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.

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**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

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**Genome Structure and Function**

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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.


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


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


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

420F Discovery of rare, diagnostic AluYb8/9 elements in diverse human populations. J. Feusier.

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


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


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.


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.


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.


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


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POSTER SESSIONS

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.


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.


466W Identifying the genetic causes underlying perinatally 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. Sinaloj.

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.


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. Paret.

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.


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.


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.


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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.


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.


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


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.


517W All in the Family: How family history affects diagnostic yield of hypertrophic cardiomyopathy 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.


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


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.


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.

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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.
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.


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.


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.


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.


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.


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.

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


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


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

591F Effectiveness of a dedicated rotation in genomics, genetic medicine, and undiagnosed diseases for internal medicine residents. M. Wheeler.

590T Measuring health outcomes in telegenetics. D. Regier.


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

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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.


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

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661W Genetic variants in the 8q24 region are associated with progression in African Americans. H. Tung.

662T Tumor profiling of separated carcinomatous and sarcomaous components from uterine carcinosarcoma biopsies provides insights into their development. Z. Weber.

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.


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.

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

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

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


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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.

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

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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.

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


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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.

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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 CiP: 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.


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


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.


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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 IncRNA transcriptome of uncharacterized samples. A. Nath.

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

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

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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.

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

780F Functional validation of pleiotropic susceptibility 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.


783T 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.


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.


789F Familiarily-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.


796W Cryptic forms of gene 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.

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

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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.

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

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

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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.


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

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


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.

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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.


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


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

842T Whole exome sequencing of patient cell lines with high-persensitivity 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.

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


853W MAT1A variants in methionine adenosyltransferase deficiency (MAT1IIII) 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.

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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.


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


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


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.O. Carvalho-Siqueira.


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.


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

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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 nephropathis in Japan. N. Morisada.


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

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.


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. Waissfisz.

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.


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 metaphyseal chondrodysplasia due to a recessively inherited mutation in salt inducible kinase 3 (SIK3). F. Csukasi.


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.


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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.


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 revealing 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.


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-(TGGA)n-associated unconventional translation in spinocerebellar ataxia type 31. N. Sato.


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


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

945F ‡ Severity of GABBR2 mutations determines neurological phenotypes ranging from Rett-like syndrome to epileptogenic 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 (PA-FAH1B1) cause a unique leukoencephalopathy. L.T. Emrick.

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


954F Rare SOX30 variants in juvenile myoclonic epilepsy. S. Jais-hankar.

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.


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

959T Exome sequencing identifies a novel FBXO38 variant inherited from a mosaic mother to cause distal hereditary motor neuronopathy Type IIID 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.
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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.


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.


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 STXB1 encephalopathy with epilepsy: 6-year-old girl with de novo missense variant in STXB1 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.


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. Hebar.

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


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


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.


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-IIb). 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.

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


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

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


1007T Further clinical and molecular characterization of the 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<sup>F468C</sup> knock-in mouse model of the CMT2Q neuropathy. M. Gu.


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-pallidolysian 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.


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<sup>e1</sup> mutation. A. Goji.

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


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.


1036W Detection 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.

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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.


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: Mendelome 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.


1052F  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.


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


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


1064T  Exome Pool-Seq in neurodevelopmental disorders. C. Wei-er.

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.


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 mut0 model recapitulates key aspects of severe methylmalonic acidemia. K.T. Ellis.


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


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.


1082T  Identification of large effect variants underlying non-syndromic MA in families segregating the disease. G.A. Arenas-Perez.
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1083F DFNA5 and the infamous skipping of exon 8. K.T. Booth.


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 Schill.

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


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.


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.


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.


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. Fanidi.

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.

1110W ‡ Genetic characteristics of an international large cohort with Stargardt disease: The progression of atrophy secondary to Stargardt disease (ProgStar) study. K. Fujinami.


1112T Unclassifiable brachydactyly (brachydactyly 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, Mexico. 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-Shaefner syndrome and aniridia. G. Delplancq.


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<td>‡ A genotype-first approach identifies genes contributing to variable phenotypic presentations in a Smith-Magenis Syndrome cohort.</td>
<td>C. Zhang.</td>
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<tr>
<td>1156W</td>
<td>A novel FBXO28 frameshift in a patient with the predominant features of 1q41-q42 deletion syndrome: A case for haploinsufficiency and primary pathogenicity.</td>
<td>C.D. Balak.</td>
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<td>1157T</td>
<td>Exome sequencing in a family with autosomal recessive amelogenesis imperfecta.</td>
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<td>1158F</td>
<td>‡ De novo coding and noncoding variants in novel disease genes account for a significant fraction of isolated and complex congenital diaphragmatic hernia.</td>
<td>H. Qi.</td>
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<tr>
<td>1161F</td>
<td>Clinical exome data analysis and novel variant identification for MPS VII disease in GUSB gene.</td>
<td>A. Bhattacherjee.</td>
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<tr>
<td>1162W</td>
<td>Mutations in the condensin II component NCAPG2 cause autosomal recessive neurodevelopmental syndrome.</td>
<td>T.N. Khan.</td>
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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.


1172T ‡ An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease-gene discovery. A. Haghighi.


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.


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1178T A combinatorial approach for the selection of novel bioactive peptides. R.R. Handley.

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1180W Challenges in translating pharmacogenetic variants from Illumina’s Multi-ethnic Genotyping array into clinical practice. N. Rafaels.

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.


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.

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1188F Evaluation of genetic sequencing to improve newborn screening for VLCAD disease. B. Cai.


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.

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1201W Exome sequencing by NimbleGen kit is not suitable for SOX2 and SOX3 molecular screening due to bald spot. A.F.F. Bene-detti.


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.


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.


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.


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.


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.


1229T Novel pathway transcriptomics method greatly increases detection of molecular pathways associated with the trait. C. Chatzinakos.


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.


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. Pounrana.
1239F Improving pathogenicity prediction of structural variation in neurodevelopmental disorders: A machine learning approach. P.T. Tandon.


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.

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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 ClinGen Pathogenicity Calculator 2.0: New features and lessons learned from the data mining of 2,400 variant interpretations according to ACMG/AMP guidelines. K. Kallberg.


1251F Examining age, tissue, and genetic effects on RNA splicing with allele-specific resolution in a diverse mouse population. D.A. Skelly.


1254F Mixture models reveal multiple positional bias types in RNA-seq data and lead to accurate transcript concentration estimates. J. Aleksejeva.


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.


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.


1262T A high-throughput pipeline for genotyping within primary health care to enable precision prevention. S. Jai Kumar Ahuja.


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.

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1269F WGS A07: 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 hematopoietic 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. Produtur.


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1281F  Cloud-based quality measurement of whole-genome cohorts. W. Salerno.


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.

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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. Plewczyński.

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.


1299F  Enhancing discoverability and reusability of 1.2 million human genomic datasets. M. Corpora.

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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.

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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. Mihailek.

1315W  A novel computational strategy for DNA methylation prediction. F. Yu.

1316T  Scaling workflows for growing microbiome applications. J. Lai.

1317F  Deriving disease signatures using data driven approaches 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.


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.

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1323F PubCases: A diagnosis assistant tool for rare diseases based on disease-phenotype associations extracted from published case reports. T. Fujiwara.


1325T Search Candidate Regulatory Elements by ENCODE (SCREEN): A web-based tool for visualizing genomic annotations. H.E. Pratt.


1327W Improving quality of variant calling by integrating whole genome and whole exome sequencing from same samples. X. Li.


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.


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.

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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.

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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.


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.


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.


1355T PathwayMatcher: Direct mapping of omics data to the Reactome pathway knowledgebase. L.F. Hernandez Sanchez.

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.
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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.


1367T Optimal workflow for next generation sequencing data processing using existing technology. J.E. Martin.

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1371F Improving SNP array copy-number variant calling using site-specific variance models and windowed intensity normalization. T. Poterba.

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1374F IMSindel: An accurate intermediate-size indel detection tool incorporating de novo assembly and gapped global-local alignment into split reads. D. Shigemizu.


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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.


1380F UDPICS, a laboratory-medical informatics system for improving translational research in the NIH Undiagnosed Diseases Program. Z.M. Valivullah.

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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.

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1385T An alignment-based method to trim adapter and filtrate low-quality reads from pair-end FASTQ files. R. Ye.


1388T‡ From quantity to quality: A population-based approach for building reference panel imputation. M. Cocca.

1389F A new diagnostic platform (Genomic Intelligence®) improves accuracy of whole exome and genome sequencing diagnosis in rare disease. A. Fisher.


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.


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.


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.


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.


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1404F  Polaris: A collection of 220 publicly available whole genomes for sharing validated structural variants. M.A. Bekritsky.


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.


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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 phenotype-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. Yu.

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.


1430T ‡ Impact of polygenic risk on changes in biomarkers over time due to lifestyle intervention and aging. M. Conomos.


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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.


1440F  PQC: A phenotype checking and tracking tool. N.W. Rayner.

1441W  Recombination rate estimation in large-scale genetic data. S. Choi.


1444W  Effects of filtration on imputation in clustered variants. C.M. Charon.

1445T  Modeling and analysis of RNA structuromes. Z. Ouyang.

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.


1454T General validation framework using semi-supervised learning on complex cDNA 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.

1458F Hypertriglyceridemia as a presenting sign of HMG-CoA synthase deficiency. E. Conboy.


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.


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.


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.


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1484T Personalized dosing of dichloroacetate by clinical genotyping assay. T. Langae.

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. Hastié.

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.


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 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. Amr.

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.


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.


1520T Automation of ultra-high molecular weight DNA isolation and labeling for genome mapping. P. Lynch.


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.

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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. Ulthe.

1533F  Targeted sequencing using a long-read sequencing technology. J. 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. 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.


1540W  Streamlined, efficient, and uniform molecular inversion probe capture for targeted sequencing. J. Uthe.

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.

1545T  The molecular basis of increased diabetes susceptibility in carriers of the PGC1α (Ser482) risk allele. R. Van den Eeck.

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.


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. A. Justice.


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-naive multiple sclerosis cases and controls exhibit differentially methylated regions in CD4+ and CD8+ T cells. B. Rhead.


1565T  Integrative analysis identified pervasive long-range regulation mediated by super-enhancers shared in multiple autoimmune diseases. X.F. Chen.

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1566F ‡ Cis regulatory variation determines dynamic HLA-DQB1 alleric expression in response to T cell activation. M. Gutierrez-Arcelus.


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.


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 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.


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.


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 associated with hidradenitis suppurativa (acne inversa). D. Jhala.


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 methylymic profiles between blood and cerebral spinal fluid in aneurysmal subarachnoid hemorrhage patients. A. Arrockiaraj.

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.


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.


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.


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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 Methylation 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 fashkodi.

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.


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. Svoboda.

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.


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.


1647F ‡ Comprehensive functional annotation of the zebrafish genome. T. Liu.

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1648W DNA methylation and its impact on inter-population differences in disease risk and prognosis. M. Loh.


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.

1655F 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 PPARGCT1A 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.

1661F 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 450K 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.

1666F 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.


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.


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.


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.

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Developmental Genetics and Gene Function

1692F Interaction of exocrine and endocrine pancreas in obesity ciliopathies. T. Hostelley.


1694T Haploinsufficiency of a histone modifier, Kmt2d, in a mouse model of Kabuki syndrome leads to widespread defects in the B cell lineage. G. Pilarskow.

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-YPos 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 NKKX2-5/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.


1705W Gene expression profiling of single oocytes reveals pathways and regulators involved in follicle activation. Y. Lyu.


1709T Missense and splicing mutations in the retinoic acid catabolizing enzyme CYPI26C1 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-Serre.

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.


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 pentylentetrazole 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. Alamedy.

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.

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1728F Pathologic characterization of a neurodevelopmental abnormality in the cerebral cortex of domestic cats with loss of PEA15. E.C. Graff.


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 knockout 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 Irdi.

1734F Neuronal inflammation and dysregulated mitophagy features in ubiquitous Clec16a knockout 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 22q11.2DS. W. Manley.


1739T The chr14.232.a pseudogene in the 14q21.2 region regulates the expression of the contiguous LRFN5 gene and is deleted in a man skeletal myotubes. R. Oliveira.

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-Rondon.

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. Gotlia.

1747W Rare family with partial duplication in 7q11.23 link four genes associated with intellectual delay and autistic phenotypes. J.R. Korenberg.


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.


1754T MeCP2 AT-hook1 mutations disrupt DNA binding and chromatin compaction in patients with intellectual disability and schizophrenia. T.I. Sheikh.

1755F De novo IRF2BP1 pathogenic variants cause severe precarious 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 homedomain 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.


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 nyctagmus in a Brazilian boy with septo-optic dysplasia. L. Gabriel.


1766T Bardet-Biedl syndrome, postaxial polydactyly, Shh signaling pathway and a founder effect in a Libyan extended consanguinous family from Tarhunah Berber tribe. N. Bouayed. Abdelmoula.
1767F Acquired ventriculomegaly in a case with SOX 9 mutation. A. Matsumoto.

1768W The role of WNT regulatory variants in nonsyndromic cleft lip and palate. L. Malii.

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 KA76A 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.


1784T Mechanistic insight of inactivation of mouse chitinase-like protein Ym1. F. Oyama.


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.


1791F 30 kDa fragment of beta-dystroglycan co-immunoprecipitates with Dp71 isoforms in PC12 cells. C.O. Azotila Vilchis.

1792W Analysis of copy number variation and association with facial shape in a large cohort of Bantu African children. F. Yilmaz.

1793T ARMCS and PMAH: From human genetic defects to the Armc5+/- mouse. F.R. Fauccz.

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.


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.


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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.


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. Soookoian.

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.


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. Roshandeh.

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 glycemic 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.


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.


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.

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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. Emma-nouillidou.


1847T Genetic evidence that early carbohydrate-stimulated insulin secretion affects accumulation and distribution of adiposity. C.M. Astley.


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.

1853F 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.


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 and their sexual dimorphism. A. Alanne-Kinnunen.

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.


1869F ‡ Genetic analysis of lipids in >300,000 participants in the Million Veteran Program. D. Klarin.


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.

1873F ‡ 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.

1875W Lipid loading in human liver cells induces differential expression of 88 genes. M. Alvarez.

1876F ‡ Genome-wide association study of anthropometric, cardiovascular, and eczema. W. Ek.

1877F Multi-omic approaches to identifying clinical biomarkers of asthma exacerbations in African Americans. H. Gui.

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.

1880F Shared genetic etiology and ancestry variations between asthma and major complex diseases. T. Mersha.


1883T Unraveling the genetic architecture of generalized vitiligo in a homogeneous, isolated Romanian village. G. Andersen.

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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.


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.


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.


1901T Identification of one novel IBD susceptibility locus through a genome-wide association study in Korean populations. B.D. Ye.


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°, are associated with drug-induced liver injury due to flucloxacinil 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.


1915W Exome sequencing identifies variants of the alkylglycerol monoxygenase 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.


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.

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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 MNM12 and
HCP5 as susceptibility loci for Kawasaki disease. J. Kim.

1926F  Immunochip v2.0 meta-analysis identifies shared genetic

1927W  GWAS identified associations of HLA-DRB1-DQB1 haplo-
types and BTNL2 gene with response to a hepatitis B vaccine. N.
Nishida.

1928T  Reduced severity of collagen-induced arthritis in peptidyl-
larginine deiminase type 4 knockout mice. A. Suzuki.

1929F  A locus on chromosome 5 shows African-ancestry-limit-
ed 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 erytheta-

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

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 ani-

1935F  Sex-specific transcriptional responses to lipopolysaccha-
ride (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 meningi-
tis: Exome sequencing and a GWAS in a South African population.
M. Möller.

1938F  Genome wide association in Peru demonstrates that pro-
gression to active tuberculosis is a polygenic and highly heritable
trait. S. Raychaudhuri.

1939W  Genetic variation in GLS2 is associated with development of

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 poly-
morphism with Kawasaki disease. Y. Kwon.

1946T  Local ancestry interaction models reveal a novel asthma
association with asthma on chromosome 1q23.1 specific to people

1947F  A genome-wide association analysis of Hashimoto’s thyroid-
itis. 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 popula-
tion. M. Kawashima.

1951W  Exome-wide association study of kidney function in 55,041
participants of the DiscovEHR cohort. C. Schurmann.


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 associat-
ed 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 myelo-
suppression 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 co-
lonic diverticula in Korean population: Genome-wide association
study. E. Choe.

1961T  Dysregulated gene and miRNA expression in different stag-

1962F  Comparative bacteria communities between gallbladder bile
and gallstone in gallstone disease patients in Taiwan. H. Yang.

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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.


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. Atkins.

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. Dasas.

1973T Genome-wide CNV analysis identifies TGFB3 as a candidate causal gene for endometriosis and infertility. E. Hatchwell.


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.


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.


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.


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.


1990W Exome sequencing in idiopathic scoliosis families implicates cilia genes in disease etiology. E.E. Baschal.


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.


1997T Polymorphisms of genes involved in extracellular matrix homeostasis may play a role in the risk to develop anterior cruciate ligament and meniscal tears. L. Casilla.

1998F Filaggrin variations associated to atopic dermatitis in Ecuadorian pediatric population. V. Romero.


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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.


2009T A whole exome study of Alzheimer’s disease which is augmented by population data found the noble AD risk genes. J. Kim.


2012T ‡ Novel Alzheimer disease loci identified in subsets of whole exome sequencing data stratified by APOE genotype. Y. Ma.


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.


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 FXB040 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. Luiz.

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. Veeerappa.

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.


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.

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POSTER SESSIONS

2040F Identification of somatic mutations in malformations of cortical development. N.G. Griffin.


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.


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.


2057T ONDRISeq: Genetic diagnosis of neurodegenerative disease patients using targeted next-generation sequencing. A.A. Dilliot.

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.


2061F The NINDS human genetics DNA and cell line repository: A publicly available biospecimen collection for neurological disease research. A. Resch.


2064F Common variants at 5q33.1 predispose to migraine in African-American children. X. Chang.


2066T No rare deleterious variants from STK32B, PPARC1A, 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.


2071W Decoding GWAS discoveries of neurodegenerative diseases: Gene expression changes in single neurons. L. Tagliaferro.

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.

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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. Haripaul.

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2082F Gene expression profiling predicts clinical outcome in autism spectrum disorder: Confirmation of potential biomarkers and initial characterization of clinically homogeneous subgroups. F. Macciardi.


2084T Epigenetic dysregulation of DYRK1A may have a role in ASD development in a discordant monozygotic twin pair. C. Sjaarda.


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.

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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 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. Ort.

2103F Copy number variation analysis of psychiatric traits in a large population-based sample of youth. M. Zarrei.


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.


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, non-coding 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. Asian.


2115F Parent-of-origin and maternal effects in attention deficit hyperactivity disorder. D. Smajagic.

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.

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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.


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-naïve schizophrenia patients: A longitudinal study of DNA methylation and neurophysiological phenotyping. C. Chen.


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.


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. Kuksal.

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 their relationship to genetic liability for schizophrenia. H. Young.

2144T Genetics of attention deficit hyperactivity disorder dimensions. T. Zayats.


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.


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.


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 homozogous 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.

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2160F A hypertension-associated mtDNA mutation alters the tertiary interaction and function of tRNALeu(UUR). M. Guan.


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.


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.


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.

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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.


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 Ca2+ 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.

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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.


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. Rajabi.

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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.

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2204T Allele-specific expression in healthy centenarians. L.C. Tindle.

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2231T Whole exome sequencing reveals a novel candidate gene, HSPA1L, for spontaneous preterm birth. J.M. Huusko.


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2256F Defining the opportunity: The IGNITE CPIC Prescribing Study. L. Wiley.


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2304F Tracing maternal lineage of Austronesian-speaking Melanesians and Micronesians in the Solomon Islands. M. Ishiki.


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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.


2321T Archaic-genome-agnostic detection of introgressed segments. S.R. Browning.

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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.


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.


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.

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2337F † A comparative study of endoderm differentiation in humans and chimpanzees. L.E. Blake.

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2339T The composition and intensity of de novo mutations in the Lithuanian exome. L. Pranckėnienė.

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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 massively 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.

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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 chronic lymphocytic leukemia. A. Dowiak.

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2387T Hereditary cancer risk testing within a Colombian cohort reveals high incidence of Lynch syndrome. J.A. Rugeles.

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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 study 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.


2648T Arrhythmia and night vision blindness: Chicken and egg? Or could be chicken or egg? A. Faucon.

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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.


2662W Uncovering the genetic determinants of variation in arterial stiffness through joint location and scale association testing. D. Soave.

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2664F Evaluating the burden of pathogenic variants for the inherited arrhythmia syndromes. Y.P. Fu.

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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.

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2671W A genome-wide gene by cigarette smoking interaction study on elevated blood pressure. M. Kang.

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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.

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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.


2682F Whole genome cardiac DNA methylation fingerprint and gene expression analysis provide new insights in the pathogenesis of chronic Chagas disease cardiomyopathy. C. Chevillard.


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.


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.


2696T The role of Kringle IV 2 copy number variation and SNPs on Lp(a) levels and cardio-metabolic risk. S.E. Ruotsalainen.


2698W Fitness, physical activity, and cardiovascular disease: Longitudinal and genetic analyses in the UK Biobank Study. E. Tikkkanen.

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.


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.


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-Martín.

2710W Family based method for the discovery of rare high penetrance sequence variants. G. Sveinbjörgsson.

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.


2714T Low frequency and rare variants of RBFOX1 are associated with blood pressure. K.Y. He.

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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. Manka.

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. Ramachandrule.

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.


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.


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. Komucu-Bayrak.

2742F Investigation of microRNA expression in coronary artery disease. N. Coban.


2745F Gender transcriptome signatures for congenital heart defects (CHD) children based on next generation sequencing technologies of cardiac tissues. C. Kim.


2747T Epigenetic regulation of PAR-4-mediated platelet activation: Understanding the mechanistic links between smoking and cardiovascular disease. N. Timpson.


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.

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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.


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-naive adults with Gaucher disease type 1: Final efficacy and safety results from a phase 2 clinical trial after 8 years of treatment. H. Lau.


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.


2780T Socioeconomic deprivation amplifies genetic susceptibility to obesity and its comorbidities. K.A. Kentistou.

2781T Effects of the interaction between a CREB5F 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.

2786F 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.


2789T Targeted sequencing of 109 genes in the eMERGEseq panel uncovers novel variants and genes influencing triglyceride levels. X. Fan.

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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.


2793F ‡ Adaptive multi-trait association test using GWAS summary data. B. Wu.


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.


2804T Association study of R3HD17 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.


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.


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.


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-occurrence 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.


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.


2830W Relationship between essential tremor and Parkinson’s Disease. A.A. Gosch.
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 nommates novel genes. J.R. Shaffer.


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.


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.


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 1L1RAPL2 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. Waksman.

2867T Genetic risk score is associated with vertical cup-disc ratio and improves prediction of primary open angle glaucoma in Latinos. D.R. Namini.

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.

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2871F Iterating from discovery to epidemiological consequence through disease mechanism. J. Brown.


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 (TLT) 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.

2883T 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.


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.


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.


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.

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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.


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.


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.


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.


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 difficile Infection (CDI) recurrence in patients treated with bezlotoxumab. J. Shen.

2934F ‡ A CREB1 missense mutation substantially affects height in Samoans. S.L. Rosenthal.


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.

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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.


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.


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.


2960T Reverse regression enables disease only case-control association studies for burden tests. J. Tom.


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.


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.


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 Detectingheritable phenotypes without a model: Fast permutation testing for heritability and set-tests. R. Schweiger.


2982F Combining sequence data from multiple studies: Impact of analysis strategies on rare variant association results. Z. Chen.


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

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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 REProductibility 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.


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. Mbachou.

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.


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.