
Genome Structure and Function

- 375F** A novel IDS gene mutation in two Japanese patients with severe mucopolysaccharidosis type II and correlation between developmental outcomes. S. Kasuga.
- 378F** Cargo of miRNAs from synovial fluid exosomes contributes to pathogenesis of primary osteoarthritis. M. Czarny-Ratajczak.
- 381F** Gene correction of epidermolysis bullosa simplex mutations using CRISPR/Cas9 technology. M. Bchetnia.
- 384F** Molecular mechanisms in C9orf72 ALS/FTD using the BAC transgenic mouse model with behavioral, neuropathological and molecular features of disease. A. Pattamatta.
- 387F** Prolonged pentylenetetrazole exposure modulates the Rho Family GTPases pathway in the zebrafish brain. M.C.S. Nunes.
- 390F** Marker chromosome architecture and temporal origin revealed in a family with pleiotropic psychiatric phenotypes. C.M. Grochowski.
- 393F** Loss of Kctd13 in mice causes short-term memory deficiency. T. Arbogast.
- 396F** Single cell allele specific expression (ASE) in T21 and common trisomies: A novel approach to understand Down syndrome and gene dosage effects in aneuploidies. G. Stamoulis.
- 399F** Trisomy 21 and chromatin. S.E. Antonarakis.
- 402F** Exon-intron architecture in high and low GC-content genes affects alternative splicing. L. Tamer.
- 405F** Revisiting the mouse reference genome: Single molecule sequencing of C57BL/6J "Eve". A. Srivastava.
- 408F** MethylHiC reveals long-range genetic-epigenetic and epigenetic-epigenetic interactions within the same single molecule. Y. Liu.
- 411F** Targeted next-generation sequencing for identifying genes related to horse temperament. S. Song.
- 414F** Study of G2 phase cell cycle arrest and its application in mediating SOX9 mutagenesis in pluripotent stem cells (PSCs). T.Y. Ha.
- 417F** Gene expression analysis and enzyme function of pig mammalian chitinase. E. Tabata.
- 420F** Discovery of rare, diagnostic *AluYb8/9* elements in diverse human populations. J. Feusier.
- 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.
- 429F** Genomic and structural integrity of human induced pluripotent stem cells. K. Kanchan.
- 432F** ‡ Germline *de novo* mutation clusters arise during oocyte aging in genomic regions with increased double-strand break incidence. C. Gilissen.

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

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

Prenatal, Perinatal, and Reproductive Genetics

- 441F** Alteration of the expression and methylation modification of RAS in cardiomyocyte tissue of the mice conceived by in-vitro fertilization. Q. Wang.
- 444F** Compromised DNA repair and genomic imbalances in human male infertility. V. Singh.
- 447F** Transethnic meta-analyses from genome-wide association studies of fibroid characteristics in African and European American women. M.J. Bray.
- 450F** Neurodevelopment in Japanese singletons, aged 4–6 years, conceived by assisted reproductive technologies. T. Shimada.
- 453F** Single-cell RNA sequencing in sperm from fathers of autistic children. J.A. Rosenfeld.
- 456F** ‡ Large-scale cytogenomic analysis of samples from conception to childhood: A comprehensive assessment of the landscape of unbalanced genomic abnormalities. T. Sahoo.
- 459F** Carrier screening for 316 monogenic recessive diseases revealed high carrier frequency of rare known pathogenic mutations. D. Bercovich.
- 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.
- 465F** Prenatal diagnosis of Roberts syndrome suspected on sonogram in a consanguineous couple: A case report and a review of the literature. J.R. King.
- 468F** WES identifies likely pathogenic *FANCG* variants in a fetus with multiple congenital anomalies. B.D. Webb.
- 471F** Prenatal evaluation of a fetal cystic hygroma: An unexpected finding of a *de novo* fetal BRCA1 deletion. N.S. Seligman.
- 474F** ‡ Uncovering novel cytogenetic and molecular etiologies for infertility. S.L.P. Schilit.
- 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.
- 480F** Modeled fetal disease risk of a 175 condition expanded carrier screening panel. K.A. Beauchamp.
- 483F** Bacterial taxonomic analysis of oral microbiome in spontaneous preterm birth for Maternity Log Study. D. Ochi.
- 486F** Correlation between nutrigenetic variants, adherence to the Mediterranean diet and obesity in male infertility. M. La Rovere.

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

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

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

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

Genetic Counseling, ELSI, Education, and Health Services Research

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

Cancer Genetics

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

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- 600F In-frame germline deletion of exons 23 and 24 results in DICER1 syndrome.** M. Apellaniz-Ruiz.
- 603F Genotoxic chemotherapies and radiotherapy contribute to the development of multiple primary tumors in patients with Li-Fraumeni syndrome.** E. Kasper.
- 606F Cancer following radiotherapy for primary cancers in Li-Fraumeni syndrome patients.** L. Oba.
- 609F Clinical and genetic analysis of patients with hamartomatous polyposis syndromes.** J. Oh.
- 612F Functional characterization of germline *TMEM127* mutations in familial pheochromocytoma.** S.K. Flores.
- 615F Germline variants in mismatch repair genes are associated with microsatellite instability in sporadic tumors.** A.R. Buckley.
- 618F CDKN2A germline polymorphisms demonstrate parallel associations of disease risk and clinical outcome in melanoma patients.** S. Fang.
- 621F Role of *HOXB13* in breast and ovarian cancer: Preliminary data from a laboratory-based multigene panel testing cohort.** C. Horton.
- 624F Rare DNA repair gene mutations predispose to young onset and lethal prostate cancer in the UK.** Z. Kote-Jarai.
- 627F BRA-STRAP: BRCA Refined Analysis of Sequence Tests: Risk And Penetrance.** T. Nguyen-Dumont.
- 630F Oral findings of cancer predisposition conditions: “Red flags” that dental, oral and craniofacial providers should recognize for early diagnosis, referral and management.** A.M. Pham.
- 633F Identification of novel prostate cancer susceptibility loci in Finnish population.** C. Sipeky.
- 636F Association of *TLL1* variant with hepatocellular carcinoma developed after eradication of hepatitis C virus.** K. Tokunaga.
- 639F No association between 135G>C polymorphism of *RAD51* and colorectal cancer in Iranian population.** N. Yazdanpanahi.
- 642F Characterization of global molecular architecture and regulatory mechanisms underlying hepatocellular carcinoma.** H.M. Natri.
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- 648F A genome-wide association study of cisplatin-induced tinnitus in adult cancer survivors.** B.L. Mapes.
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- 657F Novel analysis incorporating multiple tumor characteristics provide evidence of highly heterogeneous associations for known breast cancer risk loci.** T.U. Ahearn.
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- 663F Classification of lung adenocarcinoma using optimized support vector machines applied to gene expression data.** J.S. Diaz.
- 666F Prevalence of cancer predisposition gene mutations among unselected pancreatic cancer patients.** C. Hu.
- 669F Whole exome sequencing reveals genes with elevated germline rare variants burden in myeloid malignancy patients.** S. Li.
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- 675F A *cis*-eQTL genetic variant of the cancer-testis gene *CCDC116* is associated with risk of multiple cancers.** N. Qin.
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- 690F Detection of epigenetic field defects using weighted epigenetic distance-based method.** Y. Wang.
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- 696F Efficient gene-by-treatment interactions test to develop predictive biomarkers on genome-wide studies using multidimensional hierarchical mixture models.** T. Otani.
- 699F Proteomics and genomics integration to predict ovarian cancer survival.** U. Ozbek.
- 702F Gender differences in germline mutations in bladder cancer participants from the DiscovEHR study.** L. Bang.
- 705F Detection of signal regions in whole genome genotyping and sequencing association studies using scan statistics.** Z. Li.
- 708F Clinicopathological and prognostic significances of EGFR, KRAS, BRAF and PI3KCA mutations in biliary tract cancer.** H. Lee.
- 711F Similar frequency of ACMG-59 gene secondary findings in a large whole exome sequenced cancer cohort and ethnicity-matched controls.** J. Kim.
- 714F Systematic evaluation of copy number variations: Towards rational personalized cancer therapy.** S. Appenzeller.
- 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.

- 720F Copy number segmentation with left-to-right hierarchical Dirichlet process hidden Markov model and segment clustering.** K. Liao.
- 723F ViFi: Virus integration and fusion identification in tumor samples.** N. Nguyen.
- 726F Pathogenicity of Mutation Analyzer (PathoMAN): A fast, automation of germline genomic variant curation in clinical sequencing.** V. Ravichandran.
- 729F Practical noninvasive biomarkers: Identification of blood gene signatures for diagnosis of lung cancer.** B. Song.
- 732F Access and discover pathways from Pathway Commons.** J.V. Wong.
- 735F Evaluating relationships between pseudogenes and genes: From pseudogene evolution to their functional potentials.** Y. Zhang.
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- 741F Genomic instability phenotypes in multidimensional genomic cancer studies.** B.N. Lasseigne.
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- 753F RADAR: A RNA binDing protein regulatory network resource for cAncer Research.** J. Zhang.
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- 762F Designing for success: The right CRISPR design strategies for the right experiment.** L. Brody.
- 765F Somatic mutation identification through haplotype discrepancy in tumor-only sequencing without matched normal DNA.** W. Chen.
- 768F The NantOmics Pharmacogenomics Test: An integrative panomic approach to pharmacogenomics screening.** C. Schwartz.
- 771F Identification of potential LREA regions in prostate cancer cell lines using multi-omic analysis features of Strand NGS software.** P. Karuna.
- 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.
- 777F Digital gene expression from low sample input: Highly multiplexed and robust profiling of formalin-fixed paraffin-embedded (FFPE) and fresh frozen samples from as little as 1 ng of RNA using the nCounter® Platform.** D. Hanson.
- 780F Functional validation of pleiotropic susceptibility loci for breast and ovarian cancer using chromosome conformation capture technology.** J. Plummer.
- 783F Comparative analysis between gene expression profile and genomic profile in adrenocortical carcinoma samples.** F.P. Fortes.
- 786F DNA repair improves sequencing accuracy in FFPE DNA samples.** F. Stewart.
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- 792F Highly efficient double-stranded molecular tagging empowers improved accuracy of ultra-low frequency mutation detection.** J. Wang.
- 795F A novel NGS target enrichment technology: Improved speed, selectivity, and uniformity.** J. Pel.
- 798F Trans-eQTLs in prostate cancer risk.** M. Bicak.
- 801F The role of antioxidants in the context of carcinogen induced chromosome aberrations.** Y.C. Li.
- 804F Rhesus macaques with mutations in *MLH1* and *MSH6* develop Lynch syndrome colorectal cancers.** M. Raveendran.
- 807F ‡ Functional characterization of a novel prostate cancer candidate gene at 2q37 in normal human prostate cell line.** C. Cieza-Borrella.
- 810F De novo inference of enhancer-gene networks in diverse cellular contexts reveals the long-range regulatory impact of disease-associated variants.** J. Wang.
- 813F In-depth analysis of genomics and epigenomics identifies a novel susceptibility lncRNA *GCLET* for gastric cancer.** M. Du.
- 816F Molecular mechanisms underlying serrated polyps: Comprehensive DNA methylation analysis reveals new targets in the serrated neoplasia pathway.** V. Khammad.
- 819F Motif disruption domains lead to cancer gene expression rewiring.** F.C. Lamaze.
- 822F Genetic polymorphism and gene expression of *SHH* & *PI3K* gene in ameloblastoma.** H. Singh.
- 825F Integrating chromatin and expression variation in statistical fine-mapping.** M. Roytman.
- 828F Genome-wide map of APA in lung cancer: A pilot study.** A. Zingone.
- 831F Colorectal cancer-upregulated long non-coding RNA *linc-DUSP* regulates DNA damage response genes and promotes resistance to apoptosis.** M.E. Forrest.

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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.

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

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

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

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.

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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.

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

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

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

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

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

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

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

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

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

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

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

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

939F ‡ Expanded-(TGGAA)_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.

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

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- 966F A novel homozygous mutation in two sisters diagnosed with Joubert syndrome.** A. Sen.
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- 996F Spectrum of *TTN* variants in a patient cohort of neuromuscular disorders.** P.S. Lai.
- 999F Correction of *NAGLU* mutation p.R297X using CRISPR/Cas9 gene editing in mucopolysaccharidosis IIIB patient-derived iPSCs.** C.L. Christensen.
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- 1005F A novel pathogenic variant of *PURA* in a patient with severe developmental delay, delayed myelination and empty sella.** K. Hosoki.
- 1008F Evaluating clinical and biochemical endpoints for therapy in Pex7 deficient mouse models.** W. Fallatah.
- 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.
- 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.
- 1017F Unbalanced translocation causing unbalanced brain: A case of hemimegalencephaly.** A.R. Barone.
- 1020F Identification of novel *SNORD118* mutations in seven patients with leukoencephalopathy with brain calcifications and cysts.** K. Iwama.
- 1023F A de novo *HNRNPU* gene mutation identified in a patient with symptomatic infection-associated acute encephalopathy and developmental delay.** S. Shimada.
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- 1041F Neurodevelopmental profile for boy with unique 6p deletion.** A. Saba.
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- 1050F A 10q23.31 microduplication is associated to autosomal dominant primary microcephaly.** D. Oliveira.
- 1053F Mutation in *OASL* gene causing speech delay and intellectual disability.** M. Alfadhel.
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- 1059F A novel missense variant in the *POMK* gene causes Walker-Warburg syndrome.** E. Preiksaitiene.
- 1062F The phenotypic spectrum of Xia-Gibbs Syndrome.** Y. Jiang.
- 1065F Novel mutation in *ARHGEF9* associated with developmental delay and seizures.** E. Fattakhov.

- 1068F** Family-based whole exome sequencing for identifying novel variants in primary myopathies. J. Hwang.
- 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.
- 1074F** Novel biallelic mutations in the *PNPT1* gene encoding a mitochondrial-RNA-import protein PNPase cause delayed myelination. A. Kikuchi.
- 1077F** The clinical spectrum of *BCS1L* mutations: Case report of a novel mutation and review of the literature. J. Priestley.
- 1080F** A viable knockout murine model of *Mmaa (cbIA)* deficiency provides a platform for microbiome manipulations. A.F. Lesser.
- 1083F** *DFNA5* and the infamous skipping of exon 8. K.T. Booth.
- 1086F** In silico analysis and identification of *TYR* mutations in a Cypriot family. R. Kalkan.
- 1089F** Single base deletion in *ATOH7* gene causes abnormal eye development in a consanguineous family. M. Ansar.
- 1092F** Genetic causes of *CHARGE* syndrome identified by whole exome sequencing. D.M. Martin.
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1845F Zebrafish larvae as a model system for high-throughput, image-based screens in insulin resistance and diabetes. A. Emmanouilidou.

1848F Exome sequencing in African American children with early-onset obesity reveals new insights. A. Chesni.

1851F A dinucleotide deletion in a putative miRNA target site in long-chain fatty acid elongase Elov16 associates with higher thermogenesis and lower body mass index in Pima Indians. P. Kumar.

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.

1860F Population and medical genetics of the Kibbutzim Family Study. S. Carmi.

1863F Genome-wide association study of HDL efflux phenotypes in 5,143 French Canadians. K.S. Lo.

1866F Low LDL cholesterol concentrations are associated with increased risk of type 2 diabetes. Q. Feng.

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

1872F ‡ Gender differences in genetics of body composition and obesity traits after an intensive exercise intervention. A.I. Vazquez.

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

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.

1881F Shared genetic etiology and ancestry variations between asthma and major complex diseases. T. Mersha.

1884F Polygenic risk score predicts development of HCV-associated mixed cryoglobulinemia and response to interferon-free therapy. M. Artemova.

1887F Immune-phenotypes among patients with systemic lupus erythematosus and their association with *HLA-DRB1* alleles. L.M. Diaz-Gallo.

1890F Association of protein tyrosine phosphatase non-receptor N22 gene functional variant R620W with systemic lupus erythematosus patients from Kuwait. M.Z. Haider.

1893F *TYK2* correlates with multiple sclerosis symptomatology at onset. J. Jiangyang.

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- 1896F** Identification of rare variants in Italian multiplex families with multiple sclerosis using a next generation sequencing approach. E. Mascia.
- 1899F** Longitudinal clinical, molecular, and immunohistochemistry changes during mycophenolate mofetil therapy in patients with systemic sclerosis. D.M. Toledo.
- 1902F** The influence of human genetic variation on HIV related non-Hodgkin lymphoma. C.W. Thorball.
- 1905F** Autoimmunity genes are associated with acquired hypothyroidism in a large clinical cohort. J. Freudenberg.
- 1908F** Genome-wide association study of otitis media in children. J. Li.
- 1911F** *NKG2D* variation and viral bronchiolitis. A. Pasanen.
- 1914F** Genetic footprints and functional analysis of polymorphisms in the *PKLR* gene. O.C.L. Bezerra.
- 1917F** A long non-coding RNA in the rheumatoid arthritis risk locus at chromosome 18 is involved in T cell activities. M. Houtman.
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- 1923F** Genetic markers associated with leprosy susceptibility in a group of incident household contacts from Brazil. L. Arnez.
- 1926F** Immunochip v2.0 meta-analysis identifies shared genetic loci for inflammatory bowel disease in Korean population. S.B. Lee.
- 1929F** A locus on chromosome 5 shows African-ancestry-limited association with alloimmunization in sickle cell disease. L.M. Williams.
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- 1938F** Genome wide association in Peru demonstrates that progression to active tuberculosis is a polygenic and highly heritable trait. S. Raychaudhuri.
- 1941F** Whole genome sequencing of pharmacogenetic drug response in racially and ethnically diverse children with asthma. A.C.Y. Mak.
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- 1947F** A genome-wide association analysis of Hashimoto's thyroiditis. V. Boraska Perica.
- 1950F** *NELFCD* and *CTSZ* loci are associated with jaundice-stage progression in primary biliary cholangitis in the Japanese population. M. Kawashima.
- 1953F** ‡ Chronic obstructive pulmonary disease subtyping through multiple -omics data integration. B.D. Hobbs.
- 1956F** Effect of CAG repeat length in the *androgen receptor* gene on hirsutism among healthy Israeli women of different ethnicities. S. Ben-Shachar.
- 1959F** Exome sequencing highlights novel DNA variants with a potential role in polycystic ovary syndrome. S.G. Wilson.
- 1962F** Comparative bacteria communities between gallbladder bile and gallstone in gallstone disease patients in Taiwan. H. Yang.
- 1965F** Evaluation of candidate genes for Hirschsprung disease using target sequencing. W.Y. Lam.
- 1968F** Differential metagenomic analysis associated between alcoholic and non-alcoholic fatty liver disease using 16S rRNA gene sequencing. Y. Yun.
- 1971F** Patterns of *APOL1* G1 association with kidney function in young adults in five African diaspora populations. B. Tayo.
- 1974F** Identification of lung cell populations from single-cell RNA-seq profiling of murine emphysema model. J.H. Yun.
- 1977F** Chromatin interactions reveal novel gene targets for drug repositioning in rheumatic diseases. P. Martin.
- 1980F** Identification of genetic variants associated with knee OA in patients with knee replacement surgery. Y. Zhang.
- 1983F** Copy number variant causes the mis-calculation of single nucleotide polymorphism in association analysis. Z. Yan.
- 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.
- 1989F** Longitudinal genome-wide association analyses and heritability estimates of pediatric bone mineral density. D. Cousminer.
- 1992F** The genetic architecture and phenotypic spectrum in the skeletal ciliopathies. W. Zhang.
- 1995F** Characterizing the molecular biology of systemic sclerosis with RNA-Seq of both skin and PBMCs. E. Roberson.
- 1998F** Filaggrin variations associated to atopic dermatitis in Ecuadorian pediatric population. V. Romero.
- 2001F** Whole-exome sequencing analyses for late-onset Alzheimer's disease in Japanese. Y. Asanomi.
- 2004F** Novel candidate AD-risk loci identified through whole exome sequencing in African Americans. M.M. Carrasquillo.
- 2007F** Complement receptor 1 (*CR1*) intragenic duplication and Alzheimer's disease. E.J. Hollox.
- 2010F** Alzheimer's disease exome sequencing study in the Finnish population isolate. M.I. Kurki.
- 2013F** Ambidexterity and Alzheimer's disease risk. E.E. Mlynarski.
- 2016F** Single variant and polygenic score analysis of whole exome data from the Alzheimer's Disease Sequencing Project (ADSP). J.S. Reddy.

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- 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.
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- 2049F** Genome- and phenome-wide association analyses uncover MET as a susceptibility gene of cerebral palsy. J.J. Connolly.
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- 2055F** Somatic copy number gains of alpha-synuclein (*SNCA*) in synucleinopathies (Parkinson's disease and multiple system atrophy). C. Proukakis.
- 2058F** Genome-wide CNV analysis identifies candidate causal genes for Parkinson's disease in the lysosomal pathway. P.S. Eis.
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- 2064F** Common variants at 5q33.1 predispose to migraine in African-American children. X. Chang.
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- 2070F** RNA-sequencing reveals novel immunological pathways in neuromyelitis optica. I. Adrianto.
- 2073F** Selective activation of caspase family of genes in multiple sclerosis patients inducing neuronal apoptosis. Y. Kattimani.
- 2076F** Genetic investigation of restricted and repetitive traits in autism. M.L. Cuccaro.
- 2079F** CNV meta-analysis of major neurodevelopmental and neuropsychiatric disorders. J.T. Glessner.
- 2082F** Gene expression profiling predicts clinical outcome in autism spectrum disorder: Confirmation of potential biomarkers and initial characterization of clinically homogeneous subgroups. F. Macciardi.
- 2085F** Epigenetic factors and gene-environment interactions in autism: Prenatal maternal stress and the SERT gene. Z. Talebizadeh.
- 2088F** Chronic psychosocial stress in mice alters brain myelination in a genetic background-dependent manner. I. Hovatta.
- 2091F** Whole-genome sequencing to identify risk loci for nighttime eating in American Indians. C. Koroglu.
- 2094F** Network analysis of gene polymorphisms in GABA, dopamine, cannabinoid, mu-opioid and alcohol metabolism pathways with alcohol dependence in scheduled class (SC) population of Punjab (Northwest India). R. Sharma.
- 2097F** A multi-omics analysis towards understanding of the polygenicity in schizophrenia. P. Jia.
- 2100F ‡** Using genetic diversity from East Asia to improve the biological insight into schizophrenia. H. Huang.
- 2103F** Copy number variation analysis of psychiatric traits in a large population-based sample of youth. M. Zarrei.
- 2106F** Neurodevelopmental copy number variants and clinical risk: A pediatric record population study. K. Ahn.
- 2109F** Maternal antenatal depression and child socio-emotional outcomes: Investigating intervening child genetic risk for ADHD and biological pathways. L.M. Chen.
- 2112F** Limited contribution of rare, noncoding variation to autism spectrum disorder from sequencing of 2,076 genomes in quartet families. D.M. Werling.
- 2115F** Parent-of-origin and maternal effects in attention deficit hyperactivity disorder. D. Smajlagic.
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- 2121F ‡** Common genetic variation contributes to risk of severe developmental disorders. M. Niemi.
- 2124F** Association of *HTR3C*, *HTR3D*, *HTR5A*, and *HTR6* gene polymorphisms with treatment response to risperidone in Chinese schizophrenia patients. S. Qin.
- 2127F ‡** Major depressive disorder and nausea and vomiting during pregnancy. Shared genetic factors? L. Colodro Conde.
- 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.
- 2133F** The transcriptional consequences of bipolar disorder polygenic risk and medication use. C.E. Krebs.

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- 2136F** Increased predicted *C4A* expression is associated with cognitive deficit in both schizophrenia and Alzheimer's disease. N.S. McCarthy.
- 2139F** Enrichment of treatable metabolic disease gene variants in a large cohort of schizophrenia, bipolar and major depressive disorder patients. V. Sreiretnakumar.
- 2142F** Identifying a shared regulatory background for neurodevelopmental disorders through meta-analysis of genomewide association studies. Z. Yang.
- 2145F** Gene-based meta-analysis of GWAS in African American and European ancestry populations implicates novel genes for PTSD. H. Zhang.
- 2148F** Catalogue of 1 billion candidate ultra rare variants across 11670 Han Chinese individuals. S. Mangul.
- 2151F** Analysis of leukocyte telomere length in children and adolescents at risk of developing mental disorders. G. Xavier.
- 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.
- 2157F** The intersection of type 2 diabetes and cognitive impairment in Mexican Americans: Insights from the mitochondria. T. Silzer.
- 2160F** A hypertension-associated mtDNA mutation alters the tertiary interaction and function of tRNA^{Leu(UUR)}. M. Guan.
- 2163F** Association of PMEL missense variants with ocular pigment dispersion and pigmentary glaucoma. M.A. Walter.
- 2166F** Identifying genes that underlie eye disorders and vision loss using predicted gene expression. J.B. Hirbo.
- 2169F** Identification of rare sequence variants in genes involved in focal adhesion and Wnt signaling pathways in keratoconus human corneas. J.A. Karolak.
- 2172F** A transethnic genome-wide association study identifies five novel genetic loci associated with primary open angle glaucoma. E. Jorgenson.
- 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.
- 2178F** Common variants in *KLHL2* and *C4orf50* are associated with poorer anti-VEGF treatment response in age-related macular degeneration. O. Garcia Rodriguez.
- 2181F** Using GWAS data to identify copy number variants associated with orofacial clefts. A.L. Petrin.
- 2184F** Clinical aspects associated with orofacial clefts in patients of smile operation in a Colombian population. J. Martinez.
- 2187F** Ballooning of redundant myelin sheaths in DS may resolve with Ca²⁺ as seen in the mouse. A.N. van Hoek.
- 2190F** ‡ CRISPR/Cas9 engineering to generate an isogenic model of the 3-Mb 22q11.2 syndromic deletion. Y.T. Lin.
- 2193F** Syndromic cleft genes implicated in non syndromic forms: Towards translational phenotypes? B. Demeer.
- 2196F** African haplotypic background mitigates the effect of *APOE* ε4 risk allele in Alzheimer disease. F. Rajabli.
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- 2202F** Meta-analysis of GWAS elucidates genetic architecture of dental caries. D. Shungin.
- 2205F** The PhenX Toolkit: Adding a resource for geriatric research. M. Phillips.
- 2208F** GWAS replicates known asthma variants validating self-reported childhood asthma diagnosis in the COPDGene Study. L.P. Hayden.
- 2211F** A simulated evaluation of data-driven algorithms for addressing clinical heterogeneity in complex traits. A.O. Basile.
- 2214F** Pathogenic and likely pathogenic mutations identified in apparently normal individuals of Arab descent. A. Alkhateeb.
- 2217F** Leveraging tissue specific omics data to estimate the disease/traits-related tissues. R. Chen.
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- 2223F** GWAS of early childhood caries in an Appalachian population. E. Orlova.
- 2226F** Genomic features of loci associated with multiple complex traits in genome-wide association studies. Y.-F. Wang.
- 2229F** Genetic polymorphisms of *LIN28B* and *MKRN3* in association with precocious puberty. K. Lee.
- 2232F** Whole exome sequencing analysis in severe chronic obstructive pulmonary disease. D. Qiao.
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- 2256F** Defining the opportunity: The IGNITE CPIC Prescribing Study. L. Wiley.

- 2259F** Integration of a tool for patient self-assessment within primary care to enable precision prevention. K. Rageth.
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- 2265F** The Precision Medicine Initiative All of Us Research Program: Innovative access to unprecedented data. A. Ramirez.
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- 2280F** ‡ Transferability of polygenic risk prediction across diverse and admixed populations. A.R. Martin.

Evolution and Population Genetics

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- 2289F** Improving genotype imputation in population isolates using identity by descent. M. Abney.
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- 2301F** Haplotype map of Russian population. I.V. Evsyukov.
- 2304F** Tracing maternal lineage of Austronesian-speaking Melanesians and Micronesians in the Solomon Islands. M. Isshiki.
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- 2310F** ‡ Discovering rare variants and deciphering a population structure of 386 Mongolian individuals by whole-genome sequencing. C. Kim.
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- 2316F** Selection analysis in Chileans identify adaptation signals in Native Americans, highlighting regulatory processes. L. Vicuña.
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- 2367F** Characterizing heterogeneity within fine-scale population structure. A. Hippen Anderson.

Molecular and Cytogenetic Diagnostics

- 2370F** Assessment of BAP1 germline and somatic alterations in uveal melanoma. M.H. Abdel-Rahman.
- 2373F** Retrospective analysis of 36 fusion genes in 6170 patients of *de novo* acute leukemia and myeloid neoplasms. X. Chen.
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- 2403F** Screening for rearrangements in *RB1* gene/ 13q14 through real-time PCR. R.M. Freitas.
- 2406F** Hyb & Seq™: The next generation of simultaneous RNA and DNA detection in liquid biopsy without the library and sample preparation bottlenecks. J. Beechem.
- 2409F** What's in a VUS rate? Simulated VUS calculations for hereditary cancer genes in a general population using population frequency data and ClinVar submissions. K.E. Kaseniit.
- 2412F** Loss-of-function *POLE* and *POLD1* variants may not be associated with early-onset colon cancer/polypsis. E.K. Flynn.
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- 2490F** Genetic causes of intellectual disability in 102 consanguineous families from Jordan. T. Froukh.
- 2493F** Copy number reanalysis: The hidden contribution of *MED13L* to intellectual disability. L.K. Conlin.
- 2496F** The clinical application of chromosomal microarray in the diagnosis of children with developmental delay/intellectual disability in Korea: A single tertiary center experience. Y. Kim.
- 2499F** Targeted next-generation sequencing of 75 genes in Japanese patients with intellectual disability and multiple congenital anomalies of unknown etiology. D.T. Uehara.
- 2502F** Genetic spectrum of limb-girdle muscular dystrophy in Taiwan. Y.L. Lin.

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- 2562F** The discussion of whether in vitro fertilization (IVF) or pre-implantation genetic diagnosis (PGD) in inv(9) carriers. H. Chen.
- 2565F** Developing frameworks to evaluate diagnostic genomic testing strategies for rare disease and cancer. R. Scott.
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- 2580F** An open-source quality control monitoring system for clinical NGS. N.R. Tawari.
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- 2589F** A clinical molecular genetics laboratory experience with whole exome sequencing. J. Machado.
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- 2598F** CLIP-Cap: Combined Long-Insert Paired-End and Capture sequencing, a novel method for the analysis of complex genomic aberrations. C. Purmann.
- 2601F** Undiagnosed Diseases Network (UDN) successes in precision medicine. J. Phillips.

Cardiovascular Phenotypes

- 2604F** Loss of *ADAMTS3* activity causes Hennekam lymphangiectasia-lymphedema syndrome 3. P. Brouillard.
- 2607F** *MCTP2* gene change detected by whole exome sequencing in an infant with endocardial fibroelastosis syndrome who underwent heart transplantation. D. Ercelen.
- 2610F** Toward genetics-driven early intervention in dilated cardiomyopathy: The DCM Precision Medicine Study. D.D. Kinnamon.
- 2613F** A report of a patient with hypertrophic cardiomyopathy without myopathy associated with *FLNC* heterozygous pathogenic variant and review of literature. B. Monteleone.
- 2616F** Germline loss-of-function mutations in *EPHB4* cause a second form of capillary malformation–arteriovenous malformation (CM-AVM2) deregulating *RAS*-*MAPK* signaling. M. Viskula.

- 2619F Mutation spectrum of Long QT Syndrome in Singapore.** R.Y.Y. Yong.
- 2622F *EIF2AK4* mutations are not likely a common genetic modifier of disease in BMPR2 mutation positive pulmonary arterial hypertension patients.** K. Sumner.
- 2625F Whole-exome sequencing identified a *de novo* *PDE3A* mutation causing autosomal dominant hypertension with brachydactyly.** D. Wang.
- 2628F Exome sequencing in children with pulmonary arterial hypertension demonstrates a different genetic architecture of disease compared to adults.** C.L. Welch.
- 2631F Exome sequencing of 103 Williams syndrome cases rules out variation in the remaining elastin allele as a major contributor to variance in blood pressure and arterial stenosis.** P.C.R. Parrish.
- 2634F Probing for modifiers of X-inactivation in a cohort of Amish families with hemophilia B.** A. Ozel.
- 2637F The association of *TMPO* and *RYR1* genes with cardiovascular diseases in a Turkish Cypriot Family.** M.C. Ergoren.
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- 2646F Identification of rare variation influencing CVD risk in Mexican Americans.** J.E. Curran.
- 2649F Associations of circulating protein levels with lipid fractions in the general population.** S.M. Figarska.
- 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.
- 2655F Gender specific modification of heart failure with preserved ejection fraction risk by mitochondrial haplogroups.** R.T. Levinson.
- 2658F Identifying new therapeutical targets for congestive heart failure.** A. Moreira.
- 2661F Elevated genetic risk for coronary artery disease increases hospitalization burden and mortality.** M. Sjögren.
- 2664F Evaluating the burden of pathogenic variants for the inherited arrhythmia syndromes.** Y.P. Fu.
- 2667F Exome sequencing identifies multiple variants associated with glyca, a novel biomarker of cardiovascular events.** L.C. Kwee.
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- 2673F Sequence data processing and analysis of 70,000 human genomes in the NHLBI TOPMed sequencing program.** T. Blackwell.
- 2676F Updated genome-wide association study and functional annotation reveals new risk loci for mitral valve prolapse.** N. Bouattia-Naji.
- 2679F Genome-wide association study of transposition of the great arteries.** D. Skoric-Milosavljevic.
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- 2685F ‡ Genome-wide association study reveals novel genetic markers associated with chlorthalidone blood pressure response.** S. Singh.
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- 2697F Genome-wide trans-ethnic meta-analysis for a novel sleep apnea endophenotype.** H. Wang.
- 2700F Cross-exposure multivariate interaction tests.** J. Kim.
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- 2709F “Genotype-phenotype in Marfan syndrome patients with causative mutations in the calcium binding region of the 43 cEGF-like domains in fibrillin-1 gene (*FBN1*)”.** J.A. Aragon-Martin.
- 2712F New genetic variants unveiled using a predicted-VAT mass phenotype.** T. Karlsson.
- 2715F Incorporating multiple sources of biological knowledge into association analysis of whole genome sequencing data identifies novel trait-associated rare variants.** Y. Ma.
- 2718F Geographic distribution of polygenic risk of complex traits and diseases in Finland.** S. Kerminen.
- 2721F Association of *SELP* variants and soluble P-selectin levels with type 2 diabetes mellitus: A case-control study.** R. Kaur.
- 2724F Origins and dynamics of the Brazilian population and sickle cell mutations reveal unexpected diversity.** Y. Guo.
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2745F Gender transcriptome signatures for congenital heart defects (CHD) children based on next generation sequencing technologies of cardiac tissues. C. Kim.

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2760F Isoforms and eQTLs of the myocardial infarction gene *PHACTR1*. V. Codina-Fauteux.

Statistical Genetics and Genetic Epidemiology

2763F Type 1 diabetes progression is correlated with changes in the co-expression relationships of immune response genes. I. Braenne.

2766F ‡ Smoking-by-genotype interaction in type 2 diabetes. P. Wu.

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.

2772F Long-term response to oral eliglustat in treatment-naïve adults with Gaucher disease type 1: Final efficacy and safety results from a phase 2 clinical trial after 8 years of treatment. H. Lau.

2775F Obesity is a systemic regulatory outcome and mainly controlled by several tissues. R. Hao.

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.

2781F Effects of the interaction between a *CREBRF* missense variant and body mass index on type 2 diabetes risk in Samoans. E.M. Russell.

2784F ‡ Trans-ethnic meta-analysis of rare variants in sequencing association studies. J. Shi.

2787F PLEIOVAR, testing for association between multiple traits and multiple variants. O. Meirelles.

2790F Examining the causal relationship between vitamin D and serum metabolic measures: A Mendelian randomization study. T. Dudding.

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

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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.

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2805F ‡ Statistical framework for biological interpretation and improvement of genetic association studies. M. Artomov.

2808F Relationship of genetic and clinical factors and prevalence of CKD in a Japanese population: J-MICC Study. R. Fujii.

2811F Integrated pediatric bone density phenotypes and genetic regulation of the developing skeleton. J.A. Mitchell.

2814F ‡ Tissue-specific genetic regulated expression in late-onset Alzheimer's disease: The Alzheimer's Disease Genetics Consortium (ADGC). H.-H. Chen.

2817F Genome-wide search for genetic loci perturbing gene co-expression networks in Alzheimer's disease. L. He.

2820F Mitochondrial variants associated with increased risk of late-onset Alzheimer's disease. T.J. Zhou.

2823F Small posterior fossa in Chiari malformation affected families is significantly linked to 1q43-44 and 12q23. A. Musolf.

2826F Low-rank structure based brain connectivity GWAS study. Z. Zhu.

2829F ‡ Comparison of methods for multivariate gene-based association analysis using common variants for complex disease. J. Chung.

2832F *SORBS2* is associated with extended Alzheimer disease related phenotypes in *PSEN1* mutation carriers in Puerto Rico. R. Cheng.

2835F Diagnostic changes leading to ASDs' prevalence increase altered the disorders' average genetic architecture. E.M. Wigdor.

2838F Improved prediction of genetic predisposition to psychiatric disorders using genomic feature best linear unbiased prediction models. P.D. Rohde.

2841F A penalized parametric bootstrap approach for self-contained pathway analysis of gene-environment interaction. B.J. Coombes.

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- 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.
- 2847F** Detecting tissue-specific genetic correlation between complex psychological disorders using GWAS summaries. Q. Fan.
- 2850F** Gene interaction between DRD4 and DAT1 Loci is a ADHD-risk factor in females of Chilean ancestry. G. Pathak.
- 2853F ‡** Flipping GWAS on its head: A statistical approach to identify genetically distinct disease subphenotypes. A. Dahl.
- 2856F** Identifying highly damaging missense mutations in over 10,000 developmental disorder trios using a regional missense constraint metric. K.E. Samocha.
- 2859F** The investigation by WES of inborn errors of metabolism as an underlying cause of idiopathic intellectual disability and/or unspecified congenital malformations in a series of 550 patients. N. Houcinat.
- 2862F** Association study for common and rare genetic variation contributing to exfoliation syndrome. R.P. Igo.
- 2865F** Family based association tests of myopia reveal a potentially hidden association signal upstream of two GABA receptor genes. C.D. Middlebrooks.
- 2868F** Grouped association analysis for very rare variants using Fisher's Exact Test and external controls. A. Kwong.
- 2871F** Iterating from discovery to epidemiological consequence through disease mechanism. J. Brown.
- 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.
- 2877F** Genetics of the human microbiome and implications in obesity associated measures. C.T. Finnicum.
- 2880F** Admixture mapping of asthma in individuals of mixed African ancestry reveals a novel association on chromosome 6q23.2. M. Daya.
- 2883F** Model-based multiple variants test considering causal status. J. Joo.
- 2886F** Use low-depth and high-depth whole genome sequencing data to predict 36 blood groups. Y. Sun.
- 2889F** Family-based rare variant association study of familial myopia in Caucasian families. D. Lewis.
- 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.
- 2895F** Bayesian hierarchical modeling of genic sub-region intolerance. T.J. Hayeck.
- 2898F** Evidence for a major gene for myopia risk in Han Chinese-American families at 10q26. J.E. Bailey-Wilson.
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- 2910F** Population pharmacokinetics of sulindac and genetic polymorphisms of FMO3 and AOX1 in women with preterm labor. J. Yee.
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- 2916F** Comparison of power of summary based methods for identifying expression-trait associations. Y. Veturi.
- 2919F** Logolas: A tool for visualizing enrichment of genetic signature profiles. K. Dey.
- 2922F** Mapping genetic organization and disease liabilities of human cortical surface with summary statistics of vertexwise genome-wide association studies. C. Fan.
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- 2928F** Polymorphisms in the HSF2, LRRC6, MEIG1 and PTIP genes correlate with sperm motility. S. Rajender.
- 2931F** Simulating autosomal genotypes with realistic linkage disequilibrium and a spiked in genetic effect. M. Shi.
- 2934F ‡** A CREBRF missense mutation substantially affects height in Samoans. S.L. Rosenthal.
- 2937F** Pharmacological insights from genetic mapping of the plasma proteome. J.C. Maranhville.
- 2940F** Gene-based association testing of dichotomous traits with generalized linear mixed models using extended pedigrees. C. Chiu.
- 2943F** The SUPERBABY PROJECT: Genetic determinants of the favorable NICU course in premature newborns. K.M. Gnona.
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- 2949F** Total serum IgE whole genome sequence association analysis in families from Barbados. A. Shetty.
- 2952F** Multivariate generalized linear model for genetic pleiotropy. D.J. Schaid.
- 2955F** MEGA analysis of alcohol consumption in diverse populations: The Population Architecture using Genomics and Epidemiology (PAGE) Study. K.L. Young.
- 2958F** VikNGS: A C++ Variant Integration Kit for Next Generation Sequencing across research studies for robust rare and common variant association analysis. Z. Baskurt.
- 2961F** Estimating cell-type-specific DNA methylation effects in the presence of cellular heterogeneity. Y. Feng.
- 2964F** Fast permutation tests and related methods for association between rare variants and binary outcomes. A. Sondhi.

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- 2973F** Increasing the power of meta-analysis of genome-wide association studies to detect heterogeneous effects. C.H. Lee.
- 2976F** Testing for goodness rather than lack of fit of a X-Chromosomal SNP to the Hardy-Weinberg Model. S. Wellek.
- 2979F ‡** Using relationships inferred from electronic health records to conduct genetic studies. F. Polubriaginof.
- 2982F** Combining sequence data from multiple studies: Impact of analysis strategies on rare variant association results. Z. Chen.
- 2985F** Widespread pleiotropy confounds causal relationships between complex traits and diseases inferred from Mendelian randomization. M. Verbanck.
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- 3003F** A generalized permutation testing method for binary trait association in structured samples. J. Mbatchou.
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- 3009F** Investigating shrinkage methods to improve accuracy of GWAS and PRS effect size estimates. Y. Ruan.
- 3012F** Statistical and population genetics of extreme phenotypes. O. Soylemez.
- 3015F** Scalable Bayesian functional genome-wide association study method with summary statistics. J. Yang.
- 3018F** On simulation design for evaluating type 1 error: What is the 'correct' null model? T. Zhang.