genomics® Chromium™linked-read workflows fully optimized on PerkinElmer Sciclone® for high-throughput automation of exome and genome applications. J. Garifallou.

2549T Human ring chromosome atlas: A web-based registry and a comprehensive review of ring chromosome cases in the Chinese population. Q. Hu.

2550F Uniparental isodisomy X: Evidence for monosomy rescue. K. Rudd.

2551W Variant of Turner syndrome 45, X/46Xdel(X)(q21) mosaicism: A case report. G. Giraldo.

2552T A unique case of male/female chimerism in buccal specimen due to a bone marrow transplant. H. Risheg.

2553F Landscape of disorders of sex development with mosaicism. A. Rojas.

2554W HLA-B*1502 genotyping for the prevention of carbamazepine induced severe cutaneous adverse drug reactions (SCARs) in a children's hospital. H. Law.

2555T Clinical whole genome sequencing in a pediatric hospital. C. Saunders.

2556F Single exon resolution copy number analysis significantly increases clinical sensitivity of NGS. R.R. Kelly.

2557W A comparative study of the CYTAG® CGH and CYTAG® SuperCGH DNA labeling kits to detect CNVs with small amounts of DNA. A.L. Mosca-Boidron.

2558T Gene-specific criteria for PTEN variant curation. J. Mester.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2559F ‡ Covering all the bases: Case vignettes and diagnostic pipelines at the Stanford Center for Undiagnosed Diseases. A.M. Dries.

2560W VarSome, the Human Genomic Variant Search Engine. A. Massouras.

2561T Going beyond the ACMG recommendations for reporting secondary findings: From decision-making to follow-up. N.T. Strande.

2562F The discussion of whether in vitro fertilization (IVF) or preimplantation genetic diagnosis (PGD) in inv(9) carriers. H. Chen.

2563W Improved molecular tracking of individual genomes for clinical whole-genome sequencing. S. Batalov.

2564T Workshop in genomic medicine for paediatric specialists. A.D. Gilbert.

2565F Developing frameworks to evaluate diagnostic genomic testing strategies for rare disease and cancer. R. Scott.

2566W Benchmarking the quality of diagnostic next generation sequencing. S. Deans.

2567T ‡ Correctly building, evaluating and using clinical grade pathogenicity classifiers for variant of unknown significance. G. Bejerano.

2568F Paternal inheritance of X chromosome CNVs aid in the interpretation of pathogenicity. R. Burnside.

2569W Considering other mechanisms of gene regulation in disorders of sex development. M. Molina.

2570T Identifying single fetal trophoblastic cells in the maternal circulation: A modified NGS genotyping method. X. Zhuo.

2571F Comprehensive analysis of CYP2D6 variants and copy numbers using reverse-hybridization and real-time PCR based assays. C. Oberkanins.

2572W A highly specific, cost-effective solution utilizing a unique 2-enzyme system for SNP genotyping in pharmacogenetic studies. D. Tsang.

2573T Long read capture sequencing for clinical applications. K.C. Worley.

2574F Clinical reassessment of post-laboratory variant call format (VCF) files. L.F. Al Subaie.

2575W A single assay system for CNV, AOH, and Seq Var genetic testing. S. Shams.

2576T ‡ Copy number variant discrepancy resolution using the ClinGen dosage sensitivity map. E.R. Riggs.

2577F Defining quality standards for clinical whole exome sequencing: A national collaborative study of the Dutch Society for Clinical Genetic Laboratory Diagnostics (VKGL). M.G. Elferink.

2578W Characterization of incidental findings identified by targeted testing for gene deletions and duplications. A.M. Janze.

2579T Diagnostic digital PCR copy number assay for NKX2-1 related disorders. K.M. Robbins.

2580F An open-source quality control monitoring system for clinical NGS. N.R. Tawari.

2581W Application of comprehensive actionable medical panel and whole exome sequencing in critical care of pediatric patients. H. Wang.

2582T Structural variant detection with optical mapping and microfluidic partitioning: A t(9;13) case report. D. Baldridge.

2583F Confirmation of copy number variations from massively parallel sequencing using a chromosomal microarray with single exon level detection. J.M. Devaney.

2584W ‡ New systematic rubric for clinical interpretation of copy number variants (CNVs) improves interpretation consistency across laboratories. D. Pineda Alvarez.

2585T No consistent phenotype in patients with copy number variants of the SHOX downstream regulatory domain. A. Wray.

2586F ‡ Clinical variant reclassification and scaling support for the return of updated genetic results. H.L. Rehm.

2587W Linked-read sequencing for molecular cytogenetics. S. Garcia.

2588T High throughput linked-read sequencing for improved variant detection. A. Fehr.

2589F A clinical molecular genetics laboratory experience with whole exome sequencing. J. Machado.

2590W Diagnostic exome sequencing identifies a homozygous whole-gene deletion of *DPY19L2* that was not detected by a high-density single nucleotide polymorphism (SNP) array. S. Sajan.

2591T ‡ Towards automated variant pathogenicity assessment: A Bayesian classification framework. L.G. Biesecker.

2592F Protocols to keep NGS gene panels and annotation content current. N.A. Rouse.

2593W Elective whole genome testing in clinical practice. D. Bick.

2594T Clinically significant small indels detected by whole genome sequencing: A proof of concept case series. C.M. Brown.

2595F Safely washing and reusing pipette tips for Next Generation Sequencing (NGS) qPCR. A. Graham.

2596W A randomized controlled trial of rapid whole genome sequencing for neonatal genetic diagnosis. S. Kingsmore.

2597T Challenges and solutions for FFPE DNA quantitation. K. Plasman.

2598F CLIP-Cap: Combined Long-Insert Paired-End and Capture sequencing, a novel method for the analysis of complex genomic aberrations. C. Purmann.

2599W Frequencies of *BCHE* variants in a large cohort of US individuals. G. Zhu.

2600T WGS is an imperfect but valuable tool for predicting the risk of genetic disease in children. M.S. Meyn.

2601F Undiagnosed Diseases Network (UDN) successes in precision medicine. J. Phillips.

Cardiovascular Phenotypes

2602W Integration of sequence data from 150,000 individuals provides new insights for variants involved in cardiomyopathy. E.J. Mazaika.

2603T Analysis of 181 selected genes associated with dilated cardiomyopathy by next-generation sequencing. K. Al Harbi.

2604F Loss of ADAMTS3 activity causes Hennekam lymphangiectasia-lymphedema syndrome 3. P. Brouillard.

2605W Titin rare genetic variants in arrhythmogenic cardiomyopathy. R. Celeghin.

2606T Novel heterozygous mutations of *KCNQ1* gene in a Jervell and Lange-Nielsen syndrome patient with gastric neuroendocrine tumor. K. Choi.

2607F MCTP2 gene change detected by whole exome sequencing in an infant with endocardial fibroelastosis syndrome who underwent heart transplantation. D. Ercelen.

2608W Use of the ClinGen clinical validity framework to evaluate the strength of evidence for genes implicated in hypertrophic cardiomyopathy. J. Goldstein.

2609T Cardiologic findings in 101 patients with Williams Syndrome. R.S. Honjo.

2610F Toward genetics-driven early intervention in dilated cardiomyopathy: The DCM Precision Medicine Study. D.D. Kinnamon.

2611W Familial TAPVR with 15q11.2 (BP1-BP2) microdeletion. Y. Kuroda.

2612T Genetic testing in arrhythmogenic cardiomyopathy: New insights into the disease determinants and new doubts. E. Lazzarini.

2613F A report of a patient with hypertrophic cardiomyopathy without myopathy associated with FLNC heterozygous pathogenic variant and review of literature. B. Monteleone.

2614W Comparison of genetic architecture of isolated left ventricular noncompaction cardiomyopathy and familial dilated cardiomyopathy as assessed by whole exome sequencing. L. Piherová.

2615T ‡ High incidence of *SCN5A* c.5350 G>A (p.Glu1784Lys) heterozygotes and prolonged QT intervals in a large biobank cohort. M.L.B. Schwartz.

2616F Germline loss-of-function mutations in *EPHB4* cause a second form of capillary malformation–arteriovenous malformation (CM-AVM2) deregulating RAS-MAPK signaling. M. Vikkula.

2617W Predisposition genetic screening for actionable cardiovascular conditions in patients undergoing heritable cancer syndrome testing: Prevalence of pathogenic variants in 10,812 individuals. S. Yang.

2618T Novel *CACNA1C* variant associated with Long QT syndrome in a multigenerational family. V. Novelli.

2619F Mutation spectrum of Long QT Syndrome in Singapore. R.Y.Y. Yong.

2620W Clinical evolution and recommendations for management of the smooth muscle dysfunction syndrome due to mutations of the ACTA2 arginine 179. E. Regalado.

2621T HIVEP2 as a cause hypoplastic left heart syndrome: A report of a rare condition in an 18 month old boy. H. Holway.

2622F *EIF2AK4* mutations are not likely a common genetic modifier of disease in BMPR2 mutation positive pulmonary arterial hypertension patients. K. Sumner.

2623W Loss-of-function variant in *FNDC3B* is associated with dominant pulmonary arterial hypertension in a pedigree. M. Cousin.

2624T Mutation spectrum of the *KCNQ1*, *KCNH2*, and *SCN5A* genes for the long QT syndrome in Korea. M. Kim.

2625F Whole-exome sequencing identified a *de novo PDE3A* mutation causing autosomal dominant hypertension with brachydactyly. D. Wang.

2626W A novel workflow for analysis of whole genome sequencing in cardiac disease using tissue-specific biological datasets. S.M. Hosseini.

2627T Novel PIEZO1 mutations in patients with autosomal recessive hereditary lymphedema. L. Lai.

2628F Exome sequencing in children with pulmonary arterial hypertension demonstrates a different genetic architecture of disease compared to adults. C.L. Welch.

2629W CRISPR-Cas9 mediated knockout of SEL1L and proteasomal inhibition reveal divergent degradation pathways for corresponding LDLR and VLDLR disease-causing mutants. B.R. Ali.

2630T Thoracic aortic disease outcomes in 987 cases with pathogenic variants in *ACTA2*, *PRKG1, TGFBR1, TGFBR2* and *SMAD3* ascertained by the Montalcino Aortic Consortium. D. Milewicz.

2631F Exome sequencing of 103 Williams syndrome cases rules out variation in the remaining elastin allele as a major contributor to variance in blood pressure and arterial stenosis. P.C.R. Parrish.

2632W Treatment of RIT1-associated cardiomyopathy with trametinib: Initial results in two patients. G. Andelfinger.

2633T Genetic architecture of pulmonary arterial hypertension with congenital heart disease. N. Zhu.

2634F Probing for modifiers of X-inactivation in a cohort of Amish families with hemophilia B. A. Ozel.

2635W ‡ Novel genetic associations for blood pressure identified via gene-alcohol consumption interaction in about 570K individuals. M.F. Feitosa.

2636T Genome-wide association study of cardiotoxicity and cardiac gene expression in NCCTG N9831 (Alliance) Adjuvant Trastuzumab Trial. D.J. Serie.

2637F The association of TMPO and RYR1 genes with cardiovascular diseases in a Turkish Cypriot Family. M.C. Ergoren.

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2638W Association of a polymorphism in *ITGB3* with resistance to clopidogrel in early acute coronary syndrome in an admixed population from Colombia. A.V. Valencia-Duarte.

2639T ‡ Identifying and characterizing causal genes in GWAS-identified loci for heart rate variability using high-throughput, image-based screens in zebrafish larvae. B. von der Heyde.

2640F ‡ Combining Mendelian genetics and genetic epidemiology identifies *APOL3* as a new gene for abdominal aortic aneurysm. J.M. Albuisson.

2641W Is HDL-C causally associated with risk of cardiovascular diseases in Han Chinese? A Mendelian randomization study with 10,000 subjects. M.I. Biradar.

2642T ‡ Genome-wide association study of congenital heart disease in the UK Biobank. A. Córdova-Palomera.

2643F Genetic correlation of human lipidomic endophenotypes and cardiometabolic phenotypes in the Busselton Family Heart Study. G. Cadby.

2644W Genome-wide association analysis identifies multiple loci associated with coronary artery calcification in Koreans. S. Choi.

2645T The search for coronary heart disease biomarkers: A large scale reanalysis of gene expression data. B. Cunha.

2646F Identification of rare variation influencing CVD risk in Mexican Americans. J.E. Curran.

2647W Pharmacogenetic study on clopidogrel response among Filipinos. E.C. Cutiongco de la Paz.

2648T Arrhythmia and night vision blindness: Chicken and egg? Or could be chicken or egg? A. Faucon.

2649F Associations of circulating protein levels with lipid fractions in the general population. S.M. Figarska.

2650W Association of common variants in arrhythmogenic cardiomyopathy desmosomal genes with ECG traits in the general population. L. Foco.

2651T ‡ Genome wide association study identifies nine novel loci for subclinical atherosclerosis traits and highlights genetic correlation with clinical cardiovascular disease. N. Franceschini.

2652F ‡ 66 novel loci detected in a trans-ethnic genome-wide association study of blood pressure in over 440,000 participants from the Million Veteran Program and UK Biobank. A. Giri.

2653W Prevalence of variants of *FVL*, *PTH*, *PAI-1*, *MTHFR* and *EPCR* among Cardio vascular patients. M. Hosseini moghadam.

2654T Elucidating the molecular causes of severe hypercholesterolemia in Finland. N. Junna.

2655F Gender specific modification of heart failure with preserved ejection fraction risk by mitochondrial haplogroups. R.T. Levinson.

2656W Family study of noncompaction cardiomyopathy shows variability of cardiac phenotype within and between families. D. Majoor-Krakauer.

2657T Novel compound homozygous mutations in genes involved in mitochondrial function associated with sudden death with cardiac fibrosis in infancy. K. Mittal.

2658F Identifying new therapeutical targets for congestive heart failure. A. Moreira.

2659W Polygenic hyperlipidemias and coronary artery disease risk. P. Ripatti.

2660T Evaluating the role of genetic variants on blood cell count variability in the Jackson Heart Study. J.R. Shaw.

2661F Elevated genetic risk for coronary artery disease increases hospitalization burden and mortality. M. Sjögren.

2662W Uncovering the genetic determinants of variation in arterial stiffness through joint location and scale association testing. D. Soave.

2663T Genetics and outcome of noncompaction cardiomyopathy: A Dutch multicenter study. J. van Waning.

2664F Evaluating the burden of pathogenic variants for the inherited arrhythmia syndromes. Y.P. Fu.

2665W The importance of epistatic interactions and fitness costs in congenital heart disease. E.O. Akhirome.

2666T African ancestry genome- and transcriptome-wide association study of blood pressure detects nine novel loci in a large cohort from the Million Veteran Program. J.N. Hellwege.

2667F Exome sequencing identifies multiple variants associated with glycA, a novel biomarker of cardiovascular events. L.C. Kwee.

2668W Genetic determinants in the *LILR* gene family predicting statin intolerance. M.K. Siddiqui.

2669T Mendelian randomization to identify causal risk factors for atrial fibrillation. L. Weng.

2670F Localization and dissection of a major quantitative trait locus influencing vascular endothelial growth factor plasma levels. M. Almeida.

2671W A genome-wide gene by cigarette smoking interaction study on elevated blood pressure. M. Kang.

2672T Large-scale validation of zebrafish larvae as a model system for genetic screens in dyslipidaemia, atherosclerosis and coronary artery disease. M. Bandaru.

2673F Sequence data processing and analysis of 70,000 human genomes in the NHLBI TOPMed sequencing program. T. Blackwell.

2674W Genome-wide association study using whole-genome sequencing recapitulates both rare and common risk alleles for Brugada syndrome. R. Redon.

2675T Genome-wide association study of susceptibility to rheumatic heart disease in South Asians: Preliminary results. K. Auckland.

2676F Updated genome-wide association study and functional annotation reveals new risk loci for mitral valve prolapse. N. Bouatia-Naji.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

2677W Circulating cholesteryl ester transfer protein (CETP) concentration: A genome-wide association study followed by Mendelian randomization on coronary artery disease. D. Mook-Kanamori.

2678T A genome-wide association study identifies novel genetic signatures associated with thiazide diuretics adverse metabolic events. M.H. Shahin.

2679F Genome-wide association study of transposition of the great arteries. D. Skoric-Milosavljevic.

2680W GWAS-driven pathway analyses and functional validation reveals *GLIS1* to associate with mitral valve prolapse. M. Yu.

2681T ‡ A novel LDL-lowering missense variant in *B4GALT1* identifies novel biological connection between protein glycosylation and cardiovascular risk factors in human. M. Montasser.

2682F Whole genome cardiac DNA methylation fingerprint and gene expression analysis provide new insights in the pathogenesis of chronic Chagas disease cardiomyopathy. C. Chevillard.

2683W Primary lymphedema: A novel association with 22q11.2 deletion syndrome. M. Unolt.

2684T Utilization of drugs with evidence for pharmacogenomic testing following percutaneous coronary intervention. N. El Rouby.

2685F ‡ Genome-wide association study reveals novel genetic markers associated with chlorthalidone blood pressure response. S. Singh.

2686W Exome-chip meta-analysis identifies association between variation in *ANKRD26* and platelet aggregation. A.D. Johnson.

2687T Functional fine-mapping of coronary artery disease risk variants. B. Liu.

2688F Genetic variation in thromboxane A synthase 1 is associated with stroke risk that can be reduced by daily aspirin. S. Zajic.

2689W A longitudinal transcriptome analysis identifies novel gene expression signatures for body mass index in monocytes. C. Müller.

2690T Implementing genome-based predictive and preventive medicine: The GeneRISK follow-up study. E. Widen.

2691F The effect of genetic variation in donors and patients on rejection after heart transplantation. J. van Setten.

2692W Univariate and phenome-wide GWAS of correlated electrocardiographic traits offer novel insights into genetics of cardiac electrophysiology: the Population Architecture using Genomics and Epidemiology (PAGE) study. A.R. Baldassari.

2693T Putative loss-of-function (pLOF) genetic variants in arrhythmogenic cardiomyopathy-associated genes: Prevalence and EHR-based phenotype in 50,000 biobank participants. C.M. Haggerty.

2694F Breast cancer clinical trial of chemotherapy and trastuzumab: Potential tool to identify cardiac modifying variants of dilated cardiomyopathy. N. Norton.

2695W *APOL1* coding variants are associated with incident cardiovascular disease in community-dwelling African Americans. C.A. Winkler.

2696T The role of Kringle IV 2 copy number variation and SNPs on Lp(a) levels and cardio-metabolic risk. S.E. Ruotsalainen.

2697F Genome-wide trans-ethnic meta-analysis for a novel sleep apnea endophenotype. H. Wang.

2698W Fitness, physical activity, and cardiovascular disease: Longitudinal and genetic analyses in the UK Biobank Study. E. Tikkanen.

2699T Large-scale genomic study of >26,000 MyCode participants uncovers novel loci for hemostasis. J. Backman.

2700F Cross-exposure multivariate interaction tests. J. Kim.

2701W A Bayesian approach for detecting gene by environment interactions with common and rare variants. S.M. Lutz.

2702T StructLMM: Resolving genetic effects due to environmental sample substructure. R. Moore.

2703F Colocalization of gene-psychosocial interactions identifies novel genomic regions for blood pressure among African Americans. M.A. Richard.

2704W Race, sex, and age differences in GxE association: *EBF1* gene-by-stress interaction on central obesity differs among Blacks, Whites and male, female in multiple harmonized datasets. A. Singh.

2705T Multi-ancestry genome-wide study incorporating gene-smoking interactions identifies 139 genome-wide significant loci for systolic and diastolic blood pressure. Y.J. Sung.

2706F GWAS of red cell distribution width identifies discovery associations at GCNT4, KCNJ3, and chr6p22.1 in admixed U.S. populations: The PAGE Study. C.J. Hodonsky.

2707W Cadmium exposure, *MT* polymorphisms and subclinical cardiovascular disease in American Indians: The Strong Heart Family Study. R. Hou.

2708T Genome-wide association study of mitochondrial DNA copy number: The Cohorts for Heart & Aging Research in Genetic Epidemiology (CHARGE). R.J. Longchamps.

2709F "Genotype-phenotype in Marfan syndrome patients with causative mutations in the calcium binding region of the 43 cbEGF-like domains in fibrillin-1 gene (*FBN1*)". J.A. Aragon-Martin.

2710W Family based method for the discovery of rare high penetrance sequence variants. G. Sveinbjornsson.

2711T Association of genetic risk score with childhood obesity-related traits: The Santiago Longitudinal Cohort Study (SLCS). G. Chittoor.

2712F New genetic variants unveiled using a predicted-VAT mass phenotype. T. Karlsson.

2713W Towards precision therapy in hypertension: Genome-wide association study reveals genetic variants associated with uncontrolled blood pressure on thiazide diuretic/beta-blocker combination therapy. O. Magvanjav.

2714T Low frequency and rare variants of *RBFOX1* are associated with blood pressure. K.Y. He.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2715F Incorporating multiple sources of biological knowledge into association analysis of whole genome sequencing data identifies novel trait-associated rare variants. Y. Ma.

2716W Low frequency and rare variants in multiple genes are associated with sleep related traits using whole genome sequencing data. X. Zhu.

2717T Partitioning genome-wide summary statistics improves polygenic risk prediction. S. Chun.

2718F Geographic distribution of polygenic risk of complex traits and diseases in Finland. S. Kerminen.

2719W An information theoretic approach to filtering false signals of pathogenicity across ancestrally diverse populations. A.K. Manrai.

2720T Control of ethnically-stratified vascular risk factors in modeling of intracerebral hemorrhage. S. Marini.

2721F Association of *SELP* variants and soluble P-selectin levels with type 2 diabetes mellitus: A case-control study. R. Kaur.

2722W Whole genome sequence reveals selection for muscle and cardiovascular functions in sport hunting dog breeds. J. Kim.

2723T Heterogeneity in coronary artery disease GWAS results is associated with pan-tissue eQTL count. K.W. Johnson.

2724F Origins and dynamics of the Brazilian population and sickle cell mutations reveal unexpected diversity. Y. Guo.

2725W Integrating biological age and transcriptome markers for predicting the functional recovery potential of patients undergoing mechanical circulatory support surgery. G. Bondar.

2726T Integrated analysis using RNA-Seq and ChIP-Seq data to understand the regulation of cardiogenesis. M. Toufiq.

2727F Disease-specific variant pathogenicity prediction using machine learning methods improves interpretation in inherited cardiac conditions. X. Zhang.

2728W A disease-specific and automated variant annotator enables fast and accurate clinical variant interpretation. N. Whiffin.

2729T Data mining "normal" chromosome microarrays for gene discovery. N. Walton.

2730F Postoperative risk prediction based on preoperative leukocyte immunobiology. S. Ramachandrula.

2731W ‡ Genetic variants in familial abdominal aortic aneurysms identified by whole genome and exome sequencing. A. IJpma.

2732T ‡ High-throughput discovery of deleterious cardiac sodium channel variants. A. Glazer.

2733F ‡ Integration of exome genetic variation into mass spectrometry peptide identification to effectively identify plasma proteome QTLs. T. Solomon.

2734W ‡ Large-scale generation of iPSC-derived cardiomyocytes for functional genomic applications. M.K.R. Donovan.

2735T ‡ The iPSCORE resource: 222 iPSC lines enabling functional characterization of genetic variation across a variety of cell types. E.N. Smith.

2736F Finding biomarkers for thromboembolism leading to stroke. D.B. Dogini.

2737W Danon Disease: A lysosomal hypertrophic cardiomyopathy model created by CRISPR editing LAMP2 in iPSC and fibroblasts. C. McKinney.

2738T Chromosome 22q11 microdeletion: Modifiers of the cardiovascular phenotype identified by whole exome sequencing. G. Repetto.

2739F AIF-1 in association with TLR-2 induces proinflammatory response in monocytes after ischemia reperfusion (IR). D.Olga. McDaniel.

2740W Association of rare recurrent copy number variants in next generation sequencing data from family trios with congenital heart defects. Y. Liu.

2741T The effects of missense mutations causing PRKAG2 cardiomyopathy on expression levels of selected genes involved in AMPK pathway. E. Komurcu-Bayrak.

2742F Investigation of microRNA expression in coronary artery disease. N. Coban.

2743W An integrated genetic-epigenetic prediction model for coronary heart disease. M. Dogan.

2744T Epigenetic modulation in the pathogenesis and treatment of inherited aortic aneurysm conditions. B.E. Kang.

2745F Gender transcriptome signatures for congenital heart defects (CHD) children based on next generation sequencing technologies of cardiac tissues. C. Kim.

2746W Interpreting genetic variation in coronary artery disease (CAD). I. Selvarajan.

2747T Epigenetic regulation of PAR-4-mediated platelet activation: Understanding the mechanistic links between smoking and cardiovascular disease. N. Timpson.

2748F Silencing the miR-30c-5p pathway attenuates cocaine-induced increases in blood pressure and aortic stiffness. W. Zhu.

2749W Epigenome-wide association study of the previous number of strokes in participants from the Vitamin Intervention for Stroke Prevention (VISP) clinical trial. N.M. Davis-Armstrong.

2750T ‡ Enhancer signature of dilated cardiomyopathy. D. Hemerich.

2751F Identification of eQTLs for platelet and hemostasis related genes in platelets and leukocytes within the Framingham Heart Study. J.E. Huffman.

2752W Characterization of experimentally validated heart disease genes using functional genomic information and 3d genome structure. R. Gill.

2753T Functional analysis of *β-globin* locus control region hypersensitive site 2-associated proteins and noncoding RNA. A. Gurumurthy.

2754F RNA-seq of human heart tissue identifies shared and divergent expression signatures of heart failure. M.E. Sweet.

2755W The communal relation of MTHFR, MTR, RFC gene polymorphisms and hyperhomocysteinemia as plausible risk of congenital septal defects. S.B. Sunayana.

2756T Fibulin-4a inhibits vascular and enhances cardiac cell fate by inhibiting transforming growth factor beta signaling. Z. Urban.

2757F Genetic causes of heterotaxy identified by whole exome sequencing. A. Sridhar.

2758W A mutation in the *LMOD1* actin-binding domain segregating with disease in a large British family with thoracic aortic aneurysms and dissections. Y. Wan.

2759T Homozygous *EEF1A2* mutation causes dilated cardiomyopathy, failure to thrive, global developmental delay, epilepsy and early death. P.B. Agrawal.

2760F Isoforms and eQTLs of the myocardial infarction gene *PHACTR1*. V. Codina-Fauteux.

Statistical Genetics and Genetic Epidemiology

2761W Integration of GWAS and local genetic effects on gene expression (eQTL/ASE) highlights genes with kidney function and disease. C. Qiu.

2762T Contribution of AMD risk variants to the genetic architecture of choroidal thickness in the Amish. N. Restrepo.

2763F Type 1 diabetes progression is correlated with changes in the co-expression relationships of immune response genes. I. Braenne.

2764W Understanding progression and subtypes of prediabetes with metabolomics and genomic profiling in Starr County Mexican Americans. G. Jun.

2765T Preliminary evidence suggests that a 6.7 kb deletion polymorphism in *LILRA3* is associated with Type 1 Diabetes. C. Maroteau.

2766F ‡ Smoking-by-genotype interaction in type 2 diabetes. P. Wu.

2767W Genome-by-environment interactions have a major impact on obesity. C. Amador.

2768T Pleiotropic associations of adiposity-related genetic risk scores. Z. Fairhurst-Hunter.

2769F Investigation of the association between ITLN1 gene A326T polymorphism and in subjects with type 2 diabetes mellitus and obese: In the TARF study. F. Geyik.

2770W ‡ Fine-mapping and characterization of GWAS loci harboring extensive allelic heterogeneity. C. Spracklen.

2771T Gender-, genotype- and ethnic-specific effects of sugar-sweetened beverages on serum uric acid concentrations. X. Zhang.

2772F Long-term response to oral eliglustat in treatment-naïve adults with Gaucher disease type 1: Final efficacy and safety results from a phase 2 clinical trial after 8 years of treatment. H. Lau.

2773W Long-term results of ENGAGE: A phase 3, randomized, double-blind, placebo-controlled, multi-center study investigating the efficacy and safety of eliglustat in adults with Gaucher disease type 1. P. Mistry.

2774T Diagnostic of a worldwide cohort of Gaucher patients: Glucosylsphingosine levels in blood reflects the severity of GBA mutations. A. Rolfs.

2775F Obesity is a systemic regulatory outcome and mainly controlled by several tissues. R. Hao.

2776W Genetic polymorphism of APOA5 gene is associated with metabolic syndrome in Koreans. S.W. Oh.

2777T Gene-level differential methylation analysis. H. Xu.

2778F Newborn screening for six lysosomal storage diseases in a cohort of Mexican patients: Three-year findings from a screening program in a closed Mexican health system. J.I. Navarrete.

2779W Obesity revisited: Evidence of genetic predisposition for metabolically healthy obesity. L.O. Huang.

2780T Socioeconomic deprivation amplifies genetic susceptibility to obesity and its comorbidities. K.A. Kentistou.

2781F Effects of the interaction between a *CREBRF* missense variant and body mass index on type 2 diabetes risk in Samoans. E.M. Russell.

2782W Genetic determinants of glycemic response to metformin in the Million Veteran Program. C. Roumie.

2783T Principal component-based prediction of complex traits by using support vector machine approach. X. Li.

2784F ‡ Trans-ethnic meta-analysis of rare variants in sequencing association studies. J. Shi.

2785W A novel approach to analyze the mediation model when the mediator is a censored variable. J. Wang.

2786T Genome-wide association analysis in the UK Household Longitudinal Study offers insights into the genetic architecture of health-related biomarkers. K. Kuchenbaecker.

2787F PLEIOVAR, testing for association between multiple traits and multiple variants. O. Meirelles.

2788W Gene-depressive symptoms interactions identify novel lipid loci in multi-ethnic cohorts. S.K. Musani.

2789T Targeted sequencing of 109 genes in the eMERGEseq panel uncovers novel variants and genes influencing triglyceride levels. X. Fan.

2790F Examining the causal relationship between vitamin D and serum metabolic measures: A Mendelian randomization study. T. Dudding.

2791W Enriched loss-of-function variants associating with lipids in Finns. P. Helkkula.

2792T Do blood lipid levels influence bone mineral density? Findings from a Mendelian randomization study. J. Zheng.

2793F ‡ Adaptive multi-trait association test using GWAS summary data. B. Wu.

2794W Genetic variation associated with telomere length in African American children with and without asthma. M. White.

2795T Genome-wide haplotype-based association study reveals novel non-HLA susceptibility loci for primary biliary cirrhosis in Japanese cohorts. C. Im.

2796F Precisely controlled differential gene expression system to investigate the effect of eQTL. X. Lu.

2797W Transcriptional risk scores link GWAS to eQTL and predict complications in Crohn's disease. U.M. Marigorta.

2798T *HLA-DQ* variants interact with pregnancy to modify risk of multiple sclerosis among women of European ancestry. C. Adams.

2799F ‡ Trans-ethnic GWAS identifies genetic variants associated with white blood cell counts in the Population Architecture using Genomics and Epidemiology (PAGE) Study. K.K. Nishimura.

2800W ‡ Finding genomic variants regulating the exon-skipping. R. Liu.

2801T Variance component selection with microbiome taxonomic data. J. Zhou.

2802F Genome-wide association study identifies susceptibility loci for primary non-response to anti-TNF therapy in patients with inflammatory bowel disease. T. De.

2803W Testing for colocalization of causal variants underlying obstructive sleep apnea and immune-related phenotypes. S. Akle.

2804T Association study of *R3HDM1* variants with aspirin exacerbated respiratory disease and FEV1 decline after aspirin provocation. J. Kim.

2805F ‡ Statistical framework for biological interpretation and improvement of genetic association studies. M. Artomov.

2806W Integrated clinical genome database on hepatitis B-related diseases for genome-wide association: Project goals and utilization of materials and genomic information in the ToMMo biobank. S. Teraguchi.

2807T Trans-ethnic Bayesian meta-analysis detects novel replication evidence for multiple loci for inflammatory bowel disease in African Americans. R.Y. Cordero.

2808F Relationship of genetic and clinical factors and prevalence of CKD in a Japanese population: J-MICC Study. R. Fujii.

2809W A genome- and phenome-wide association study of diverticular disease using electronic health records. Y.J. Joo.

2810T Gene-based pathway analysis for osteoporosis: Insights from genomic-wide association. K.J. Su.

2811F Integrated pediatric bone density phenotypes and genetic regulation of the developing skeleton. J.A. Mitchell.

2812W ‡ Penalized regression for detecting rare variant effects under extreme phenotype sampling for continuous traits. C. Xu.

2813T Alzheimer's Disease Sequencing Project: Case-control analyses of over 10,000 whole exomes. J.C. Bis.

2814F ‡ Tissue-specific genetic regulated expression in late-onset Alzheimer's disease: The Alzheimer's Disease Genetics Consortium (ADGC). H.-H. Chen.

2815W Convergent evidence for *LRP2BP* in resilience to Alzheimer's disease. D. Felsky.

2816T Cell free single stranded DNA concentration in CSF as biomarker to diagnose Alzheimer's disease status. J.D. Gonzalez Murcia.

2817F Genome-wide search for genetic loci perturbing gene co-expression networks in Alzheimer's disease. L. He.

2818W ‡ Genome-wide rare variant imputation and tissue-specific transcriptomic analysis identify novel rare variant candidate loci in late-onset Alzheimer's Disease: The Alzheimer's Disease Genetics Consortium (ADGC). A.C. Naj.

2819T Complex disease prediction: A framework to integrate SNP and imaging data. B. Zhao.

2820F Mitochondrial variants associated with increased risk of late-onset Alzheimer's disease. T.J. Zhou.

2821W Genetic association study on white matter microstructure by integrating multiple neuroimaging datasets. J. Zhang.

2822T Investigating the underlying genetic basis of the co-occurence of epilepsy and psychiatric disorders. H.O. Heyne.

2823F Small posterior fossa in Chiari malformation affected families is significantly linked to 1q43-44 and 12q23. A. Musolf.

2824W Genetics of age-related cognitive decline and relationship to Alzheimer's and other neurodegenerative diseases. M.P. Reeve.

2825T Gene-gene interaction tests for genetic-imaging data analysis. W. Peng.

2826F Low-rank structure based brain connectivity GWAS study. Z. Zhu.

2827W Genetic causes of death in US infants: Findings from the National Center for Health Statistics. C. Lally.

2828T Rare coding mutations in Alzheimer Disease. D. Patel.

2829F ‡ Comparison of methods for multivariate gene-based association analysis using common variants for complex disease. J. Chung.

2830W Relationship between essential tremor and Parkinson's Disease. A.A. Gosch.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

2831T ‡ Multivariate genome-wide association study for volumes of structural MRI regions of interest measures via a genetic correlation network modular analysis. J. Liang.

2832F *SORBS2* is associated with extended Alzheimer disease related phenotypes in *PSEN1* mutation carriers in Puerto Rico. R. Cheng.

2833W Polygenic risk scores applied to UK Biobank data highlight the interplay between behaviour and psychiatric disorders. P.F. O'Reilly.

2834T Severity modifiers in autism spectrum disorder: WGS perspective. S.P. Smieszek.

2835F Diagnostic changes leading to ASDs' prevalence increase altered the disorders' average genetic architecture. E.M. Wigdor.

2836W CNVs among Japanese individuals with neuropsychiatric diseases effect dosage sensitivity in ohnologs and genes expressions. M. Yamasaki.

2837T Testing the moderation of quantitative gene by environment interactions in unrelated but dependent individuals. R. Tahmasbi.

2838F Improved prediction of genetic predisposition to psychiatric disorders using genomic feature best linear unbiased prediction models. P.D. Rohde.

2839W Quantifying the effect of copy-number variants on general intelligence in unselected populations. G. Huguet.

2840T Genome-wide association study of dental treatment-related fear and anxiety nominates novel genes. J.R. Shaffer.

2841F A penalized parametric bootstrap approach for self-contained pathway analysis of gene-environment interaction. B.J. Coombes.

2842W Identification of novel genetic variants of DSM-5 alcohol use disorder: Genome-wide association study in NESARC-III. J. Jung.

2843T Allelic heterogeneity across psychotic disorders and related phenotypes. T. Polushina.

2844F Meta-analysis of *de novo* variants from 9246 probands finds that genes previously associated with autism spectrum disorder harbor more *de novo* variants in probands with intellectual disability/developmental delay without autism. J.A. Kosmicki.

2845W Smoking and neuroticism: Using Mendelian randomization to investigate causality. H. Sallis.

2846T Genetic validation of bipolar disorder identified by automated phenotyping using electronic health records. C. Chen.

2847F Detecting tissue-specific genetic correlation between complex psychologic disorders using GWAS summaries. Q. Fan.

2848W Genetically predicted gene expression in the brain and peripheral tissues associated with PTSD. L.M. Huckins.

2849T Whole genome sequencing in families with bipolar 1 disorder implicates cysteine transport process and synaptic neurotransmission pathway. A. Parrado.

2850F Gene interaction between DRD4 and DAT1 Loci is a ADHD-risk factor in females of Chilean ancestry. G. Pathak.

2851W Proper joint and conditional meta-analysis of sequence data in the presence of missing summary association statistics. D. Liu.

2852T Joint analysis of rare and common variants with the adaptive combination of Bayes factors method. W. Lin.

2853F ‡ Flipping GWAS on its head: A statistical approach to identify genetically distinct disease subphenotypes. A. Dahl.

2854W FOLD: A method to optimize power in meta-analysis of genetic association studies with overlapping subjects. E. Kim.

2855T ‡ Integrating eQTL data with GWAS summary statistics identifies novel genes and pathways associated with schizophrenia. C. Wu.

2856F Identifying highly damaging missense mutations in over 10,000 developmental disorder trios using a regional missense constraint metric. K.E. Samocha.

2857W Genotype-phenotype study of *OPHN1* and *IL1RAPL2* genes mutations in children with intellectual disability. Y.M. Khimsuriya.

2858T Global developmental delay: Genetic causes in a group of Mexican patients. M.L. Arenas-Sordo.

2859F The investigation by WES of inborn errors of metabolism as an underlying cause of idiopathic intellectual disability and/or unspecific congenital malformations in a series of 550 patients. N. Houcinat.

2860W Significant association at the Duffy blood group locus with mitochondrial copy number. X. Geng.

2861T Targeted sequence analysis of human mitochondrial DNA using an IDT xGen® Lockdown® probe panel. R. Lopez.

2862F Association study for common and rare genetic variation contributing to exfoliation syndrome. R.P. Igo.

2863W Whole exome sequencing reveals candidate variants for elevated intraocular pressure in the Beaver Dam Eye Study. W. Li.

2864T Individualized glaucoma risk evaluation using the genomic profile. X. Gao.

2865F Family based association tests of myopia reveal a potentially hidden association signal upstream of two *GABA* receptor genes. C.D. Middlebrooks.

2866W Examination of a rare risk variant in complement factor H for age-related macular degeneration in the Amish. A.R. Waksmunski.

2867T Genetic risk score is associated with vertical cup-disc ratio and improves prediction of primary open angle glaucoma in Latinos. D.R. Nannini.

2868F Grouped association analysis for very rare variants using Fisher's Exact Test and external controls. A. Kwong.

2869W The genetic architecture of the AVSD risk in Down syndrome: Results from chromosome 21 genome sequencing. X. Blanc.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2870T Tracing the dark matter: Prevalence of copy number variants across Mendelian disorders. R. Truty.

2871F Iterating from discovery to epidemiological consequence through disease mechanism. J. Brown.

2872W Genome-wide association analyses in large-scale multi-ancestry cohorts: Statistical challenges and opportunities. C. DeBoever.

2873T Genetic factors that modulate the relationship between education and Alzheimer's disease. R.A. Bhatta.

2874F Vitamin D deficiency: Analysis shows season and dietary vitamin D intake influence the effect of *GC*, *CYP2R1*, *DHCR7* and *CYP24A1* genes on vitamin D levels. K.E. Hatchell.

2875W Evidence of *ZKSCANS, SULT2A1, TRIM4* and *BCL2L11* for serum dehydroepiandrosterone sulfate (DHEAS) levels: Replication from the Long Life Family Study (LLFS). P. An.

2876T Gene by environment interaction in human longevity as observed in Danish birth cohorts from 1905 to 1915. Q. Tan.

2877F Genetics of the human microbiome and implications in obesity associated measures. C.T. Finnicum.

2878W Leukocyte Telomere Length (LTL) as a marker of biological aging in Iranian healthy adult population: Report on assay establishment and recent finding. F. Larti.

2879T Old before our time: Biological ageing in an ethnically diverse cohort of preschool children. K.N. Ly.

2880F Admixture mapping of asthma in individuals of mixed African ancestry reveals a novel association on chromosome 6q23.2. M. Daya.

2881W Methods to estimate heritability of complex traits under a variety of complex genetic architectures. L. Evans.

2882T Robust inference of population structure from next-generation sequencing data with systematic differences in sequencing. Y.J. Hu.

2883F Model-based multiple variants test considering causal status. J. Joo.

2884W Caring without sharing: Genome-wide association and mapping on cohorts fragmented across institutional silos. A. Pourshafeie.

2885T Some novel methods of detecting gene-drug interactions. M. Rao.

2886F Use low-depth and high-depth whole genome sequencing data to predict 36 blood groups. Y. Sun.

2887W Modeling the interactions between coding and non-coding RNA by kernel machines in binary phenotypes. S. Yang.

2888T High frequency of the MEFV c.1437C>G, p.F479L allele among Druze FMF patients. V. Adir.

2889F Family-based rare variant association study of familial myopia in Caucasian families. D. Lewis.

2890W Exploring the effect of minor allele frequency on the inflation of type I error rates for GWA studies of family data with non-normally distributed traits. J.A. Sabourin.

2891T To ERV is human: A phenotype-wide scan linking polymorphic human endogenous retrovirus-K insertions to tissue-specific gene expression and complex diseases. A.D. Wallace.

2892F Robust, accurate, and efficient pedigree reconstruction and pedigree-aware distant relatedness detection in 120 rhesus macaques (*Macaca mulatta*) from the Tulane National Primate Research Center using dense whole genome sequence data. L.E. Petty.

2893W Genome-wide scan of pulmonary phenotypes on local ancestry in African Americans reveals novel genes interacting with smoking. A. Ziyatdinov.

2894T Genetic analyses for antiepileptic drug-induced cutaneous adverse reaction in a HK population. J. Ding.

2895F Bayesian hierarchical modeling of genic sub-region intolerance. T.J. Hayeck.

2896W Mixed-model adjustments for tests of epistasis reduce confounding by other loci. N. Patel.

2897T The usage of local ancestry to Inform eQTL mapping in African Americans. Y. Zhong.

2898F Evidence for a major gene for myopia risk in Han Chinese-American families at 10q26. J.E. Bailey-Wilson.

2899W Platelet-derived growth factor genes, maternal binge drinking and obstructive heart defects. M.A. Cleves.

2900T ‡ Iranome: A human genome variation database of eight major ethnic groups that live in Iran and neighboring countries in the Middle East. M.R. Akbari.

2901F Genome-to-genome analysis of host-pathogen interactions in human tuberculosis. N. Chaturvedi.

2902W Genotype imputation performance using an African-American population. L. Franco.

2903T Data-driven genetic encoding (DAGE) allows flexible identification of novel main effects and SNP-SNP interactions. M.A. Hall.

2904F The genetic architecture of 25-hydroxyvitamin D. X. Jiang.

2905W ‡ Improved genotype imputation in disease-relevant regions with inclusion of patient sequence data: Lessons from cystic fibrosis. N. Panjwani.

2906T Assessing pleiotropy and mediation in loci associated with chronic obstructive pulmonary disease. M.M. Parker.

2907F ‡ Genetic determinants of urinary biomarkers in the UK Biobank. D. Zanetti.

2908W Whole genome sequencing association analysis of red blood cell traits in a multi-ethnic population from the Trans-Omics for Precision Medicine (TOPMed) Project. X. Zheng.

2909T Large-scale inference in population cohorts. M.A. Rivas.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

2910F Population pharmacokinetics of sulindac and genetic polymorphisms of *FMO3* and *AOX1* in women with preterm labor. J. Yee.

2911W Genome wide meta-analysis for dental caries in childhood. S. Haworth.

2912T ‡ Leveraging whole genome sequence data to improve imputation and increase power in GWAS of diverse populations. C. Quick.

2913F GWAS data analysis with non-local prior based Bayesian iterative variable selection-regression. N. Sanyal.

2914W Genome-wide association study of HIV-1 subtype C in Botswana population. A.K. Shevchenko.

2915T Genome-wide association study identifies novel susceptibility loci for tanning ability in Japanese population: From ToMMo cohort study. K. Shido.

2916F Comparison of power of summary based methods for identifying expression-trait associations. Y. Veturi.

2917W Improving imputation by maximizing power. Y. Wu.

2918T Genome-wide analysis of age-related macular degeneration progression. Q. Yan.

2919F Logolas: A tool for visualizing enrichment of genetic signature profiles. K. Dey.

2920W A semi-supervised method for predicting functional consequences of genome-wide coding and noncoding variants. Z. He.

2921T Genome-to-genome analysis: Correcting for population stratification in joint association analysis of host and pathogen genomes (G2G) reduces false positive and negative results. O. Naret.

2922F Mapping genetic organization and disease liabilities of human cortical surface with summary statistics of vertexwise genome-wide association studies. C. Fan.

2923W Gene-based pleiotropic analysis of multiple survival traits via functional regressions with applications to eye diseases. R. Fan.

2924T Exploring genetic associations using self-reported phenotypes in genes for good. A. Pandit.

2925F The 1M Africa genotype array: A powerful tool for medical genetic research globally. T. Carstensen.

2926W Clonal hematopoiesis: Genetic and phenotypic associations. C. Tian.

2927T Summary statistic GWAS joint analyses across 50+ traits. H. Aschard.

2928F Polymorphisms in the *HSF2, LRRC6, MEIG1* and *PTIP* genes correlate with sperm motility. S. Rajender.

2929W Transformation of summary statistics from linear mixed model association on all-or-none traits to odds ratio. L.R. Lloyd-Jones.

2930T Assessing the causal impact of smoking and drinking on human health outcomes: Using 239 novel genetic associations for smoking and drinking addictions and a robust ensemble method for causal inference. F. Chen.

2931F Simulating autosomal genotypes with realistic linkage disequilibrium and a spiked in genetic effect. M. Shi.

2932W X wide association analysis identifies a novel *FRMPD4* locus associated with the differential sex risk for multiple sclerosis. Y. Zhou.

2933T Genome-wide association study identifies novel genetic loci in the Major Histocompatibility Complex (MHC) associated with reduction in *Clostridium difficil*e Infection (CDI) recurrence in patients treated with bezlotoxumab. J. Shen.

2934F ‡ A *CREBRF* missense mutation substantially affects height in Samoans. S.L. Rosenthal.

2935W Investigation of post-colonial demographic structure and the implications for association analyses. K.A. Rand.

2936T Comparison of PC-based and LME-based population structure adjustment using GWAS and WES markers. Y. Chen.

2937F Pharmacological insights from genetic mapping of the plasma proteome. J.C. Maranville.

2938W Measuring the rate and heritability of aging using machine learning methods. J. Ding.

2939T Potentially causal rare variants identified using whole genome sequencing of distant relatives from multiplex families with oral clefts. F. Begum.

2940F Gene-based association testing of dichotomous traits with generalized linear mixed models using extended pedigrees. C. Chiu.

2941W ‡ TRUFFLE: Tests of undetermined relationships between founders - fast, light and efficient. A. Dimitromanolakis.

2942T A fast algorithm for Bayesian multi-locus model in genome-wide association studies. W. Duan.

2943F The SUPERBABY PROJECT: Genetic determinants of the favorable NICU course in premature newborns. K.M. Gnona.

2944W Integration statistics suggest gene expression in the exocrine pancreas may contribute to intestinal obstruction in cystic fibrosis. J. Gong.

2945T Bayesian methods for genetic associations and causal inference yield potential biological insight for genetics of gene regulation. B. Jo.

2946F Human knockouts in the Ashkenazi Jewish population. A. Kleinman.

2947W A genetic variants simulation program to simulate high order epistatic interactions for family-based studies. Q. Li.

2948T Modeling functional enrichment improves polygenic prediction accuracy in UK Biobank and 23andMe data sets. C. Marquez-Luna.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.

2949F Total serum IgE whole genome sequence association analysis in families from Barbados. A. Shetty.

2950W Sum ranking, simple but powerful method for detecting pleiotropic loci. G.V. Roshchupkin.

2951T ‡ Heritability enrichment of specifically expressed genes identifies disease-relevant tissues and cell types. H.K. Finucane.

2952F Multivariate generalized linear model for genetic pleiotropy. D.J. Schaid.

2953W Quantification of MAF-dependent architectures in 14 UK Biobank traits reveals strength of genome-wide negative selection. A. Schoech.

2954T ‡ Local genetic correlation gives insights into the shared genetic architecture of complex traits. H. Shi.

2955F MEGA analysis of alcohol consumption in diverse populations: The Population Architecture using Genomics and Epidemiology (PAGE) Study. K.L. Young.

2956W Association detection between ordinal trait and rare variants based on adaptive combination of p-values. Y. Zhou.

2957T A large-scale genome-wide enrichment analysis identifies new trait-associated genes, pathways and tissues across 31 human phenotypes. X. Zhu.

2958F VikNGS: A C++ Variant Integration Kit for Next Generation Sequencing across research studies for robust rare and common variant association analysis. Z. Baskurt.

2959W GLMM-seq: Gene-based detection of allele-specific expression by RNA sequencing. J. Fan.

2960T Reverse regression enables disease only case-control association studies for burden tests. J. Tom.

2961F Estimating cell-type-specific DNA methylation effects in the presence of cellular heterogeneity. Y. Feng.

2962W Generalizing genetic risk scores from Europeans to Hispanics/Latinos. T. Sofer.

2963T Imputation of exome array variants to the Haplotype Reference Consortium (HRC). S. Bomotti.

2964F Fast permutation tests and related methods for association between rare variants and binary outcomes. A. Sondhi.

2965W Epistasis detection for human complex diseases in structured populations. K. Van Steen.

2966T Leveraging polygenic functional enrichment to improve GWAS power. G. Kichaev.

2967F Development of an evidence based sequence variant interpretation tool based upon ACMG and AMP variant interpretation consensus guidelines. F. Suer.

2968W POLARIS: Polygenic LD-Adjusted Risk Score approach for analysis of GWAS data. E. Baker.

2969T Sequential fine-mapping from summary statistics in meta-analyses of genome-wide association studies. C. Benner.

2970F A meta-analysis strategy based on the SPA test to combine multiple PheWAS studies. R. Dey.

2971W Admixture mapping: Controlling for multiple testing and spurious associations in the presence of population structure. K. Grinde.

2972T Incorporating multiple functional annotations to infer trait-relevant tissues in genome-wide association studies. X. Hao.

2973F Increasing the power of meta-analysis of genome-wide association studies to detect heterogeneous effects. C.H. Lee.

2974W ‡ Integrative analysis of GWAS summary statistics and imputed gene expression in 44 tissues deciphers genetic architecture for many complex traits. M. Li.

2975T A hierarchical clustering method for joint analysis of multiple phenotypes. X. Liang.

2976F Testing for goodness rather than lack of fit of a X-Chromosomal SNP to the Hardy-Weinberg Model. S. Wellek.

2977W Robust genetic prediction of complex traits with latent Dirichlet process regression models. X. Zhou.

2978T Estimating higher-order heritability components in GWAS data from 133,515 individuals. S.R. McCurdy.

2979F ‡ Using relationships inferred from electronic health records to conduct genetic studies. F. Polubriaginof.

2980W Detecting heritable phenotypes without a model: Fast permutation testing for heritability and set-tests. R. Schweiger.

2981T Estimating effect-size distributions using summary level statistics from genomewide association studies and projecting trajectories of future discoveries for 32 complex traits. Y. Zhang.

2982F Combining sequence data from multiple studies: Impact of analysis strategies on rare variant association results. Z. Chen.

2983W ‡ Pervasive pleiotropy in the human genome revealed by a novel quantitative analysis of summary association statistics. D.M. Jordan.

2984T GWAS genes whose expression is implicated by Mendelian randomization are highly connected in tissue-specific regulatory circuits. E. Porcu.

2985F Widespread pleiotropy confounds causal relationships between complex traits and diseases inferred from Mendelian randomization. M. Verbanck.

2986W Allele specific information in Mendelian randomization. X. Wang.

2987T A highly adaptive microbiome-based survival analysis method. H. Koh.

2988F Lower frequency of genetic mosaicism observed on the X chromosomes of males relative to the X chromosomes of females. M.J. Machiela.

2989W Identifying the clinical impact of loss-of-function intolerant genes using SKAT-O PheWAS. R. Sivley.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists

2990T PheCLC: A novel statistical method for phenome-wide association studies. H. Zhu.

2991F Simulation study on different sample sizes for rare-variant association analysis. X. Zhang.

2992W ‡ DESCEND: Expression distribution deconvolution in scRNA-seq and characterization of transcriptional bursting and expression dispersion. J. Wang.

2993T A novel approach for parsing distribution of polygenic risk. L. Almasy.

2994F Statistical framework for integrating biological knowledge to accelerate discovery from GWAS data. S. Bhattacharjee.

2995W Improved methods to estimate functional enrichment from genome-wide summary association data. K. Burch.

2996T HiREPRO: Evaluating Hi-C data REProducibility via Regression. C. Crowley.

2997F Powerful and robust cross-phenotype association test for case-parent trios. T. Fischer.

2998W Learning causal networks of molecular phenotypes with Mendelian randomization. A.Q. Fu.

2999T A test for Hardy-Weinberg equilibrium in structured populations. W. Hao.

3000F PolyGEE: A generalized estimating equation approach to the efficient and robust estimation of polygenic effects in large-scale association studies. J. Hecker.

3001W Estimating genetic correlations in functionally annotated regions using genome-wide summary association statistics. D.L. Kassler.

3002T Estimating the contribution of gene-environment interactions to phenotypic variance. V. Laville.

3003F A generalized permutation testing method for binary trait association in structured samples. J. Mbatchou.

3004W ‡ Integrative analysis of eQTL and GWAS summary statistics to identify functional relationships. J. Morrison.

3005T Modeling ancestry-dependent phenotypic variance increases power in multi-ethnic association studies and enables detection of variance effects. S. Musharoff.

3006F ‡ Y chromosome variants associate with height but not disease risk factors: The Ygen consortium. N. Pirastu.

3007W ‡ Heritability informed power optimization (HIPO) leads to improved methods of discovering genetic association across multiple traits. G. Qi.

3008T We didn't see this in GWAS: Understanding and fixing unfamiliar problems in association analyses, when pooling whole genome sequence data from multiple studies. K. Rice.

3009F Investigating shrinkage methods to improve accuracy of GWAS and PRS effect size estimates. Y. Ruan.

3010W A simple, consistent estimator of heritability from GWAS summary statistics. A. Schork.

3011T GWAS meta-analysis allowing for sample overlap estimated using summary statistics. S. Sengupta.

3012F Statistical and population genetics of extreme phenotypes. O. Soylemez.

3013W Bayesian model averaging for the X-chromosome inactivation dilemma in genetic association study. L. Sun.

3014T Estimating the proportions of additive, dominant and recessive genetic effects. H. Wu.

3015F Scalable Bayesian functional genome-wide association study method with summary statistics. J. Yang.

3016W Using imputation for haplotype association. G.J.M. Zajac.

3017T Robustly doubling the sample size: A unifying regression framework for allele-based association test. L. Zhang.

3018F On simulation design for evaluating type 1 error: What is the 'correct' null model? T. Zhang.

3019W ‡ Efficiently controlling for unbalanced case-control sampling and sample relatedness for binary traits in PheWAS by large cohorts. W. Zhou.

3020T A Bayesian framework for transcriptome-wide association studies. J.D. Rosen.

---

‡ Indicates Reviewers' Choice Abstract; "ES" Indicates Epstein Trainee Award Semifinalist; "EF" indicates Epstein Trainee Award Finalists
The author listed is the first/presenting author of the abstract. The letter following each poster number indicates the day that the author will present at their posters. W=Wednesday; T=Thursday; F=Friday.