Genome Structure and Function

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1370F VCPA: Genomic variant calling pipeline and data management tool for Alzheimer’s disease sequencing project. Y. Chou.

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### Omics Technologies

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1736F Exome analyses in subfamily trios from large family tree in the South-Eastern Moravia (Czech Republic) population with high incidence of parkinsonism. R. Vodicka.

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1823F Molecular barcode thresholding and unique dual sample indices reduce/eliminate index cross talk/hopping between NGS libraries to improve variant calling accuracy. B. Mullinax.

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1844F Exploration of DNA methylation sites associated with adiponectin levels based on a gene co-expression network and DNA methylation data analysis. M. Nakatoci.

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2162F Association study of three single nucleotide polymorphisms (SNPs) of TERC- a telomeric gene, in diabetic nephropathy patients. G. Singh.

2165F Single cell heterogeneity analysis and CRISPR screen identify key β cell-specific disease genes. C. Weng.

2168F Exome sequencing identifies a nonsense variant in DAO associated with reduced energy expenditure and respiratory quotient in American Indians. P. Piaggi.

2171F Genome wide assessment for resting heart rate and shared genetics with type 2 diabetes and metabolic traits. Y. Guo.

2174F Targeted metabolomics and genome-wide association study identifies plasma mimo acids and genetic variants associated with non-alcoholic fatty liver disease. A. Huertas-Vazquez.


2183F Togetherness of lysinuric protein intolerance and HOIP deficiency in a boy: SLC7A7 and RNF31 gene disruptions. L. Aliyeva.

2186F Reassessing the significance of the PAH c.158G>A (p.Arg53His) variant in Korean patients with hyperphenylalaninemia. J. Kim.

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3128F The roles of SMYD4 in epigenetic regulation of cardiac development in zebrafish. D. Ma.

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3134F A Mendelian randomization analysis of hemostatic factors in peripheral artery disease. A.M. Small.

3137F Assessing and characterising the causal association of glycemic with risk of coronary heart disease. L.B.L. Wittemans.

3140F Genetic variants of electrical storm in post myocardial infarction patients. A. Rangaraju.

3143F Genetic risk scores for cardiometabolic traits in sub-Saharan Africans. K. Ekoru.


3149F ‡ GWAS of resistant hypertension in over 70,000 participants from the Million Veteran Program identifies genes with altered expression in the adrenal gland. J.N. Hellwege.

3152F Bilirubin and ischemic heart disease: A two-sample Mendelian randomization study. J. Lee.


3161F Genome-wide meta-analysis in >30,000 Caucasian subjects identify 7 novel loci for glycine. Q. Jia.

3164F Genomic loci associated with lipid traits in Latinos. I. Kullo.

3167F Towards a multi-modal health assessment: Gaining insights into cardio-metabolic conditions. I. Shomorony.

3170F Genome-wide association study of angioedema induced by agents acting on the angiotensin system. M. Wadelius.

3173F Replication of a single nucleotide polymorphism variant in CETP gene associated with HDL level with tiled regression in the ClinSeq® Study. H. Sung.


3179F Investigating pleiotropic architecture of 92 plasma proteins related to cardiovascular disease. L. Repetto.

3182F Mutation spectrum in a large cohort of hypertrophic cardiomyopathy patients in Korea. D. Yang.

3185F Heritability of atrial fibrillation across a spectrum of clinical risk factors. L. Weng.

3188F Natural genetic variation of the human atrial transcriptome, proteome and metabolome. I. Assum.

3191F Mega-analysis of the EBF1 GxE association: Evaluation of differences in gene-by-stress interaction across race, sex, and age in harmonized datasets of over 27,000 individuals. A. Singh.

3194F Hyperuricemia is an early onset metabolic disorder causally associated with cardiovascular disease event in Han Chinese. K.M. Chiang.

3197F NOTCH3 p.R544C mutation is a common and significant risk factor for ischemic stroke in Taiwanese population. Y.C. Liao.

3200F Whole genome sequencing identifies multiple rare variants in GTPase-activating protein (DLC1) associated with sleep disordered breathing. J. Liang.

3203F Bayesian whole genome association analysis of gene-age interactions. M. Kerin.

3206F Multi-phenotype genome-wide association study of protein levels in individuals with pulmonary arterial hypertension. E. Pileckyte.

3209F Inter-species differences in response to hypoxia and oxidative stress in iPSC-derived cardiomyocytes from humans and chimpanzees. M.C. Ward.


3215F A comprehensive and accurate knowledgebase system for variant clinical interpretation using ACMG-AMP standard guideline in inherited cardiac diseases and hereditary cancers. C. Chen.

3218F An artificial intelligence-based next-generation integrated genetic-epigenetic prediction of 5-year risk for symptomatic coronary heart disease. M.V. Dogan.

3221F ‡ Metabolic profiling and sequencing at biobank scale for prioritizing drug targets. S. Ruosaari.

3224F Comprehensive analysis of chromatin organization delineates regulatory programs of human cardiomyocyte differentiation. Y. Zhang.

3227F Methylation at nucleotide C62 in spliceosomal RNA U6 alters mRNA splicing which is important for embryonic development. A. Ogren.

3230F Using endothelial cell molecular QTLs to dissect coronary artery disease risk. L.K. Stolze.

3233F ‡ Genetic and epigenetic fine mapping of complex trait associated loci in the human liver. M. Caliskan.

3236F Enhancer-mediated enrichment of interacting JMJD3-DDX21 to ENPP2 locus prevents R-loop formation and promotes transcription elongation. D. Argaud.

3239F Large chromosome 4q25 intergenic deletion disrupts chromatin structure and leads to familial sinus node dysfunction. H. Murata.


3245F External chromatin accessibility quantitative trait loci (caQTLs) are enriched for association with human GWAS data independently of internal caQTLs. W.W. Greenwald.

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**Statistical Genetics and Genetic Epidemiology**

3251F ‡ Polygenic risk stratified by eQTL influence for obesity and related comorbidities for patients from the Geisinger MyCode Community Health Initiative. M. Butkiewicz.

3254F Genome-transcriptome-wide association of renal and morphometric traits across glomerular and tubular tissues in diabetic Pima Native Americans. A. Liu.

3257F Undiscovered novel polymorphisms in the SOWAH-C-RGPD5 intergenic region on chromosome 2q are associated with diabetic retinopathy. S.Y. Rhee.

3260F Rare variation in and near \textit{CREBBP} and association with fasting glucose in non-Polynesian participants in the TOPMed Program. R.L. Minster.

3263F ‡ A genome-wide and phenome-wide association study in individuals of African Ancestry identified novel loci of body mass index and obesity-related diseases. Y. Huang.

3266F Non-alcoholic Fatty Liver Disease (NAFLD) increases risk for Type 2 Diabetes (T2D) but decreases susceptibility to obesity: A Mendelian randomization study. Z. Liu.

3269F Metabolite-GWAS of obesity in the Atherosclerosis Risk in Communities (ARIC) study. K.L. Young.


3275F Polymorphism rs822396 (-3971 A/G) of \textit{ADIPQ} gene as a risk factor to develop metabolic syndrome in Mexican population: Preliminary results. L. Rubio Chavez.

3278F ‡ Using short identical-by-descent (IBD) segments to map disease genes in unrelated case-control studies. W.-M. Chen.

3281F High dimensional mediation for causal gene screening. Q. Zhang.

3284F Genomewide association for high-density lipoprotein change levels over time in three large combined studies. M.F. Feitosa.

3287F Variation in serum PCSK9, cardiovascular disease risk and potential unanticipated effects of PCSK9 inhibition: A GWAS and Mendelian randomization study in the HUNT study, Norway. B. Brumpton.

3290F \textit{TBC1D29} is associated with response to inhaled corticosteroids in adult asthmatics. A.L. Wang.

3293F GWAS of six red blood cell traits identifies multiple associations with ancestry-specific lead variants: The PAGE study. C.J. Hodonsky.

3296F Characterisation and somatic mosaicism of the human glycophorin DUP4 structural variant, and association with haemoglobin levels in a malaria-endemic village. W. Algady.

3299F Investigating the burden of nonsynonymous variation in PIK3delta pathway genes of individuals affected with idiopathic bronchiectasis. E.M. Schmidt.

3302F ‡ Genome-wide scan identifies loci for white blood cell and platelet traits in a multi-ethnic study from population architecture using genetic epidemiology (PAGE). S.A. Bien.

3305F Understanding the genetic determinants of peanut-specific IgG4 in the learning early about peanut allergy (LEAP) study. A.H. Winters.


3311F Identifying modifying genes to explain the variation in severity of fragile X-associated primary ovarian insufficiency. C.E. Trevino.

3314F Gene prioritization in kidney function associated loci through systematic colocalization with gene expression. K.B. Sieber.

3317F ‡ A weighted genetic risk score of 279 signals discovered in individuals of European descent is associated with impaired lung function and chronic obstructive pulmonary disease in multiple ancestries. A.L. Guyatt.


3323F \textit{HLA-DQB1}^*03:01 as a biomarker for genetic susceptibility to bullous pemphigoid induced by DPP-4 inhibitors. T. Ozeki.

3326F Shared causal paths underlying Alzheimer’s disease and type 2 diabetes. Z. Hu.


3332F Genome-wide association of plasma tau levels in the Framingham study. C. Sarowski.


3338F Analysis of \textit{APOE} and known depression SNPs in patients with multiple sclerosis. S.W. Brugger.

3341F Replication of the \textit{SCFD1} associated locus in ALS using linear mixed models. A. Nicolas.

3344F ‡ Genome-wide association analysis of excessive daytime sleepiness in the UK Biobank identifies 42 loci. R. Saxena.

3347F Regional collapsing of rare variation implicates specific regions in known and novel ALS genes. S. Gelfman.

3350F Bivariate causal discovery and its applications to gene expression and imaging data analysis. R. Jiao.

3353F Prioritizing risk genes for neurodevelopmental disorders using pathway information. T. Nguyen.

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3359F Significance testing for allelic heterogeneity. Y. Deng.

3362F Describing the genetic architecture of psychiatric disorders from population scale genealogies. A.J. Schork.

3365F Integration of mQTL data and enhancer-promoter interactions with GWAS summary results identifies novel schizophrenia-associated genes. C. Wu.

3368F Excess of rare, inherited variation in children with developmental disorders. K.E. Samocha.

3371F Gene-based analysis with multiple longitudinal glaucoma related phenotypes shows evidence for genetic factors in primary open angle glaucoma progression. A. Athanas.

3374F Association of variants on the X chromosome with age-related macular degeneration. M. Grunin.

3377F Association of low-frequency variants in regulatory regions with non-syndromic orofacial clefts. J.R. Shaffer.

3380F Association between distance from hospital birth and patient’s home with morbimortality outcomes in Bogotá and Cali in years 2015 - 2016. C. Tovar-Sánchez.

3383F ¶ Changing frequencies of the main autosomal trisomies in years 2015 - 2016. C. Wang.

3386F Genome-wide causal studies on Alzheimer’s disease. H. Qin.


3392F Inferring the rate of aging from quantitative traits using machine learning methods. J. Ding.

3395F Fine-mapping and colocalization at the IL1RL1 locus to identify asthma susceptibility effector genes. S. Ghosh.

3398F Gene-based kernel machine test for longitudinally measured binary phenotype. W. Wu.

3401F Leveraging functional enrichment improves polygenic prediction accuracy. C. Marquez Luna.

3404F Estimating the autocorrelation of causal SNP effect size magnitudes as a function of genomic distance. A.P. Schoech.

3407F On performing gene expression imputation and gene-level association analysis in multiethnic or admixed populations. A.I. Konkashbaev.

3410F Mixed-linear model adjustment recalibrated with LD structure improves statistical power for the detection of significant eQTLs. N. Patel.

3413F ¶ Phenome-wide analyses of pharmacomimetic variants in their genomic context: New opportunities for genetic validation of therapeutic targets in UK Biobank. J.D. Hoffman.

3416F Gene-based rare variant association tests for ancestry-matched case-control data. C. Wang.

3419F Genetics and nicotine dependence: Heaviness of smoking index in European individuals. C. Batini.

3422F Genetic associations with sleep apnea in a European population. Y. Huang.

3425F Three novel loci were identified for plasma acylcarnitine levels and their ratios in genome-wide association study. H. Li.

3428F Genetic correlation between polyoidal choroidal vasculopathy and age-related macular degeneration. Q. Fan.

3431F Previously ignored high-frequency common variants greatly contribute to heritability of a common disease. Y. Nagao.

3434F Assessment of genome-wide significance of conditionally independent signals. S. Ghasemi.

3437F ¶ Exploring the effect of parental height on a newborn’s birth weight. Y. Lee.

3440F ¶ Phenome-wide Mendelian randomization study mapping the influence of the plasma proteome on complex diseases. J. Zheng.

3443F The influence of mitochondrial DNA variation on complex traits. E.J. Grzeszkowiak.

3446F NAT2 genetic variants and toxicity related to anti-tubercular agents: A systematic review and meta-analysis. M. Richardson.

3449F Reducing the blind spots in target validation, examples from UK Biobank. L.M. Yerges-Armstrong.

3452F Extensive fine-scale population structure in the UK Biobank. S. Hu.

3455F A unified method for rare variant analysis of GxE interactions with application to GxSmoking interactions on obesity-related traits. E. Lim.

3458F Two-phase sampling designs for post-GWA fine-mapping studies: Optimality criteria. O. Espin-Garcia.


3464F A general statistic to test an optimally weighted combination of common and/or rare variants in association studies. J. Zhang.

3467F Identifying hidden ancestries in publicly available summary genetic data. A.E. Hendricks.

3470F Leveraging allele-specific expression to refine fine-mapping for eQTL studies. J. Zou.

3473F Why cross-study common controls are hard—and what can we do about it? S. Buyske.

3476F Epigenetics-prioritized imputation of gene expression identifies more tissue-specific disease genes. W. Liu.


3482F Enrichment of survey responses by genetic community within a large genetic genealogy database. E.P. Sorokin.

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3485F Robust and powerful analysis of gene-environment interactions using polygenic risk scores in case-control studies. N. Chatterjee.

3488F Robust region-based test for unbalanced case-control phenotypes. Z. Zhao.

3491F Refining fine-mapping: Effect sizes and regional heritability. C. Benner.


3497F A meta-analysis method to detect pleiotropic loci of correlated traits. C.H. Lee.

3500F Power and sample size calculations for genetic association studies in the presence of genetic model misspecification. C.M. Moore.


3506F Implications of post-colonial demographic structure on interpretations of association analyses. M. Zhang.

3509F Joint analysis of multiple phenotypes using a clustering linear combination method based on hierarchical clustering. X. Li.

3512F Reliable heritability estimation in admixed populations using covariate-adjusted LD score regression. X. Li.

3515F Relatedness disequilibrium regression estimates heritability without environmental bias. A.I. Young.

3518F Finding rare variants of large effect using identity-by-descent mapping. S. Shringarpure.

3521F Heterogeneity of causal estimates inferred from single genetic variants as instrumental variables in Mendelian randomization across complex traits and diseases. A. Dobbyn.

3524F ‡ Leveraging genome-wide significant loci to increase the power of S-LDSC to detect enrichment. K. Tashman.

3527F FinnGen: Towards a comprehensive catalogue of genomes and major health events for 500,000 Finnish residents. J. Karjalainen.

3530F Spatially-dense 3D facial heritability and genetic correlations in a heterogeneous sample. J. Li.

3533F Importance of protected statistical testing and replicate number in RNA-sequencing studies. R.Z. Blumhagen.

3536F Deconvolution of bulk tissue cell type proportions from single-cell RNA-seq. X. Wang.

3539F General retrospective mega-analysis framework for rare variant association tests. Y. Chiu.

3542F ‡ Improving gene expression prediction accuracy in transcriptome-wide association studies. J. Fryett.

3545F A universal and nearly optimal permutation testing approach and an application to association analysis of quantitative traits in whole-genome sequencing studies. J. Hecker.

3548F An analysis of the genetic overlap of 20 complex traits under a non-infinitesimal model. R.D. Johnson.

3551F A selection operator for summary association statistics reveals allelic heterogeneity of complex traits. Z. Ning.

3554F Accounting for cryptic relatedness across families between subjects with no genotype data. M. Saad.

3557F ‡ A statistical framework to quantify shared genetic variability in de novo mutations for complex diseases. Y. Shi.

3560F Building multi-predictor models of quantitative phenotypes from the Trinity Student Study with tiled regression. A.J.M. Sorant.

3563F Simulation study of double FDR method on LD block structure in GWAS. J. Xu.

3566F Genetic mapping of human diseases using gene damage scores. X. Zhu.