Genome Structure and Function

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Prenatal, Perinatal, and Reproductive Genetics

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Genetic Counseling, ELSI, Education, and Health Services Research

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1888W GWAS polygenic model approach applied to primary biliary cholangitis (PBC) in a Japanese population. O. Gervais.

1891W ‡ Transcription factors are associated with disease risk loci: Epstein-Barr virus nuclear antigen 2 (EBNA2) is an environmental factor associated with multiple autoimmune diseases. J.B. Harley.

1894W Genetic screening of Galectin-3 CRD variants in RA: A case-control association study. T. Kaur.

1897W Genetic association between not related to HLA immune gene polymorphisms and development of specific autoantibody is limited to few genetic loci in patients with rheumatoid arthritis. L. Padyukov.

1900W Integrative analysis of genetic, gene expression and DNA methylation data on systemic lupus erythematosus (SLE). W. Yang.

1903W *IL1RN* variants influence systemic juvenile idiopathic arthritis susceptibility and are a biomarker of non-response to treatment with anakinra. E.G. Shuldiner.

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1909W Multiple HLA B*57 alleles, sharing the amino acid residue valine^{97}, are associated with drug-induced liver injury due to flucloxacillin in a European population. P. Nicoletti.

1912W Human genetic variation impacts total IgA levels and pathogen-specific IgG levels. P. Scepanovic.

1915W Exome sequencing identifies variants of the alkylglycerol monooxygenase gene (*AGMO)* as a cause of relapses in visceral leishmaniasis in Sudan. S. Marquet.

1918W WGS identifies rare variants influencing variation in blood cell traits in Mexican American families. N.B. Blackburn.

1921W GWA and MHC-fine mapping analyses of multiple sclerosis (MS) age at diagnosis (AAD) identify novel associations with an HLA-DQ heterodimer and *GZMA*. P.G. Bronson.

1924W Genome-wide association study identifies *HLA-DR/DQ* region for childhood nephrotic syndrome in Japanese. X. Jia.

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

1930W GWAS meta-analysis in Chinese and European populations identified a novel locus associated with systemic lupus erythematosus on Xp11. H. Zhang.

1933W Association analysis of rheumatoid arthritis through whole-exome sequencing in a Singapore Chinese cohort. V. Kumar.

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1939W Genetic variation in *GLS2* is associated with development of complicated *Staphylococcus aureus* bacteremia. W.K. Scott.

1942W *ITPKC* and *SLC11A1* gene variations are associated with Kawasaki disease patients. Y. Bae.

1945W Male-specific association of the *FCGR2A* His167Arg polymorphism with Kawasaki disease. Y. Kwon.

1948W HLA-DPB1 and Graves disease in Han Chinese. Y. Lee.

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

1954W Identifying genetic determinants of age at menarche and age at menopause in the Japanese population. M. Horikoshi.

1957W Rare variation associated with immunosuppressant drug concentrations: Moving beyond common SNPs in predicting drug metabolism. A.A. Seyerle.

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1996W Developement of the scleroderma genotype-phenotype map for assessing phenotypic relationships and drug repurposing. K. Chesmore.

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2002W Identification and validation of novel key drivers of Alzheimer's disease from multiscale causal networks that integrate large-scale DNA, RNA, and proteomic data. N.D. Beckmann.

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2017W The roles of *CD33* and *TREM2* in neurodegeneration associated with Alzheimer's disease (AD) and frontotemporal dementia (FTD). A. Rendina.

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2053W A longitudinal metabolome-wide association study on beta amyloid in adults with increased risk for Alzheimer's disease. B.F. Darst.

2056W Protective effect of smoking and caffeine on Parkinson's disease: A gene-environment study. V. Altmann.

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2128W Whole genome sequence association analysis of tobacco use in the Trans-Omics for Precision Medicine Whole Genome Sequencing Program (TOPMed). G. Datta.

2131W Copy number variation in Thai individuals with schizophrenia and schizoaffective disorder. N. Jinawath.

2134W Rare heterozygous mutation in glutamate receptor gene segregating in a schizophrenia family. P. Kukshal.

2137W BBS1 M390R/M390R mice have impaired anxiety-like behavior. T. Pak.

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2173W Trial to make the combined genotyping to detect high-risk individuals for cold medicine related Stevens-Johnson syndrome (CM-SJS) with severe ocular complications (SOC). M. Ueta.

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2611W Familial TAPVR with 15q11.2 (BP1-BP2) microdeletion. Y. Kuroda.

2614W Comparison of genetic architecture of isolated left ventricular noncompaction cardiomyopathy and familial dilated cardiomyopathy as assessed by whole exome sequencing. L. Piherová.

2617W Predisposition genetic screening for actionable cardiovascular conditions in patients undergoing heritable cancer syndrome testing: Prevalence of pathogenic variants in 10,812 individuals. S. Yang.

2620W Clinical evolution and recommendations for management of the smooth muscle dysfunction syndrome due to mutations of the ACTA2 arginine 179. E. Regalado.

2623W Loss-of-function variant in *FNDC3B* is associated with dominant pulmonary arterial hypertension in a pedigree. M. Cousin.

2626W A novel workflow for analysis of whole genome sequencing in cardiac disease using tissue-specific biological datasets. S.M. Hosseini.

2629W CRISPR-Cas9 mediated knockout of SEL1L and proteasomal inhibition reveal divergent degradation pathways for corresponding LDLR and VLDLR disease-causing mutants. B.R. Ali.

2632W Treatment of RIT1-associated cardiomyopathy with trametinib: Initial results in two patients. G. Andelfinger.

2635W ‡ Novel genetic associations for blood pressure identified via gene-alcohol consumption interaction in about 570K individuals. M.F. Feitosa.

2638W Association of a polymorphism in *ITGB3* with resistance to clopidogrel in early acute coronary syndrome in an admixed population from Colombia. A.V. Valencia-Duarte.

2641W Is HDL-C causally associated with risk of cardiovascular diseases in Han Chinese? A Mendelian randomization study with 10,000 subjects. M.I. Biradar.

2644W Genome-wide association analysis identifies multiple loci associated with coronary artery calcification in Koreans. S. Choi.

2647W Pharmacogenetic study on clopidogrel response among Filipinos. E.C. Cutiongco de la Paz.

2650W Association of common variants in arrhythmogenic cardiomyopathy desmosomal genes with ECG traits in the general population. L. Foco.

2653W Prevalence of variants of *FVL*, *PTH*, *PAI-1*, *MTHFR* and *EPCR* among Cardio vascular patients. M. Hosseini moghadam.

2656W Family study of noncompaction cardiomyopathy shows variability of cardiac phenotype within and between families. D. Majoor-Krakauer.

2659W Polygenic hyperlipidemias and coronary artery disease risk. P. Ripatti.

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

2665W The importance of epistatic interactions and fitness costs in congenital heart disease. E.O. Akhirome.

2668W Genetic determinants in the *LILR* gene family predicting statin intolerance. M.K. Siddiqui.

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

2674W Genome-wide association study using whole-genome sequencing recapitulates both rare and common risk alleles for Brugada syndrome. R. Redon.

2677W Circulating cholesteryl ester transfer protein (CETP) concentration: A genome-wide association study followed by Mendelian randomization on coronary artery disease. D. Mook-Kanamori.

2680W GWAS-driven pathway analyses and functional validation reveals *GLIS1* to associate with mitral valve prolapse. M. Yu.

2683W Primary lymphedema: A novel association with 22q11.2 deletion syndrome. M. Unolt.

2686W Exome-chip meta-analysis identifies association between variation in *ANKRD26* and platelet aggregation. A.D. Johnson.

2689W A longitudinal transcriptome analysis identifies novel gene expression signatures for body mass index in monocytes. C. Müller.

2692W Univariate and phenome-wide GWAS of correlated electrocardiographic traits offer novel insights into genetics of cardiac electrophysiology: the Population Architecture using Genomics and Epidemiology (PAGE) study. A.R. Baldassari.

2695W *APOL1* coding variants are associated with incident cardiovascular disease in community-dwelling African Americans. C.A. Winkler.

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

2701W A Bayesian approach for detecting gene by environment interactions with common and rare variants. S.M. Lutz.

2704W Race, sex, and age differences in GxE association: *EBF1* gene-by-stress interaction on central obesity differs among Blacks, Whites and male, female in multiple harmonized datasets. A. Singh.

2707W Cadmium exposure, *MT* polymorphisms and subclinical cardiovascular disease in American Indians: The Strong Heart Family Study. R. Hou.

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

2713W Towards precision therapy in hypertension: Genome-wide association study reveals genetic variants associated with uncontrolled blood pressure on thiazide diuretic/beta-blocker combination therapy. O. Magvanjav.

2716W Low frequency and rare variants in multiple genes are associated with sleep related traits using whole genome sequencing data. X. Zhu.

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2719W An information theoretic approach to filtering false signals of pathogenicity across ancestrally diverse populations. A.K. Manrai.

2722W Whole genome sequence reveals selection for muscle and cardiovascular functions in sport hunting dog breeds. J. Kim.

2725W Integrating biological age and transcriptome markers for predicting the functional recovery potential of patients undergoing mechanical circulatory support surgery. G. Bondar.

2728W A disease-specific and automated variant annotator enables fast and accurate clinical variant interpretation. N. Whiffin.

2731W ‡ Genetic variants in familial abdominal aortic aneurysms identified by whole genome and exome sequencing. A. IJpma.

2734W ‡ Large-scale generation of iPSC-derived cardiomyocytes for functional genomic applications. M.K.R. Donovan.

2737W Danon Disease: A lysosomal hypertrophic cardiomyopathy model created by CRISPR editing LAMP2 in iPSC and fibroblasts. C. McKinney.

2740W Association of rare recurrent copy number variants in next generation sequencing data from family trios with congenital heart defects. Y. Liu.

2743W An integrated genetic-epigenetic prediction model for coronary heart disease. M. Dogan.

2746W Interpreting genetic variation in coronary artery disease (CAD). I. Selvarajan.

2749W Epigenome-wide association study of the previous number of strokes in participants from the Vitamin Intervention for Stroke Prevention (VISP) clinical trial. N.M. Davis-Armstrong.

2752W Characterization of experimentally validated heart disease genes using functional genomic information and 3d genome structure. R. Gill.

2755W The communal relation of MTHFR, MTR, RFC gene polymorphisms and hyperhomocysteinemia as plausible risk of congenital septal defects. S.B. Sunayana.

2758W A mutation in the *LMOD1* actin-binding domain segregating with disease in a large British family with thoracic aortic aneurysms and dissections. Y. Wan.

Statistical Genetics and Genetic Epidemiology

2761W Integration of GWAS and local genetic effects on gene expression (eQTL/ASE) highlights genes with kidney function and disease. C. Qiu.

2764W Understanding progression and subtypes of prediabetes with metabolomics and genomic profiling in Starr County Mexican Americans. G. Jun.

2767W Genome-by-environment interactions have a major impact on obesity. C. Amador.

2770W ‡ Fine-mapping and characterization of GWAS loci harboring extensive allelic heterogeneity. C. Spracklen.

2773W Long-term results of ENGAGE: A phase 3, randomized, double-blind, placebo-controlled, multi-center study investigating the efficacy and safety of eliglustat in adults with Gaucher disease type 1. P. Mistry.

2776W Genetic polymorphism of APOA5 gene is associated with metabolic syndrome in Koreans. S.W. Oh.

2779W Obesity revisited: Evidence of genetic predisposition for metabolically healthy obesity. L.O. Huang.

2782W Genetic determinants of glycemic response to metformin in the Million Veteran Program. C. Roumie.

2785W A novel approach to analyze the mediation model when the mediator is a censored variable. J. Wang.

2788W Gene-depressive symptoms interactions identify novel lipid loci in multi-ethnic cohorts. S.K. Musani.

2791W Enriched loss-of-function variants associating with lipids in Finns. P. Helkkula.

2794W Genetic variation associated with telomere length in African American children with and without asthma. M. White.

2797W Transcriptional risk scores link GWAS to eQTL and predict complications in Crohn's disease. U.M. Marigorta.

2800W ‡ Finding genomic variants regulating the exon-skipping. R. Liu.

2803W Testing for colocalization of causal variants underlying obstructive sleep apnea and immune-related phenotypes. S. Akle.

2806W Integrated clinical genome database on hepatitis B-related diseases for genome-wide association: Project goals and utilization of materials and genomic information in the ToMMo biobank. S. Teraguchi.

2809W A genome- and phenome-wide association study of diverticular disease using electronic health records. Y.J. Joo.

2812W ‡ Penalized regression for detecting rare variant effects under extreme phenotype sampling for continuous traits. C. Xu.

2815W Convergent evidence for *LRP2BP* in resilience to Alzheimer's disease. D. Felsky.

2818W ‡ Genome-wide rare variant imputation and tissue-specific transcriptomic analysis identify novel rare variant candidate loci in late-onset Alzheimer's Disease: The Alzheimer's Disease Genetics Consortium (ADGC). A.C. Naj.

2821W Genetic association study on white matter microstructure by integrating multiple neuroimaging datasets. J. Zhang.

2824W Genetics of age-related cognitive decline and relationship to Alzheimer's and other neurodegenerative diseases. M.P. Reeve.

2827W Genetic causes of death in US infants: Findings from the National Center for Health Statistics. C. Lally.

2830W Relationship between essential tremor and Parkinson's Disease. A.A. Gosch.

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2836W CNVs among Japanese individuals with neuropsychiatric diseases effect dosage sensitivity in ohnologs and genes expressions. M. Yamasaki.

2839W Quantifying the effect of copy-number variants on general intelligence in unselected populations. G. Huguet.

2842W Identification of novel genetic variants of DSM-5 alcohol use disorder: Genome-wide association study in NESARC-III. J. Jung.

2845W Smoking and neuroticism: Using Mendelian randomization to investigate causality. H. Sallis.

2848W Genetically predicted gene expression in the brain and peripheral tissues associated with PTSD. L.M. Huckins.

2851W Proper joint and conditional meta-analysis of sequence data in the presence of missing summary association statistics. D. Liu.

2854W FOLD: A method to optimize power in meta-analysis of genetic association studies with overlapping subjects. E. Kim.

2857W Genotype-phenotype study of *OPHN1* and *IL1RAPL2* genes mutations in children with intellectual disability. Y.M. Khimsuriya.

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2863W Whole exome sequencing reveals candidate variants for elevated intraocular pressure in the Beaver Dam Eye Study. W. Li.

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2869W The genetic architecture of the AVSD risk in Down syndrome: Results from chromosome 21 genome sequencing. X. Blanc.

2872W Genome-wide association analyses in large-scale multi-ancestry cohorts: Statistical challenges and opportunities. C. DeBoever.

2875W Evidence of *ZKSCANS, SULT2A1, TRIM4* and *BCL2L11* for serum dehydroepiandrosterone sulfate (DHEAS) levels: Replication from the Long Life Family Study (LLFS). P. An.

2878W Leukocyte Telomere Length (LTL) as a marker of biological aging in Iranian healthy adult population: Report on assay establishment and recent finding. F. Larti.

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2884W Caring without sharing: Genome-wide association and mapping on cohorts fragmented across institutional silos. A. Pourshafeie.

2887W Modeling the interactions between coding and non-coding RNA by kernel machines in binary phenotypes. S. Yang.

2890W Exploring the effect of minor allele frequency on the inflation of type I error rates for GWA studies of family data with non-normally distributed traits. J.A. Sabourin.

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2899W Platelet-derived growth factor genes, maternal binge drinking and obstructive heart defects. M.A. Cleves.

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2911W Genome wide meta-analysis for dental caries in childhood. S. Haworth.

2914W Genome-wide association study of HIV-1 subtype C in Botswana population. A.K. Shevchenko.

2917W Improving imputation by maximizing power. Y. Wu.

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2926W Clonal hematopoiesis: Genetic and phenotypic associations. C. Tian.

2929W Transformation of summary statistics from linear mixed model association on all-or-none traits to odds ratio. L.R. Lloyd-Jones.

2932W X wide association analysis identifies a novel *FRMPD4* locus associated with the differential sex risk for multiple sclerosis. Y. Zhou.

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2938W Measuring the rate and heritability of aging using machine learning methods. J. Ding.

2941W ‡ TRUFFLE: Tests of undetermined relationships between founders - fast, light and efficient. A. Dimitromanolakis.

2944W Integration statistics suggest gene expression in the exocrine pancreas may contribute to intestinal obstruction in cystic fibrosis. J. Gong.

2947W A genetic variants simulation program to simulate high order epistatic interactions for family-based studies. Q. Li.

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2950W Sum ranking, simple but powerful method for detecting pleiotropic loci. G.V. Roshchupkin.

2953W Quantification of MAF-dependent architectures in 14 UK Biobank traits reveals strength of genome-wide negative selection. A. Schoech.

2956W Association detection between ordinal trait and rare variants based on adaptive combination of p-values. Y. Zhou.

2959W GLMM-seq: Gene-based detection of allele-specific expression by RNA sequencing. J. Fan.

2962W Generalizing genetic risk scores from Europeans to Hispanics/Latinos. T. Sofer.

2965W Epistasis detection for human complex diseases in structured populations. K. Van Steen.

2968W POLARIS: Polygenic LD-Adjusted Risk Score approach for analysis of GWAS data. E. Baker.

2971W Admixture mapping: Controlling for multiple testing and spurious associations in the presence of population structure. K. Grinde.

2974W ‡ Integrative analysis of GWAS summary statistics and imputed gene expression in 44 tissues deciphers genetic architecture for many complex traits. M. Li.

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2980W Detecting heritable phenotypes without a model: Fast permutation testing for heritability and set-tests. R. Schweiger.

2983W ‡ Pervasive pleiotropy in the human genome revealed by a novel quantitative analysis of summary association statistics. D.M. Jordan.

2986W Allele specific information in Mendelian randomization. X. Wang.

2989W Identifying the clinical impact of loss-of-function intolerant genes using SKAT-O PheWAS. R. Sivley.

2992W ‡ DESCEND: Expression distribution deconvolution in scRNA-seq and characterization of transcriptional bursting and expression dispersion. J. Wang.

2995W Improved methods to estimate functional enrichment from genome-wide summary association data. K. Burch.

2998W Learning causal networks of molecular phenotypes with Mendelian randomization. A.Q. Fu.

3001W Estimating genetic correlations in functionally annotated regions using genome-wide summary association statistics. D.L. Kassler.

3004W ‡ Integrative analysis of eQTL and GWAS summary statistics to identify functional relationships. J. Morrison.

3007W ‡ Heritability informed power optimization (HIPO) leads to improved methods of discovering genetic association across multiple traits. G. Qi.

3010W A simple, consistent estimator of heritability from GWAS summary statistics. A. Schork.

3013W Bayesian model averaging for the X-chromosome inactivation dilemma in genetic association study. L. Sun.

3016W Using imputation for haplotype association. G.J.M. Zajac.

3019W ‡ Efficiently controlling for unbalanced case-control sampling and sample relatedness for binary traits in PheWAS by large cohorts. W. Zhou.

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