Precision Medicine, Pharmacogenomics, and Genetic Therapies

372W ‡ A mechanism and treatment strategy for the sexual dimorphism seen in vascular Ehlers-Danlos syndrome. C.J. Bowen.

375W Nanog-ligated protein replacement therapy for Tgase-1-deficient congenital ichthyosis: Results from the proof of concept study. K.M. Eckl.


381W ‡ Genetic variants that associate with liver fibrosis and cirrhosis have pleiotropic effects on human traits. E. Speliotes.


387W An African ancestry uterine fibroids polygenic risk score (PRS) identifies associations with other gynecologic conditions in the clinical phenome. J.N. Hellwege.


393W Pharmacogenomics and opioid use patterns in patients with chronic spine pain. K. Fisch.

396W Fine mapping of human leukocyte antigen complex to study asthma in African Americans. H. Gui.

399W The beliefs and values of patients with hemophilia and their caregivers about gene therapy and gene editing. T. Vasquez.


408W A polygenic risk score to improve screening for fracture risk. J. Keller-Baruch.

411W Cell autonomous and cell non autonomous effects of NAGLU deficiency in Sanfilippo B syndrome. I. Esharkawi.


417W ‡ Utilizing gene expression to prioritize genes in medical patients. P.E. Evans.

420W Lessons from the TOTEM trial, a phase 1/2 multicentre, open-label, single-arm study of low-dose PI3K inhibitor taselisib in adult patients with PI3KCA-related overgrowth (PROS). M. Luu.


426W ‡ Zebrafish modeling for the clinic: Rapid in vivo functional testing of patient variants for clinical applications. B. Jussila.

429W PLP1 gene suppression therapy for Pelizaeus-Merzbacher disease using artificial miRNA. K. Inoue.

432W Long-term effect of the gene therapy using AAV vectors with the human intrinsic GLUT1 promoter for Glut1-deficient mice. S. Nakamura.

435W A comprehensive approach to developing biomarkers tracking the progression of Parkinson disease. J. Zhu.

438W Antisense oligonucleotide therapy in a humanized mouse model of MECP2 duplication syndrome. Y. Shao.

441W Translating behavioral epigenetics into the primary care setting. J.R. Murphy.

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447W Genetic polymorphisms in CYP2D6: Antipsychotic-induced weight gain in the UK Biobank sample. I. Austin-Zimmerman.

450W Pharmacological annotation of polygenic risk in individuals to direct precision intervention in complex disorders. M. Cairns.

453W The effect of startle-related genes on anxiety symptom severity at baseline and following pharmacological treatment. J. Tomasi.

456W Concurrent hearing, genetic and HCMV screening in a general newborn population. Z. Peng.

459W Rapid and ultra-rapid whole genome sequencing in acutely ill neonates: University of California Davis experience. S. Shankar.

462W First-line preventative genetic screening: Disease penetrance in Tier 1 inherited diseases in an all-comers population is similar to family history selected populations. J. Lu.


468W Host genome-wide association study of infant susceptibility to shigella-associated diarrhea. D. Duchen.


474W The design and implementation of a cohort-level pharmacogenomics program at UnitedHealth Group. D. Crosslin.

477W Analyses of core and extended pharmacogenes in sickle cell disease patients in Africa. K. Mnika.


483W Precision medicine 2.0: Integrate precision medicine into clinical practice. Z. Gu.

489W Multi-ethnic single nucleotide polymorphism (SNP)-based screening of pharmacogenomic HLA alleles implicated in drug-induced hypersensitivity reaction. P. Nicoletti.

492W Homology aware design facilitates microarray CNV detection within gene families. K. Aull.

495W Family history vs. polygenic risk scores as the predictors of genetic risk. R. Mägi.


504W Integrating longitudinal omics for detection of adverse events in deep space missions. G. Mias.

Prenatal, Perinatal, Reproductive, and Developmental Genetics

507W Targeted sequencing and RNA assay reveal a noncanonical-JAG1 slicing variant causing Alagille syndrome. Y.Y. Chen.

510W Analysis of ultrasonic manifestation of fetus with 17q12 microdeletion. L. Zhang.

513W The role of TWIST1 phosphorylation and miR10 in regulating neural crest cell fate and migration. W.D. Fakhouri.

516W Involvement of transcription factors that control embryonic morphogenesis in the repair of DNA-DSBs after fetus radiation exposure. A. Noda.

519W ‡ In utero endotoxin exposure decreases subsequent inflammatory bowel disease severity in mice. E. Banfield.

522W When one tissue is not enough: Child with developmental delays and mosaic marker chromosome duplicating 11p12-11p11.2, present in amniocytes and buccal cells but not in blood cells. C. Melver.

525W Placental epigenetic and gene expression changes associated with maternal dyslipidemia in early pregnancy. M. Ouidir.

528W The landscape of variations of embryonic mitochondrial genome. S. Madjunkova.

531W Cytogenetic abnormalities found in 675 Mexican patients with diagnosis associated with abnormal sexual development. C. Alonso Muñoz.

534W Sleep disturbances in school age children with PTHS, SMS, and MAND mirror sleep abnormalities in autism spectrum disorder. A. Gandhi.

537W The mortality and morbidity of very low birth weight infants with trisomy 21 in Japan. H. Kawasaki.

540W Placental transcriptome analysis using RNA sequencing in monochorionic twin pregnancies complicated by selective growth restriction. T. Leung.


546W Exome sequencing enhances the diagnostic rate of perinatal autopsy: A prospective multicentre clinical utility trial with implications for perinatal diagnosis. A. Vasudevan.

549W An infant with ambiguous genitalia and complex mosaicism for Y-chromosome abnormalities not detected by prenatal cell-free fetal DNA testing. W. Wilson.

552W Likely pathogenic de novo variants in congenital diaphragmatic hernia patients are associated with clinical outcomes. L. Qiao.


558W Associated anomalies in cases with agenesis of the corpus callosum. C. Stoll.

561W Investigating evolving interactions between Zika virus and host pathways using Drosophila. J. Hamish.

564W Evidence of pervasive anxiety-related symptoms in 47,XXY (Klinefelter syndrome) and the potential impact of hormonal replacement therapy (HRT). C. Samango-Sprouse.

567W Insights of the role of FOXC1 gene in ocular development using a double foxc1a/foxc1b knockout zebrafish line. J.J. Ferre Fernandez.

570W Mathematical modeling reveals non-independence of meiosis I and meiosis II errors during human oogenesis. J. Xing.


576W Expanded carrier screening using next generation sequencing: An experience from India. S. Sharda.

579W It’s not all about the extra X: The potential role of copy number variants (CNVs) in the neurodevelopmental variability of 47,XXY (Klinefelter syndrome). S. Tran.

582W Exome sequencing expands knowledge of early phenotypes in well-known genetic conditions. S. Rego.

585W Utility of whole genome sequencing in an undiagnosed fetus with increased nuchal translucency: A case illustration. M.M. Shi.

588W Genetic evidence on the role of maternal vitamin D status during pregnancy in obstetric outcomes. M.C. Borges.

591W Cell-free DNA analysis in twin pregnancies after reduction because of fetal aneuploidy. K.M. Chen.

594W Combined preimplantation genetic testing for CFTR gene mutations and aneuploidy using a single WGA PCR and ampiclon panel-specific enrichment. M. Jasper.

597W The application of ACMG variant classification guidelines for the variant assessment of non-invasive prenatal sequencing (NIPS). L. Lu.

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855W Cancer care and a tale of three molecular "genomic" tests. J. Wallace.

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867W Transcriptome-wide association study of anthracycline-induced cardiotoxicity in pediatric cancer patients. E.N. Scott.

870W Intelligent biomarker selection and individualized therapy. Q. Ge.

873W Effect of cisplatin on MAPK pathway gene expression is reversed by naringenin in HCT116 colorectal cancer cell lines. A.G. Thornburg.

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879W Cancer gene mutations in congenital pulmonary airway malformation patients. J. Hsu.


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891W Associations of BRCA1/2 mutations with Chinese breast/ovarian cancer patients. M. He.

894W SVFX: A machine learning framework to identify pathogenic structural variations in disease studies. S. Kumar.

897W Population-wide screening for germ line variants of hereditary cancer genes in 12K unselected colorectal cancer and 27K controls in Japan. H. Nakagawa.

900W Partitioned heritability and functional enrichment reveal ovarian cancer risk variants in histotype-specific enhancers that disrupt transcription factor binding sites. P.-C. Peng.

903W CRISPR-Cas9 generated allelic series of rat mutations confirms Tox3 as a breast cancer susceptibility gene. L. Shunkwiler.

906W Characterization of 331g/a polymorphism of rp gene and identification of viral oncogene hmtv virus as genetic markers for the improvement of breast cancer management in CHU Yaounde Cameroon. N. Nguedia Kaze.

909W Candidate gene polymorphism study in South Indian men with prostate cancer. S. Daram.

912W Evaluation of copy-number variants as modifiers of breast and ovarian cancer risk for BRCA1 and BRCA2 pathogenic variant carriers. C. Hakkaart.

915W Distinct microbial communities that differ by race, stage, or breast-tumor subtype in breast tissues of non-Hispanic black and non-Hispanic white women. A. Stariad-Davenport.

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924W Dissecting the chromatin landscape of rela-fusion driven ependymoma. S.C. Mack.

927W A pan-cancer approach to predict core transcriptional regulatory circuits identifies important nodes in high-grade serous ovarian cancer. K. Lawrenson.

930W Enhancer-promoter chromatin interactions in endometrial cells reveals biologically relevant target genes at endometrial cancer risk regions. T. O’Mara.

933W Single cell transcriptomic and epigenetic characterization of inter and intra tumor heterogeneity in Glioblastoma. A.M. Raman.

936W Prdm14 transcriptional output is a determinant of testicular germ cell risk. N.J. Webster.

939W Chromatin folding domain disruptions by somatic genomic rearrangements in human cancers. K. Akdemir.

942W Competing endogenous RNA network signature in glioblastoma. E. Toraih.

945W A two-stage epigenome wide association study identifies novel pancreatic cancer susceptibility loci by leveraging public controls. Z. Wang.

948W Epigenetic regulation of the mir-137 Host Gene Cpg island that spans mir-137 and adjacent downstream sequence is associated with head and neck cancer. H. Celesnik.

951W Genetically predicted methylation biomarkers and pancreatic cancer risk: A comprehensive study using genetic instruments. L. Wu.

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966W Ethno-geographic germline protein-altering mutations in ATM predispose to lung adenocarcinoma. X. Ji.
969W Validation of previously identified breast cancer SNPs in Black women. J. Moore.

972W Cervical somatic mutations detected in HPV18 and HPV45 precancerous lesions are related to disease progression. M. Pinheiro.

975W Exploring the causal role of the human gut microbiome on colorectal cancer: Application of Mendelian randomization. K.H. Wade.

978W The first study evaluating the distribution of gBRCA1/2 mutations within the ovarian cancer cluster region in ovarian cancer patients using Japan CHARLOTTE study data (Characterizing the cross-sectional approach to ovarian cancer: Genetic testing of BRCA). K. Yoshihara.

981W Characterization of BRCA1 and BRCA2 genetic variants in Guatemala population with breast cancer. C. Carranza.


987W Highly aggregated lung cancer families show significant linkage to chromosome 12q23.3 for cancer risk. A. Musolf.

990W Leveraging tumor characteristics to predict germline variant pathogenicity in mismatch repair genes. S. Li.

993W Characterization of candidate functional variants and susceptibility genes from a melanoma and multi-cancer risk locus at chr2q33.1 near CASP8. H. Kong.

996W An overview of causal associations between risk factors' genetic signals and breast cancer survival. M. Escala Garcia.

999W An empirical comparison of approaches to discovering novel cancer predisposition genes by rare variant analysis using public available summary counts or full genotype data. W. Chen.


1008W Comprehensive assessment of gene fusion detection, prioritization, annotation, and characterization. V. Chakraborty.

1011W Representing cancer-relevant variants in variation graphs. E.T. Dawson.

1014W Imputation of the GSTM1 deletion in the 1000 Genomes Project populations to develop a reference dataset for epidemiological studies. O. Florez-Vargas.

1017W The Reactome Pathway Knowledgebase and ReactomeFIViz app. R. Haw.


1023W QC measurements of exome chip sequence data in a family-based study. Q. Li.

1026W Allelic-imbalance in DNA inferred from RNA-seq data using a haplotype-based method. Z. Ozcan.


1032W Reducing false positive somatic variant calls arising from breakpoint-associated alignment artifacts. Q. Zeng.

1035W ‡ Transcriptomic deconvolution reveals cell-type specific signals across cancer types. S. Cao.


1041W ‡ Oncogene.iobio: A web app for real-time, integrative examination and functional prioritization of tumor mutations. S.J. Georges.

1044W A powerful method to estimate cell-type specific QTLs from bulk expression by leveraging allelic imbalance. C. Kalita.

1047W Pan cancer genomic characterization of patient-matched primary and brain metastases. O. Lee.

1050W A flexible, high-throughput software pipeline for identifying candidate variants and their effects on protein structure, starting from NGS data or VCF files. K. Maxfield.

1053W ‡ Associations between cancer and non-cancer-related Mendelian disease genes and specific cancer types in TCGA. B. Salisbury.

1056W Mutational signatures and clinicopathological relationship extracted from clinical sequence data of 201 cases with colorectal cancer. S. Takeuchi.


1065W Identification of a novel group of endometrial cancer patients with loss of thyroid hormone receptor expression and improved survival. D. Piqué.

1068W Comprehensive structural and copy-number variant detection with long reads. A.M. Wenger.

1071W Computational strength of evidence for clinical sequence variant classification without circular reasoning. S.V. Tavtigian.

1074W Targeting noncoding RNAs can improve the effectiveness of radiation therapy for cancer patients. H.S. Chiu.

1077W Inference and characterization of copy number variants from bulk and single cell RNA-sequencing datasets. A. Harmanci.

1080W Sparse conditional generate adversarial networks (CGAN) for individualized biomarker selection and treatment effect estimation. S. Fang.

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Mendelian Phenotypes

1083W Clinical, biochemical, and pathologlcal investigatiion of a patient with musculocontractual Ehlers-Danlos syndrome caused by a novel pathogenic variant in DSE. M. Minatogawa.

1086W Probing a novel familial lipomatosis syndrome with whole genome sequencing. J. de Vegvar.

1089W Detection of a novel large deletion in FBN1 gene by MLPA. J. Bene.


1095W Biallelic KRT5 mutations in epidermolysis bullosa simplex, including a complete human keratin 5 “knock-out”. H. Vahidnezhad.

1098W‡ A genetic retrospective study of maturity-onset diabetes of the young (MODY) in two population health studies. A. Bolze.


1104W Novel KMT2D mutation in Kabuki syndrome with midgut malrotation and hyperinsulinism. T. Phetthong.

1107W Expanding the phenotype of inflammatory bowel disease in Hermansky-Pudlak syndrome. B.R. Gochuico.

1110W Exome sequencing in biliary atresia family trios: Investigating ciliopathy genes. A. Sabo.

1113W‡ Overcoming challenges in identification, annotation, and interpretation of variants in DSD genes. E.C. Delot.


1119W X-linked hypophosphatemia: All eight individuals representing separate American families carrying the PHEX 3’UTR mutation c.231A>G tested positive for an exon 13-15 duplication. S. Mumm.

1122W GNPNAT1 variant causes a novel form of rhizomelic skeletal dysplasia due to impaired chondrocyte differentiation. S. Naz.

1125W MPS-specific physical symptom score (PSS) and adaptive functions in MPS IVA: A cross sectional study. A. Ahmed.

1128W Anabolic effects of nitric oxide on bone revealed by deficiency of arginosuccinate lyase. Z. Jin.

1131W‡ Respiratory function in the CrtapKO mouse model of osteogenesis imperfecta. R. Morello.


1137W Variant in OTUD6B gene cause intellectual developmental disorder with dysmorphic facies, seizures, and distal limb anomalies: A case report. J. Rojas Martinez.

1140W Expanding the spectrum of KLHL7, novel mutation, and evidence for multilocus pathogenic variation. A. Gezdirici.


1149W Severe Noonan syndrome phenotype associated with a germline Q71R MRAS variant. H. Suzuki.

1152W Updates to the NIAID Genomic Research Integration System (GRIS): Integrated phenotype-genotype search, laboratory test results integration and visualization, variant tagging, and automating clinical reporting workflows. S. Xirasagar.

1155W A single center experience in Turkey in the molecular diagnosis of hemophilia B. H. Onay.

1158W Novel ELANE gene mutation in a Vietnamese boy with severe congenital neutropenia. C. Bui.

1161W Biallelic loss-of-function JAK1 variants detected by whole exome sequencing in a patient with immunodeficiency and multiple congenital anomalies. J. Ji.

1164W De novo loss-of-function variants in CAPRIN1 in patients affected by severe autism and language delay. A. Brusco.

1167W Haploinsufficiency of SPTBN1 is associated with syndromic intellectual disability and autism. J. Posey.

1170W Biallelic variants in IQSEC1 cause intellectual disability, developmental delay, and short stature. S.A. Paracha.

1173W‡ Complex genetic network underlying the convergent of Rett Syndrome like (RTT-L) phenotype in neurodevelopmental disorders. S. Rangasamy.

1176W In vivo role of FMRP phosphorylation in fragile X syndrome. Y. Wen.

1179W Expanding the AUTS2-associated syndrome: Novel variants in AUTS2 are associated with a severe neurodevelopmental disorder that overlaps with Rubenstein-Taybi syndrome. K. Aldinger.

1182W Neuropsychological study in 14 French patients with White-Sutton syndrome and POG2 mutations. A. Garde.

1185W‡ Truncated AHDC1 protein induces a nucleolar stress response in Xia-Gibbs syndrome. M.M. Khayat.

1188W Mitochondrial-processing peptidase subunit beta (PMPCB) rare compound heterozygous variants in a patient with developmental delay, neurodegeneration, cerebellar vermis agenesis and cerebellar atrophy. K. Belhassan.

1191W Long-term survival of mice with deletion of BRCA1 as adults. J.O. Olayiwola.

1194W Metabolomics analysis of SLC13A5 deficiency in mice. A.C. Santos de Medeiros.


1200W Collaborative development of therapeutics for sialic acid storage disease. M.E. Hack Barth.

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1209W Novel mutation in \textit{MT-ND1} m.3955G>A related to neonatal onset Leigh syndrome with spinal lesion. T. Kumaki.

1212W Genetic diagnosis has been provided for 260 patients with inherited metabolic diseases positively screened by newborn screening in Japan since 2014. T. Fukao.

1215W The G6PD-mediated mechanism of inhibiting the invasion and proliferation of \textit{plasmodium falciparum} by dehydroepiandrostosterone and nicotinamide. W. Jiang.

1218W The CHD4-related syndrome: Investigation of the clinical spectrum, genotype-phenotype correlations and molecular basis. K. Weiss.

1221W Co-occurrence of two syndromes resulting in a mixed phenotype-Prader Willi syndrome and KBG syndrome. P. Gupta.

1224W Inhibition of G-protein signaling in cardiac dysfunction of Intellectual Developmental Disorder with Cardiac Arrhythmia (ID-DCA) syndrome. P. De Nittis.

1227W Expanding the spectrum of novel \textit{ARID1B} mutations in Coffin-Siris syndrome. E... Tan.

1230W Genome sequencing analysis of a family with a child displaying severe abdominal distention and recurrent hypoglycemia. J.L. Liu.

1233W \textdagger{} Linked-read whole genome sequencing identifies biallelic mutations in \textit{DONSON} as a novel cause of Meier-Gorlin syndrome. L.S. Bicknell.

1236W Functional characterization of lymphatic anomaly-associated, ERK-activating mutations in primary lymphatic endothelial cells. M.E. March.

1239W An integrated genomic medicine program for Mendelian disease gene discovery in Qatar. W. Aamer.

1242W A homozygous \textit{TANGO2} deletion identified in a Brazilian patient. Y. Lei.

1245W Familial cases of DeSanto Shinawi syndrome with previously unreported nonsense and missense variants in \textit{WAC}. M. Shinawi.

1248W Clinical exome sequencing data reveals high diagnostic rates and new susceptibility genes for congenital diaphragmatic hernia plus (CDH+). T.M. Scott.

1251W Biallelic novel variants in \textit{SPEG} causing arthrogryposis and neuromuscular phenotype. J. Fatih.

1254W Compound heterozygous \textit{TRAPPC1} mutations in a Chinese patient with limb girdle muscle dystrophy-18. C. Bharatendu.

1257W Clinical and neuropathological features of four familial and two sporadic cases of Neuronal Intranuclear Inclusion Disease (NIID). H. Furuya.

1260W A coexisting case of Eaton Lambert disease and Charcot Marie Tooth Type 20 syndrome. L. Mora Barreto.

1263W \textdagger{} Autosomal recessive infection-induced encephalopathy due to biallelic pathogenic variants in \textit{NUP214}. C. Applegate.

1266W Mutations in \textit{PIGB} cause an inherited GPI biosynthesis defect. B. Lace.

1269W Novel mutations of \textit{COL4A1} in four Japanese patients with variable CNS involvements. N. Nishimura.


1278W \textit{DYNC1H1}-related disease: Reviewing the literature, describing three novel de novo variants, and expanding the phenotypic spectrum. L. Jeffries.

1281W Clinical and molecular spectrum of \textit{WWOX}-related epileptic encephalopathy (WOREE). Y. Cao.

1284W \textdagger{} After the clinical exome: Identifying the genetic etiology of refractory pediatric epilepsies. J.D. Calhoun.

1287W \textit{De novo} \textit{ZBTB47} missense variants in two individuals with generalized epilepsy, developmental delays, and movement abnormalities. J.A. Rosenfeld.

1290W Identification of novel variants causing leukodystrophies in Sudan. M. Amin.


1296W Multi-omic analyses in a family with Gaucher and Gaucher-Parkinson diseases reveal several potential modifier genes in \textit{GBA1}-associated Parkinson disease. E. Garcia.

1299W Genetics of a novel neonatal neuromuscular disorder in dogs. K.J. Dillard.

1302W \textdagger{} Mutation in \textit{CADM3} cause upper limb predominant neuropathy with pyramidal signs. A.P. Rebeiro.


1308W Upstream missense variants approximately located in \textit{Hikeshi} gene cause hypomyelinating leukodystrophy. S. Ben-Shachar.

1311W \textdagger{} A mix of "old-school" and up-to-date familial genetic approaches led to identify a new genetic modifier of the age at onset in \textit{C9orf72} frontotemporal dementia. M. Barbier.


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1323W Exploring the molecular basis of hereditary spinocerebellar degeneration in a large Sudanese family. A. Babal.

1326W Mutational analysis of SOST in the first Italian family with sclerosteosis. M. Gagliardi.

1329W Biallelic pathogenic variants in TUBGCP2 cause microcephaly and lissencephaly spectrum disorders in humans. T. Mitani.

1332W Phenotypic similarities in one case with a microdeletion and two cases with single nucleotide variants in BCL11A: Cerebellar expressions of motor speech disorders. B. Peter.


1338W Brain region specific contribution of the ATXN1/CIC interaction to spinocerebellar ataxia type 1. S.L. Coffin.

1341W The C9orf72 repeat expansion, associated with ALS/FTD, is a rare folate-sensitive fragile site. M. Mirceta.

1344W Catatonia is a clinical feature of the natural progression of NLGN2-related autism spectrum disorder. A. Shillington.

1347W GJB2 and GJB6 mutations in non-syndromic childhood hearing impairment in Ghana. S.M. Adadey.

1350W Heimler syndrome: Hypomorphic mutations in the peroxisome-biogenesis genes PEX1 and PEX6 are in cause. I. Ratbi.

1353W A novel mutation in MERTK for rod-cone dystrophy in a North Indian family. S. Bhatia.

1356W Molecular genetic analysis of rare non-syndromic pre-lingual hearing disorders in a Romanian patient population. R. Birkenhager.


1362W Copy-number variation contributes 9% of pathogenicity in the inherited retinal degenerations. K.M. Bujakowska.

1365W Hidden genetic variations in PAX6-associated aniridia revealed by whole genome sequencing and machine learning based functional annotation. J. Han.


1371W Mouse models of glycopathology-related diseases: Identification of understudied genes. M. Tomczuk.


1377W Potential involvement of the novel gene ArfGAP2 in inherited retinal degeneration. R. Ayyagari.

1380W Evolution of the facial features in a patient with the rare Siderius type XLID syndrome caused by hemizygous loss of PHF8. O. Petryna.

1383W A new global Slc7a7 knockout mouse models human lysinuric protein intolerance. B.M. Stroup.


1389W Optical mapping of the schizophrenia-associated 3q29 deletion reveals new features of genomic architecture. T. Mosley.

1392W Pathogenic variants of EPHB4 cause lymphatic anomalies through over-activation of mTOR. M.R. Battig.


1398W Aromatic amino acid decarboxylase deficiency in 17 Mainland China patients: Clinical phenotype, molecular spectrum and therapy overview. D. Qian.

Bioinformatics and Computational Approaches


1404W In silico analysis of coding/non-coding SNPs of human RETN gene and characterization of their impact on resistin stability and structure. I. Morjane.

1407W Machine learning based histology phenotyping to investigate epidemiologic and genetic basis of adipocyte morphology and cardiometabolic traits. C.A. Glastonbury.

1410W A deep learning model for accurate variant calling in congenital adrenal hyperplasia. S. Hong.

1413W BD-CoCoLasso: Alternating block descent penalized regression adapted for the presence of a block of inaccurate features. C. Escribe.

1416W Accurate and rapid prediction of first-line tuberculosis drug resistance using machine learning method. X. Kuang.

1419W The investigation of cell separation-induced gene expression via a penalized deconvolution approach. A. Tai.


1428W Game theoretic centrality supports link between autism spectrum disorder and the HLA complex. M.W. Sun.

1431W Precision genotyping of APOE from whole genome sequencing. C. Zhu.

1434W Computational pipeline for RNA-seq based genetic diagnosis. M.F. Müller.
1437W Assessing and improving mitochondrial haplogroup assignments with low-coverage, genotype data. E.L. Drummond.

1440W Development and implementation of a computational workflow for the molecular diagnosis of constitutional syndromes using genome-wide DNA methylation analysis. E. Aref-Eshghi.


1446W CAGI SickKids challenges: Assessment of bioinformatic strategies for predicting patients’ phenotypes and identifying potential pathogenic variants and disease genes from their genomes. M. Meyn.

1449W Validation of a clinical germline exome sequencing assay and bioinformatics pipeline at UPMC Genome Center. R.O. Rosti.


1458W Integration of interactome perturbation and cell-type-specific transcriptome implicates novel autism risk genes and cell types where they are expressed. S. Chen.

1461W Systematic approach for drug repositioning of anti-epileptic drugs. Y... Ko.

1464W Integrated SV/structural variation analysis on the genome research platform GENESIS. M.C. Danzi.

1467W Non-synonymous co-variation is crucial for SUDEP in SCN1A driven epilepsy. P. Vandeventer.

1470W A new machine-learning based method to accurately assess copy number variants from whole genome sequencing data and its application on the analysis of the Hirschsprung disease genome. X.H. Zhuang.


1482W Shared genetic etiology underlying LOAD and MDD. J. Barrera.


1488W Automated processing of phenotypic data submissions. R. Mayani.


1494W Dissecting phenotypic variation in PTSD using genetically-regulated transcriptional variation. N.P. Daskalakis.


1500W New candidate Alu sequences in AK1 Korean reference genome. K. Bahk.

1503W Variant reclassification in a routine diagnostic setting on a highly heterogeneous patient population. P. Bauer.

1506W nanotatoR: An annotation tool for genomic structural variants. S. Bhattacharya.


1515W Decoding regulatory structures and features from epigenomics profiles: A Roadmap-ENCOD Variational Auto-Encoder (RE-VAE) model. R. Hu.

1518W Investigating the associations between epigenomic and metabolomic data using an integrative multivariate approach. V. Karhunen.

1521W BioTuring Browser: A modern data visualization and analytics platform for single-cell sequencing data. T. Le.

1524W Quality control for full genome sequencing with Sequila. A. Lesniewska.


1530W Machine learning approach to literature mining for the genetics of complex diseases. J. Schuster.


1536W Type TE: A tool for genotyping mobile element insertions in whole-genome sequencing data. J. Thomas.

1539W An automated functional annotation pipeline rapidly prioritizes clinically relevant genes for neurodevelopmental disorders which may impact treatment. O.J. Veatch.

1542W Fast estimation of genome-wide and partitioned genetic correlation for complex traits. Y. Wu.


1554W A new software ecosystem for high resolution ribosome profiling. H. Ozadam.

1557W MMIRNA_ER: A software package for studying miRNA and mRNA expression relationships. Z. Xu.

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1563W Undiagnosed Diseases Program Database (UDPdb) of genomic data: A model for cohort data consolidation, insights and reanalysis. N.J. Balandra.

1566W ‡ IncRNAKB: A comprehensive knowledgebase of long non-coding RNAs. F. Seifuddin.


1572W From NGS data to diagnosis: Fast HPO-based variant filtering of NGS data using Moon by Diploid. F. Sleutels.

1575W Leveraging large datasets accumulated through population carrier screening to inform variant classification. D.H. Tran.

1578W Expanding the scientific scope of the GWAS Catalog to include targeted and exome array studies. A. Buniello.

1581W Compound heterozygotes with ocularcutaneous albinism as a special case of outbreeding depression in an admixed Colombian population. W.A. Cardenas.

1584W The Alliance of Genome Resources: A next-generation comparative genomics data ecosystem for functionalizing the human genome. C. Bult.


1590W Pretrel: PRedicting and ETHnicity and RELatedness from genotyping data - an R Package. S. Tuna.

1593W A robust clustering algorithm for identifying cell types from single-cell RNA sequencing data. X. Liang.

1596W Analysis of sequencing error profile across different next-generation sequencing platforms. J. Bai.

1599W Scalable probabilistic PCA for large-scale genetic variation data. A.M. Chiu.

1602W High-throughput genetic variant classification for inherited cancer gene panels through an artificial intelligence inference engine. F.M. De La Vega.

1605W ‡ A framework for mapping GWAS loci to causal genes for common disease and traits. V. Forgetta.

1608W Multisample metrics for genome-wide single-position-resolution sequence quality assessment. N. Johnson.

1611W Identifying clinically relevant results in 145,000 exomes: Experiences and lessons learned from population genomic screening in the Geisinger MyCode® Community Health Initiative. M.A. Kelly.

1614W Database and supercomputer resources for human data at DDBJ. Y. Kodama.

1617W Machine learning models for classification of small sample size RNA sequencing data. S. Listopad.

1620W ClinGen Linked Data Hub: Scalable infrastructure for aggregation of diverse types of variant information to support phenotypicity assessment. A. Milosavljevic.

1623W OpenCRAVAT: A customizable annotation and prioritization pipeline for genes and variants. K. Pagel.

1626W Detecting transcriptional regulatory modules using machine learning. N. Patel.

1629W Integration of best practice RNA-Seq workflows into cloud-based translational analysis platform. M. Rickles-Young.

1632W Improved detection of allele-specific expression via personalised mapping approaches. A. Saukkonen.

1635W CAVERN: Computational and visualization environment for RNA-seq analyses. A.C. Shetty.


1641W Scandium: Sequencing coverage annotation database with uniformity metric. K. Walker.

1644W MARRVEL 2.0 (Model organism Aggregated Resources for Rare Variant ExpLoration): A web-tool for human genetics and cross-species data collection. J. Wang.

1647W De novo assembly and characterization of a breast cancer reference sample using multiple whole genome sequencing technologies. C. Xiao.

1650W CentroFlye: Assembling centromeres with long error-prone reads. A. Bzikadze.

1653W Accurate and high-resolution copy number variant detection in clinical germline screening. J. Li.

1656W Polygenic risk score ensembles across methods and diseases increase disease prediction accuracy. S. Kulm.


1662W Representation learning for variant interpretation. R. Chung.

1665W Predicting missense variant pathogenicity with C-MIP. P. Fradkin.

1668W Exploring the feasibility of data augmentation while using smaller biobank data sets. C.J. Lee.


1674W Human Cell Atlas data distribution: Curation of harmonized open access single cell datasets. J. Zamanian.


1680W ‡ ClipSV: Structural variation detection by read extension, spliced alignment, and local de novo assembly. P. Xu.


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1686W Quantifying the regulatory effect size for conditional eQTL data. N. Ehsan.

1689W Aggregated genome quality metrics can predict variant calling reproducibility in individual samples. C. Colombo.

1692W High-quality human genomes achieved through HiFi sequence data and FALCON-Unzip Assembly. Z.N. Kronenberg.


1698W Joint modeling of winner’s curse and study-specific confounding explains replication variability in genome-wide association studies. J. Zou.

1701W WGS sequencing at scale: Experiences from a national genome sequencing project. P. Fox.


1707W Comprehensive haplotype resolved MHC sequences from whole genome shotgun sequencing from single individual. J. Chin.


1716W Using mutual information to detect correlations between nucleotide positions in noncoding DNA. J. Dannemiller.

1719W Biological discovery and consumer genetics activate latent privacy risk in omics data. S.E. Brenner.


1725W Covariate adjusted rare-variant association (CARVA) testing for millions of samples. R. Bohlender.

1728W Assessment of statistical batch effect correction methods for different RNA sequencing data types. G. Cai.

1731W Visualization and analysis of single-cell RNA-seq data by alternative clustering. S. Liang.

1734W An Omics Analysis, Search and Information System (OASIS) for mining association summary statistics from biobanks and knowledge portals. J.A. Perry.

1737W SAIGEdgs: An efficient statistical tool for large-scale PhE-WAS with mixed models. X. Zheng.

1740W A method to discover combinatorial variants within transcription factors that are associated with gene expression variability. Y. Tang.

1743W Quantifying the quality of small INDELs called in sequencing analysis. H. Dai.

1746W Performance evaluation of different approaches for insertion and deletion variation detection from next generation sequencing data. S. Khan.

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Molecular Phenotyping and Omics Technologies

1749W Genomic landscape of conjunctival melanoma. K. Cisarova.

1752W Gene expression profiling from cervix cancer patients living in Senegal. G. Diop.

1755W Understanding genetic variation in cancer using targeted long-read sequencing. S. Iyer.


1761W Crowdsourced RNA-seq and gene ontology analysis of a human breast tumor in the context of an undergraduate course. P. Soneral.

1764W Structurally resolved immune profiling in ovarian carcinoma using whole-transcriptome spatial sequencing. S.R. Williams.

1767W Comparison between mutation profiles of paired whole blood and cDNA samples. A. Patel.

1770W Genomic landscape of subependymal giant cell astrocytomas: Unique gene expression profile with few somatic events. K. Giannikou.

1773W Identification of germline variants for hereditary cancer using the rapid, CleanPlex® multiplexed NGS panel. K. Pendleton.

1776W Low allele fraction variant discovery via electrical detection of nucleotide incorporations. J. Bell.

1779W An automated high-throughput custom single-cell targeted DNA sequencing platform to reveal rare clones and clonal evolution in cancer and characterize mutation profiles in CRISPR-edited cells. D. Mendoza.

1782W Single-cell RNA sequencing of B16 melanoma model reveal the trajectory of distant metastasis of melanoma. Y. Kim.

1785W Imputation of plasma metabolites using whole exome sequencing data. N.A. Bihlmeyer.

1788W Using genetic information for the reconstruction of metabolic processes underlying coronary heart disease: An example of the use of metabolomics in systems epidemiology of lipids. F. Drenos.


1794W Clinical, genetic and biochemical findings in a cohort of >1,000 patients with Gaucher disease. C. Beetz.


1800W Somatic hypermutation (SHM) in the human heavy chain immunoglobulin receptor: Regions of low and high mutational burden. D. Hospodsky.

1803W TotalSeq™ antibodies standardized antibody-oligo conjugates for CITE-Seq™, or single-cell proteogenomics. A. Chowdhury.
1806W The utility of WGS-derived telomere length in population health studies: A proof-of-concept using consumer wearable sleep metrics. J.X. Teo.

1809W Diagnosis of guanidinoacetate methyltransferase (GAMT) deficiency by untargeted metabolomic analysis in plasma. L. Lizcano-Gil.

1812W Transcriptional changes in circulating immune cells of patients with Gulf War Illness. L. Nathanson.

1815W Somatic mosaicism and mono-allelic expression across six brain cell types from single-cell RNA-seq in 48 AD individuals. M. Kousi.

1818W Concordance of germline and non-germline genotypes in glioblastoma multiforme patients. P. Dharia.


1833W A 12 minute, single tube, nanogram input library prep for WGS, FFPE, WES, and RNAseq. J. Dunham.

1836W The value of long read amplicon sequencing for clinical applications. L. Aro.

1839W Single enzymatic DNA fragmentation workflow simplifies library preparation from FF/FFPE samples of varying quality. M. Borns.

1842W A highly multiplexed target enrichment approach for sample identification and tracking using the NEBNext Direct Genotyping Solution. A.B. Emerman.

1845W Improving transcriptome analysis by incorporating unique molecular identifiers in RNA-sequencing. D. Posfai.

1848W High-throughput encapsulation of single cells in agarose microdroplets for 3D culture. T. Ayers.

1851W Resolution of a highly complex rearrangement of an SV40 insertion in an immortalized cell line using a combination of short and long read sequencing technologies. A. Sivachenko.

1854W Comparing whole exome sequencing probes at the sample and exon level for the human genome. C. Vaccaro.

1857W The impact of DNA source on genetic variant detection from human whole-genome sequencing data. B. Trost.

1860W Whole exome sequencing is “still” a valuable platform for variant detection in research: A 2019 overview of capture methods and improvements. R. Pellegrino.

1863W A versatile microfluidic platform for NGS library preparation enables multi-omics studies for a broad range of research samples and laboratory settings. Y. Chen.

1866W Detection and phasing of small variants in Genome in a Bottle samples with highly accurate long reads. W.J. Rowell.

1869W NGS DreamPrep™: A fully automated NGS library preparation solution from sample to quantified, pooled, and sequence-ready libraries. K. Modi.


1875W Clonal analysis, genetic screening, and expression profiling at a single-cell level. A. Chenchik.

1878W NuQuant: A fluorescence-based method for accurate quantification of NGS libraries in minutes. A. Saraiya.


1884W Electrical detection of single base incorporations enables high accuracy sequencing on a small, scalable platform. T. Clark.

Complex Traits and Polygenic Disorders


1890W Identifying candidate genes for hEDS: Initial results of the hEDS gene study. K. Martinez.


1899W Discovery of 159 signals for random plasma glucose and dissection of its relationship with type 2 diabetes pathophysiology using GWAS within UK biobank. M. Kaakinen.

1902W Sex-specific changes in fat accumulation and metabolic parameters in a murine knockout model of Lyplalf. R. Vohnoutka.

1905W Discovery of kidney function loci and rare variants in multi-generational diabetes pedigrees using variance component linkage analyses and next generation sequencing. S. Frodsham.

1908W Genomics as a personalized medicine approach in type 2 diabetes risk prediction - P5 FinHealth. H. Marjonen.

1911W Family-based association analysis of type 1 diabetes replicates case-control associations and reveals sequence homology as a source of spurious association in exome chip analysis. C.C. Robertson.


1917W Insights into the correlation between gluten intolerance and metabolic disorders: A case study. P. Kaur.

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1926W ‡ Body mass index and risk of dying from a bloodstream infection: A Mendelian randomization study of 56,000 subjects from the HUNT Study with 23 years follow-up. T. Rogne.


1932W Whole exome sequencing to identify genetic risk variants underlying obesity in pediatric patients. J.B. Cordero.


1938W Association study of LEP -2548 G/A polymorphism with obesity in North Indian Punjabi population. H. Kaur.

1941W Adiposity genetic risk score predicts the development of obesity from childhood to adulthood. M. Shi.

1944W Association between genetic admixture estimates for 5 ancestral populations and serum lipid levels in Pacific Islanders. W.-C. Hsueh.

1947W ‡ Genetic architecture of molecular phenotypes and their relationship with the genetics of T2D: A DIRECT study. A. Vifuela.


1953W ATP-binding cassette (ABC) transporter recessive mutations in biliary atresia cases. W.Y. Lam.


1959W Large-scale genome-wide association study identifies candidate genes for childhood steroid-sensitive nephrotic syndrome. X. Jia.


1971W Genetic studies of height-associated protein expression levels. E. Bartell.


1977W Exome sequencing of a multiplex family with idiopathic scoliosis implicates KIF7 in IS pathogenesis. M. Cuevas.


1986W Kabuki syndrome: Long time follow-up of two patients and the importance of a multidisciplinary team. D. Sabau.

1989W A locus on 4q28 is a laterality modifier with opposite effects for left- and right-sided cleft lip. E.J. Leslie.


1995W ‡ES Large-scale genetic analysis identifies 66 novel loci for asthma. Y. Han.

1998W Whole-genome sequencing and cell-type specific RNA sequencing of a multiplex pedigree identifies genes for juvenile idiopathic arthritis. C.N. Avery.

2001W Complement contributes to sex differences in risk of lupus and schizophrenia. N. Kamitaki.


2013W ‡ Trans-ancestral dissection of urate and gout associated major loci SLC2A9 and ABCG2 reveals primate-specific regulatory effects. W. Wei.

2016W Integrative analysis of disease association and blood eQTL data of Korean patients with Crohn’s disease identifies TNFSF15 and GPR35 at two reported loci. S. Jung.

2019W Multiethnic GWAS identifies novel genetic regulators of the IgA system with relevance to human disease. L. Liu.


2025W Identifying novel high-impact rare disease-causing mutations, genes, and pathways in exomes of Ashkenazi Jewish inflammatory bowel disease patients. Y. Wu.

2028W A GWAS of neonatal sepsis shows significant genetic differences between males and females. S. Williams.

2031W The effects of common TMRPSS6 and TF variants on iron status in healthy Gambian women of reproductive age. M. Jallow.

2034W The emerging role of RBFOX1 in visual impairment genetics. F. Bolduc.

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2037W Genetics of hypertriglyceridemic acute pancreatitis: Analysis of a prospective acute pancreatitis cohort and a complex family. P.J. Greer.

2040W A novel variant in CETP is associated with higher HDL-cholesterol in people of Polynesian ancestry. M. Krishnan.

2043W A phenome-wide and genome-wide analysis of triglyceride levels in the Healthy Nevada Project cohort. J.J. Grzymski.

2046W Executive function GWAS in a multi-ethnic cohort implicates region on chromosome 1. N. Dueker.

2049W BET1L is associated with survival time in the Japanese population. M. Akiyama.

2052W ‡ Genetic determinants of mosaic loss of the Y chromosome and their impact on age-related disease in the VA's Million Veteran Program. B. Gorman.

2055W Modified ACMG classification criteria for mitochondrial tRNA variants. T. Chen.

2058W Atypical nested (B-D) 22q11.2 deletion involving CRKL in a fetus with severe growth restriction and mother with severe systemic autoimmune disease. I. Lin.

2061W A case series of a mother and two daughters with a GLI2 gene deletion demonstrating variable expressivity. C. Elward.

2064W ‡ Integration of polygenic risk score with newborn metabolomics provides increased accuracy in profiling risk of infantile hypertrophic pyloric stenosis. J. Fadista.

2067W Whole exome sequencing identifies a rare functional variant SHARPIN G186R associated with increased risk of late-onset Alzheimer’s disease. Y. Asanomi.

2070W Dissecting the local ancestry effect on APOE4 and Alzheimer association in an admixed Peruvian population. M. Cornejo-Olivas.

2073W Stratified case-control association mapping of Alzheimer’s disease using family history from UK Biobank. S. Esmaeili.

2076W Genetic associations with cognitive performance inform Alzheimer’s disease etiology. L. Gurski.

2079W Next generation sequencing in dementia subgroup in Thailand. P. Lertwilaiwittaya.

2082W Genome wide association study for dementias in a Japanese population. R. Mitsumori.


2091W Circadian-relevant gene PERIOD3 is related to autism spectrum disorder and has function neuronal development. A. Miyachi.


2100W Rare genomic variants impacting neuronal functions modify the Dup7q11.23 phenotype. F. Qaiser.

2103W Assessment of focal epilepsy subtypes using polygenic risk scores. M. Gramm.

2106W Association of expression quantitative trait loci in blood with ischemic stroke. H. Amini.

2109W Genetic profile of a multiplex Hirschsprung disease family. C.S. Tang.

2112W A genome-wide meta-analysis of migraine with over 102,000 cases identifies 124 risk loci and provides first genetic insights to new migraine therapeutics targeting CGRP pathway. H. Hautakangas.

2115W Characterization to the genomic landscape in SMS with whole genome sequencing. M.A. Fisher.

2118W Longitudinal positron emission tomography of dopamine synthesis in subjects with GBA1 mutations. E. Sidransky.


2127W Genetic analysis of risk alleles associated with frontotemporal dementia in a group of Colombian patients. A.I. Sanchez.

2130W Longitudinal analysis of 4500 whole blood transcriptomes across over 1500 whole-genome sequenced individuals within the Parkinson’s Progression Markers Initiative. D.W. Craig.

2133W ‡ Human iPSC-derived microglia are genetically relevant to Alzheimer’s disease. R.R. Butler.

2136W De novo/shared mutations identified for autism in the US Hispanic population. P. Acevedo.

2139W Bionformatics analysis of GWAS for autism spectrum disorders and related traits. R.M. Cantor-Chiu.


2145W Rare recurrent copy number variations in children with neurodevelopmental disorders. M.E. Khan.

2148W Non-additive genetic effects in autism spectrum disorder and specific language impairment on reading achievement and symptoms of the broader autism phenotype. L.L. Mason.

2151W Autism spectrum disorder in the Amish: Exome sequencing unveils a novel missense variant in EvC ciliary complex subunit 1 (EVC), a known regulator of the sonic hedgehog signaling pathway. C.G. Tise.


2157W Rare variants involved in risk of suicide death identified from targeted genome-wide array analysis. E. DiBlasi.

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2163W Prenatal alcohol exposure increases risk for ADHD after accounting for genetic liability. L. Wetherill.


2169W Novel mutation in PPEF2 reported in a family affected with schizophrenia: Whole genome sequencing study. R. Torres.


2175W Applying reverse linkage as a method in 1,589 Finnish migraine families. K. Veerapen.

2178W Evaluating the phenotype of the 15q13.3 deletion using clinical MRI data. A. Hare-Harris.

2181W Genetics of severe mental illness in a Colombian population isolate. L. Olde Looohuis.

2184W Exome variation in ethanol metabolizing enzyme genes in Irish severe Alcohol Dependence (AD) cases. M. Ahangari.

2187W Cell-type specific expression quantitative trait loci during human neocortical differentiation. N. Aygun.

2190W Whole genome sequencing determines contributions of rare and common genetic variants to risk for schizophrenia. B. AdhamiMojarad.

2193W‡ Genome-wide association meta-analysis of nicotine dependence reveals novel loci and shared genetic influences with multiple traits: Findings from the iNDiGO (Nicotine Dependence GenOmics) consortium. B.C. Quach.

2196W The brain transcriptomic profiles of immunogenic system in major neuropsychiatric disorders: A cross-disorder study. Y. Chen.

2199W Polygenic risk scores for intelligence and schizophrenia: Whole genome sequencing study. L. Xiong.

2200W Genome-wide association study in multiplex consanguineous Pakistani pedigrees with schizophrenia and bipolar disorder. L. Xiong.

2203W‡ Discovery of novel genetic risk loci for acute central serous chorioretinopathy and genetic pleiotropic effect with age-related macular degeneration. Y. Ren.

2206W Expression and alternative splicing QTLs integrated with GWAS reveal novel genetic associations and causal genes for glaucoma. A.R. Hamel.

2209W Identification of genetic markers associated with variable response in total sleep time in the first two-thirds of the night of tasimelteon-treated subjects. J. Brzezynski.

2212W A Scandinavian whole genome sequencing study of familial asthma. S. Cameron-Christie.

2215W 11,364 whole-genome sequences in the cloud for autism spectrum disorder research. S.W. Scherer.


2224W Assaying lung-specific accessible chromatin to predict the causal variants in COPD. C.J. Benway.

2227W Association of lung function polygenic risk scores with chronic obstructive pulmonary disease, quantitative computed tomography imaging features, and lung growth patterns. B.D. Hobbs.

2229W A GWAS identifies novel susceptibility loci for irritable bowel syndrome. J. Li.

2232W‡ Beyond GWAS: Fine-mapping, functional, and phenotypic follow-up of 44 genome-wide significant endometriosis associations in 61K cases and 71K controls. N. Rahmioglu.

2235W A GWAS identifies novel susceptibility loci for irritable bowel syndrome. J. Li.

2242W‡ The genetic overlap between body composition, glycemic, and psychiatric traits is age- and sex-dependent. C. Hübel.


2248W‡ Examining mtDNA variants and sleep disturbances in major depressive disorder. L. Melhuish Beaupre.

2251W Examining mtDNA variants and sleep disturbances in major depressive disorder. L. Melhuish Beaupre.

2254W Examining mtDNA variants and sleep disturbances in major depressive disorder. L. Melhuish Beaupre.

2257W Examining mtDNA variants and sleep disturbances in major depressive disorder. L. Melhuish Beaupre.

2260W Examining mtDNA variants and sleep disturbances in major depressive disorder. L. Melhuish Beaupre.

2263W Examining mtDNA variants and sleep disturbances in major depressive disorder. L. Melhuish Beaupre.

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2280W Pedigree based analysis of dizygotic twinning from whole genome sequencing. J.J. Beck.

2283W‡ Cross-population fine-mapping of 50 complex traits and diseases in ~675,000 individuals across three global biobanks. M. Kanai.


2289W Cross-population fine-mapping to identify shared and population-specific causal effects. Z. McCaw.

2292W A large-scale genome-wide association scan in East Asians revealed genetic mechanisms underlying variations of human facial morphology. S. Wu.

2295W Overfitting in polygenic risk score analyses: Exploring the impact of sample overlap and first degree relatives. S. Medland.

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**Evolution and Population Genetics**


2301W GATTC1 haplotype in CD36 gene may predict the risk of developing T2DM associated BMI and hypertension. S. Gautam.

2304W Exploring health, disease, and diet in Ancient Rome through paleogenomics. H. Moots.

2307W Higher Native American ancestry proportion is associated with TB progression risk in the Peruvian population. S. Asgari.

2310W Complete mitogenome sequencing of 605 CEPH family pedigree samples reveals spurious substitutions attributed to cell culture. C. Marshall.

2313W‡ The spectrum of mitochondrial genomic variation across 250,000 individuals. N.L. Washington.


2319W Genome-wide detection and characterization of de novo repeat expansions in the general population. I. Mitra.

2322W DCDC2 READ1 regulatory element: How perception shapes language. M.M.C. DeMille.

2325W Whole genome sequence data reveal global demographic dynamics respond to the last ice age. X. Liu.


2331W‡ A functional genomics investigation reveals the molecular basis for adaptation to diving in Sea Nomads. M. Ilardo.

2334W Evolutionary dynamics of sex-biased gene expression in mammalian tissues. S. Naqvi.

2337W A catalogue of human ancestry components from population studies. C. Barbieri.


2343W Understanding the biological meaning of the human genome sequence by studying diverse indigenous populations in East Asia and Southeast Asia. S. Xu.

2346W Patterns of cross ancestry constraint in regulatory regions. T.J. Hayeck.

2349W The genetic consequences in the Americas of the transatlantic slave trade. S. Micheletti.

2352W Reconstructing the peopling of old world South Asia: From modern to ancient genomes. N. Rai.


2361W‡ Imputing gene regulation in ancient humans. L.L. Colbran.

2364W‡ Evolutionary modeling of the differential contribution of Neanderthal ancestry to complex traits provides insights into selective forces that shape trait variation. C.R. Robles.

2367W Putting RFMix and ADMIXTURE to the test in a complex admixed population. C. Uren.


2373W Prioritizing natural selection signals from the deep-sequencing genomic data suggests multi-variant adaptation in Tibetan highlanders. L. Deng.

2376W Distinct patterns of genetic ancestry and adaptation in the islanders of Taiwan. W.Y. Ko.

2379W Ancestral origins and natural selection of Highland Tajik people in Xinjiang. J. Liu.

2382W‡ A theoretical framework for interpreting the effect of geographic structure on GWAS. C. Porras.


2388W Imputation in South Asian individuals using a high-cover-age South Asian reference panel. S. Belsare.

2391W‡ Re-examining the evidence for polygenic adaptation at height-associated loci in mainland Europe and Sardinia. C. Chiang.

2394W Leveraging chromosomal linkage to infer natural selection from full-genome sequencing data. E. Friedlander.


2400W Study genetic ancestry and homozygosity disequilibrium in global populations. H.-C. Yang.


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Molecular and Cytogenetic Diagnostics

2412W ‡ Optimized early cancer detection from whole genome sequencing of cell-free DNA. C. Melton.

2415W Distinctive expression of specific microRNAs in breast cancer patients and change during chemotherapy. Y. Balcha.


2424W ‡ Common variants in PMS2CL that can present in PMS2 as pathogenic variants with extremely low frequencies. S. Pan.


2430W ADAM32 amplification in pediatric malignancies. V.T. Tonk.


2436W Next generation sequencing of BRCA1 and BRCA2 genes among women with breast cancer in Burkina Faso. M. Biancolella.

2439W DNA-depleted plasma-based extraction controls enable effective assessment of specificity and sensitivity of liquid biopsy assays. Y. Lu.

2442W Gene panel based prediction of homologous recombination deficiency in adolescent and young adult breast cancers. T. Watanabe.

2445W High accuracy NGS platform based on electrical impedance detection: Applications for oncology research. S. Paliwal.

2448W PAP technology: Ultrahigh sensitivity and specificity in detection of nucleotide variants using cell-free DNA from liquid biopsies. C. Boysen.

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2616W Implementation of sex identification and sample tracking into clinical genomic analysis workflows. J. Hu.

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Cardiovascular Phenotypes

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2628W The Cardiac Genome Clinic: Genome sequencing in families with congenital heart disease. M.S. Reuter.

2631W Changes in variant classification between KCNQ1-modified criteria and standard ACMG 2015 guidelines. M. Care.

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2637W Mutation spectrum of dilated cardiomyopathy from Korean patients. J. Lee.

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3351W Characterizing the regulatory architecture of essential genes in the human genome. S. Ramdas.

3354W Gene circuitry mapping assigns validated functional annotation to GWAS non-coding hits: from primary human hepatocytes to culprit disease-associated genes. Y. Liu.

3357W MAPS: Model-based analysis of long-range chromatin interactions from PLAC-seq and HiChIP experiments. M. Hu.

3360W Human enhancers with complex age architectures are more active than enhancers with simple architectures. S. Fong.

3363W Incomplete X chromosome inactivation is reflected in complex trait genetics architecture. Y. Fu.