

Supplementary Table S1. Number of association entries for each rare variant association method.

<i>Method/Software</i>	<i>#Number of association entries</i>	<i>PMID of the corresponding publication of Method/Software</i>
collapsing analysis	48529	23176082; 34375979; 18691683
SKAT	10768	21737059
Burden test	9733	23483651; 36755099; 19214210; 20471002
BOLT-LMM	1499	25642633
SAIGE-GENE+	1426	36138231
SKAT-O	1053	22863193
STAAR-SKAT	736	36303018
aSPU	470	24831820
ACAT-V	467	30849328
F-distributed statistics	163	26058849
LRT	137	27980649
RLRT	131	36088354
TADA model	125	23966865
GATK	108	20644199
F-approximation tests	95	25809955
MetaSTAAR	94	36564505
VEGAS	86	20598278
CMC method	77	18691683
SAIGE-GENE	64	32424355
ExactVCTest	52	27646141
STAR Method	44	32492392
VT	43	20471002
C-alpha	39	21408211
two-stage method	35	19847924
maxT	19	21738570
WSS	18	19214210
AdaJoint	17	24022295
rareMETALS	15	24894501
LF-KM	14	27161037
RQTests	12	28633423
Meta-MultiSKAT	11	31433078
MetaSKAT-O	11	23768515
fitDNM	10	26235986
MetaSKAT	10	23768515
FLM	9	22889854
MAGMA	9	25885710
pVAASST	9	24837662
ARTP method	7	21209154
TASER	5	27152526
MURAT	4	26860061
Cox BT LRT	3	26782979
FB-GGRF	3	27061818
Rasch-based multi-marker association test	3	26379234

ANRV	2	19810025
aSKAT-O	2	22863193
ensemble learning algorithm	2	26619286
GEE	2	24704269
metaFARVAT	2	31275357
uFineMap	2	26458888
ARIEL	1	22441326
GATES	1	21397060
GESE	1	28191685
haplotype method	1	27980634
Hybrid BOMP	1	23358228
Hybrid BOMP+VEST	1	23358228
MiSTF	1	23483651
Positional BOMP	1	23358228
qMSAT	1	22262732
RAML	1	16986161
VT+VEST	1	23358228

Supplementary Table S2. Information for publications collected in the RAVAR database.

PMID	Title	First Author	Journal /Book	Publication Year	DOI
37301943	Rare genetic variants impact muscle strength	Huang Y	Nat Commun	2023	10.1038/s41467-023-39247-1
37262146	Rare penetrant mutations confer severe risk of common diseases	Fiziev PP	Science	2023	10.1126/science.abo1131
36914870	Schizophrenia risk conferred by rare protein-truncating variants is conserved across diverse human populations	Liu D	Nat Genet	2023	10.1038/s41588-023-01305-1
36809768	Effects of protein-coding variants on blood metabolite measurements and clinical biomarkers in the UK Biobank	Nag A	Am J Hum Genet	2023	10.1016/j.ajhg.2023.02.002
36778668	Systematic single-variant and gene-based association testing of thousands of phenotypes in 394,841 UK Biobank exomes	Karczewski KJ	Cell Genom	2022	10.1016/j.xgen.2022.100168
36672889	Skin Phototype and Disease: A Comprehensive Genetic Approach to Pigmentary Traits Pleiotropy Using PRS in the GCAT Cohort	Farré X	Genes (Basel)	2023	10.3390/genes14010149
36672803	Exome Array Analysis of 9721 Ischemic Stroke Cases from the SiGN Consortium	Xu H	Genes (Basel)	2022	10.3390/genes14010061
36635386	Genomic atlas of the plasma metabolome prioritizes metabolites implicated in human diseases	Chen Y	Nat Genet	2023	10.1038/s41588-022-01270-1
36568030	Whole-exome sequence analysis of anthropometric traits illustrates challenges in identifying effects of rare genetic variants	Young KL	HGG Adv	2022	10.1016/j.xhgg.2022.100163
36564505	Powerful, scalable and resource-efficient meta-analysis of rare variant associations in large whole genome sequencing studies	Li X	Nat Genet	2023	10.1038/s41588-022-01225-6
36450729	Whole-exome sequencing study identifies rare variants and genes associated with intraocular pressure and glaucoma	Gao XR	Nat Commun	2022	10.1038/s41467-022-35188-3
36419110	Whole-exome sequencing identifies novel protein-altering variants associated with serum apolipoprotein and lipid concentrations	Sandholm N	Genome Med	2022	10.1186/s13073-022-01135-6
36333282	The contribution of common and rare genetic variants to variation in metabolic traits in 288,137 East Asians	Kim YJ	Nat Commun	2022	10.1038/s41467-022-34163-2
36311265	Whole-exome sequencing in 415,422 individuals identifies rare variants associated with mitochondrial DNA copy number	Pillalamarri V	HGG Adv	2022	10.1016/j.xhgg.2022.100147
36303018	A framework for detecting noncoding rare-variant associations of large-scale whole-genome sequencing studies	Li Z	Nat Methods	2022	10.1038/s41592-022-01640-x
36297015	Whole Exome Sequencing Study Identifies Novel Rare Risk Variants for Habitual Coffee Consumption Involved in Olfactory Receptor and Hyperphagia	Cheng B	Nutrients	2022	10.3390/nu14204330
36280733	A combined polygenic score of 21,293 rare and 22 common variants improves diabetes diagnosis based on hemoglobin A1C levels	Dornbos P	Nat Genet	2022	10.1038/s41588-022-01200-1
36224396	A saturated map of common genetic variants associated with human height	Yengo L	Nature	2022	10.1038/s41586-022-05275-y

36220816	Whole genome sequence analysis of blood lipid levels in >66,000 individuals	Selvaraj MS	Nat Commun	2022	10.1038/s41467-022-33510-7
36168886	Differences and commonalities in the genetic architecture of protein quantitative trait loci in European and Arab populations	Thareja G	Hum Mol Genet	2023	10.1093/hmg/dac243
36138231	SAIGE-GENE+ improves the efficiency and accuracy of set-based rare variant association tests	Zhou W	Nat Genet	2022	10.1038/s41588-022-01178-w
36088354	Identifying interpretable gene-biomarker associations with functionally informed kernel-based tests in 190,000 exomes	Monti R	Nat Commun	2022	10.1038/s41467-022-32864-2
36050321	Genetic analyses of the electrocardiographic QT interval and its components identify additional loci and pathways	Young WJ	Nat Commun	2022	10.1038/s41467-022-32821-z
36038634	Large-scale sequencing identifies multiple genes and rare variants associated with Crohn's disease susceptibility	Sazonovs A	Nat Genet	2022	10.1038/s41588-022-01156-2
35982160	Rare coding variation provides insight into the genetic architecture and phenotypic context of autism	Fu JM	Nat Genet	2022	10.1038/s41588-022-01104-0
35954343	Germline Variants Associated with Nasopharyngeal Carcinoma Predisposition Identified through Whole-Exome Sequencing	Lee NY	Cancers (Basel)	2022	10.3390/cancers14153680
35935937	Gene-Based Variant Analysis of Whole-Exome Sequencing in Relation to Eosinophil Count	Höglund J	Front Immunol	2022	10.3389/fimmu.2022.862255
35741860	Identifying Rare Genetic Variants of Immune Mediators as Risk Factors for Autism Spectrum Disorder	Cai C	Genes (Basel)	2022	10.3390/genes13061098
35580180	Impact of natural selection on global patterns of genetic variation and association with clinical phenotypes at genes involved in SARS-CoV-2 infection	Zhang C	Proc Natl Acad Sci U S A	2022	10.1073/pnas.2123000119
35552711	Whole-exome sequencing of 14 389 individuals from the ESP and CHARGE consortia identifies novel rare variation associated with hemostatic factors	Pankratz N	Hum Mol Genet	2022	10.1093/hmg/dac100
35401413	Deciphering Genetic Susceptibility to Tuberculous Meningitis	Schurz H	Front Neurol	2022	10.3389/fneur.2022.820168
35361970	Polygenic prediction of educational attainment within and between families from genome-wide association analyses in 3 million individuals	Okbay A	Nat Genet	2022	10.1038/s41588-022-01016-z
35177841	Analysis of rare genetic variation underlying cardiometabolic diseases and traits among 200,000 individuals in the UK Biobank	Jurgens SJ	Nat Genet	2022	10.1038/s41588-021-01011-w
35115689	Combined effects of host genetics and diet on human gut microbiota and incident disease in a single population cohort	Qin Y	Nat Genet	2022	10.1038/s41588-021-00991-z
35078996	A genome-wide association study of serum proteins reveals shared loci with common diseases	Gudjonsson A	Nat Commun	2022	10.1038/s41467-021-27850-z
34887591	The power of genetic diversity in genome-wide association studies of lipids	Graham SE	Nature	2021	10.1038/s41586-021-04064-3
34737426	A generalized linear mixed model association tool for biobank-scale data	Jiang L	Nat Genet	2021	10.1038/s41588-021-00954-4
34662886	Exome sequencing and analysis of 454,787 UK Biobank participants	Backman JD	Nature	2021	10.1038/s41586-021-04103-z
34648354	Mapping the proteo-genomic convergence of human diseases	Pietzner M	Science	2021	10.1126/science.abj1541

34626176	Whole-exome sequencing reveals a role of HTRA1 and EGFL8 in brain white matter hyperintensities	Malik R	Brain	2021	10.1093/brain/awab253
34610981	Genetic Studies of Metabolomics Change After a Liquid Meal Illuminate Novel Pathways for Glucose and Lipid Metabolism	Li-Gao R	Diabetes	2021	10.2337/db21-0397
34594039	A cross-population atlas of genetic associations for 220 human phenotypes	Sakaue S	Nat Genet	2021	10.1038/s41588-021-00931-x
34376796	Analyses of biomarker traits in diverse UK biobank participants identify associations missed by European-centric analysis strategies	Sun Q	J Hum Genet	2022	10.1038/s10038-021-00968-0
34375979	Rare variant contribution to human disease in 281,104 UK Biobank exomes	Wang Q	Nature	2021	10.1038/s41586-021-03855-y
34321204	Testosterone and socioeconomic position: Mendelian randomization in 306,248 men and women in UK Biobank	Harrison S	Sci Adv	2021	10.1126/sciadv.abf8257
34260947	Common haplotypes at the CFH locus and low-frequency variants in CFHR2 and CFHR5 associate with systemic FHR concentrations and age-related macular degeneration	Lorés-Motta L	Am J Hum Genet	2021	10.1016/j.ajhg.2021.06.002
34244158	Genetic Analysis of Functional Rare Germline Variants across Nine Cancer Types from an Electronic Health Record Linked Biobank	Shivakumar M	Cancer Epidemiol Biomarkers Prev	2021	10.1158/1055-9965.EPI-21-0082
34230933	Impact of natural selection on global patterns of genetic variation, and association with clinical phenotypes, at genes involved in SARS-CoV-2 infection	Zhang C	medRxiv	2021	10.1101/2021.06.28.21259529
34226706	Whole-exome imputation within UK Biobank powers rare coding variant association and fine-mapping analyses	Barton AR	Nat Genet	2021	10.1038/s41588-021-00892-1
34131117	Genome sequencing unveils a regulatory landscape of platelet reactivity	Keramati AR	Nat Commun	2021	10.1038/s41467-021-23470-9
34125832	Whole-exome sequencing reveals insights into genetic susceptibility to Congenital Zika Syndrome	Borda V	PLoS Negl Trop Dis	2021	10.1371/journal.pntd.0009507
34016428	Identifying susceptibility genes for primary ovarian insufficiency on the high-risk genetic background of a fragile X premutation	Trevino CE	Fertil Steril	2021	10.1016/j.fertnstert.2021.04.021
33919687	Genetic Analysis Reveals Rare Variants in T-Cell Response Gene MR1 Associated with Poor Overall Survival after Urothelial Cancer Diagnosis	Bang L	Cancers (Basel)	2021	10.3390/cancers13081864
33804025	Set-Based Rare Variant Expression Quantitative Trait Loci in Blood and Brain from Alzheimer Disease Study Participants	Patel D	Genes (Basel)	2021	10.3390/genes12030419
33763119	Genome-Wide Identification of Rare and Common Variants Driving Triglyceride Levels in a Nevada Population	Read RW	Front Genet	2021	10.3389/fgene.2021.639418
33679876	Whole-Exome Sequencing and hiPSC Cardiomyocyte Models Identify MYRIP, TRAPPC11, and SLC27A6 of Potential Importance to Left Ventricular Hypertrophy in an African Ancestry Population	Irvin MR	Front Genet	2021	10.3389/fgene.2021.588452
33654115	Identification of candidate genes and pathways in retinopathy of prematurity by whole exome sequencing of preterm infants enriched in phenotypic extremes	Kim SJ	Sci Rep	2021	10.1038/s41598-021-83552-y
33574263	Rare genetic variants affecting urine metabolite levels link population variation to inborn errors of metabolism	Cheng Y	Nat Commun	2021	10.1038/s41467-020-20877-8

33432171	Exome-wide evaluation of rare coding variants using electronic health records identifies new gene-phenotype associations	Park J	Nat Med	2021	10.1038/s41591-020-1133-8
33257650	Identity-by-descent detection across 487,409 British samples reveals fine scale population structure and ultra-rare variant associations	Nait Saada J	Nat Commun	2020	10.1038/s41467-020-19588-x
33230300	Discovery of rare variants associated with blood pressure regulation through meta-analysis of 1.3 million individuals	Surendran P	Nat Genet	2020	10.1038/s41588-020-00713-x
33226994	A phenome-wide association study of 26 mendelian genes reveals phenotypic expressivity of common and rare variants within the general population	Tcheandjie u C	PLoS Genet	2020	10.1371/journal.pgen.1008802
33097823	The genetic architecture of appendicular lean mass characterized by association analysis in the UK Biobank study	Pei YF	Commun Biol	2020	10.1038/s42003-020-01334-0
33093519	Identifying genetic factors that contribute to the increased risk of congenital heart defects in infants with Down syndrome	Trevino CE	Sci Rep	2020	10.1038/s41598-020-74650-4
32929287	Complex genetic signatures in immune cells underlie autoimmunity and inform therapy	Orrù V	Nat Genet	2020	10.1038/s41588-020-0684-4
32888494	The Polygenic and Monogenic Basis of Blood Traits and Diseases	Vuckovic D	Cell	2020	10.1016/j.cell.2020.08.008
32888493	Trans-ethnic and Ancestry-Specific Blood-Cell Genetics in 746,667 Individuals from 5 Global Populations	Chen MH	Cell	2020	10.1016/j.cell.2020.06.045
32492392	Exome Sequencing Identifies Genes and Gene Sets Contributing to Severe Childhood Obesity, Linking PHIP Variants to Repressed POMC Transcription	Marenne G	Cell Metab	2020	10.1016/j.cmet.2020.05.007
32436959	Analysis of putative cis-regulatory elements regulating blood pressure variation	Nandakumar P	Hum Mol Genet	2020	10.1093/hmg/ddaa098
32424355	Scalable generalized linear mixed model for region-based association tests in large biobanks and cohorts	Zhou W	Nat Genet	2020	10.1038/s41588-020-0621-6
32415273	The exhaustive genomic scan approach, with an application to rare-variant association analysis	Kanoungi G	Eur J Hum Genet	2020	10.1038/s41431-020-0639-3
32100372	Convex combination sequence kernel association test for rare-variant studies	Posner DC	Genet Epidemiol	2020	10.1002/gepi.22287
32100327	Risk factors for progression of age-related macular degeneration	Heesterbeek TJ	Ophthalmic Physiol Opt	2020	10.1111/opo.12675
32042192	Using human genetics to understand the disease impacts of testosterone in men and women	Ruth KS	Nat Med	2020	10.1038/s41591-020-0751-5
31992710	Genome-wide rare variant analysis for thousands of phenotypes in over 70,000 exomes from two cohorts	Cirulli ET	Nat Commun	2020	10.1038/s41467-020-14288-y
31866045	UK Biobank Whole-Exome Sequence Binary Phenome Analysis with Robust Region-Based Rare-Variant Test	Zhao Z	Am J Hum Genet	2020	10.1016/j.ajhg.2019.11.012
31748686	An exome-wide rare variant analysis of Korean men identifies three novel genes predisposing to prostate cancer	Oh JJ	Sci Rep	2019	10.1038/s41598-019-53445-2
31433078	Meta-MultiSKAT: Multiple phenotype meta-analysis for region-based association test	Dutta D	Genet Epidemiol	2019	10.1002/gepi.22248
31367044	Exome sequencing of Finnish isolates enhances rare-variant association power	Locke AE	Nature	2019	10.1038/s41586-019-1457-z

31338326	Exome-Wide Rare Variant Analysis From the DiscovEHR Study Identifies Novel Candidate Predisposition Genes for Endometrial Cancer	Shivakumar M	Front Oncol	2019	10.3389/fonc.2019.00574
31275357	metaFARVAT: An Efficient Tool for Meta-Analysis of Family-Based, Case-Control, and Population-Based Rare Variant Association Studies	Wang L	Front Genet	2019	10.3389/fgene.2019.00572
31242253	Pharmacogenomics of statin-related myopathy: Meta-analysis of rare variants from whole-exome sequencing	Floyd JS	PLoS One	2019	10.1371/journal.pone.0218115
31235378	Effectiveness of Intensive Rehabilitation Therapy on Functional Outcomes After Stroke: A Propensity Score Analysis Based on Japan Rehabilitation Database	Kamo T	J Stroke Cerebrovasc Dis	2019	10.1016/j.jstrokecerebrovasdis.2019.06.007
31136621	Exome chip association study excluded the involvement of rare coding variants with large effect sizes in the etiology of anorectal malformations	van de Putte R	PLoS One	2019	10.1371/journal.pone.0217477
31034468	FunSPU: A versatile and adaptive multiple functional annotation-based association test of whole-genome sequencing data	Ma Y	PLoS Genet	2019	10.1371/journal.pgen.1008081
31019915	Novel Complex Interactions between Mitochondrial and Nuclear DNA in Schizophrenia and Bipolar Disorder	Schulman A	Mol Neuropsychiatry	2019	10.1159/000495658
30976013	Exome array analysis of rare and low frequency variants in amyotrophic lateral sclerosis	Dekker AM	Sci Rep	2019	10.1038/s41598-019-42091-3
30718457	Genetic loci for alcohol-related life events and substance-induced affective symptoms: indexing the "dark side" of addiction	Peng Q	Transl Psychiatry	2019	10.1038/s41398-019-0397-6
30679032	Exome Chip Meta-analysis Fine Maps Causal Variants and Elucidates the Genetic Architecture of Rare Coding Variants in Smoking and Alcohol Use	Brazel DM	Biol Psychiatry	2019	10.1016/j.biopsych.2018.11.024
30665703	Assessing the Pathogenicity, Penetrance, and Expressivity of Putative Disease-Causing Variants in a Population Setting	Wright CF	Am J Hum Genet	2019	10.1016/j.ajhg.2018.12.015
30659681	A large-scale exome array analysis of venous thromboembolism	Lindström S	Genet Epidemiol	2019	10.1002/gepi.22187
30578418	Trans-ethnic association study of blood pressure determinants in over 750,000 individuals	Giri A	Nat Genet	2019	10.1038/s41588-018-0303-9
30382371	Rare variants in the neuronal ceroid lipofuscinosis gene MFSD8 are candidate risk factors for frontotemporal dementia	Geier EG	Acta Neuropathol	2019	10.1007/s00401-018-1925-9
30377230	Genetic Analysis of 400 Patients Refines Understanding and Implicates a New Gene in Atypical Hemolytic Uremic Syndrome	Bu F	J Am Soc Nephrol	2018	10.1681/ASN.2018070759
30315176	Large-scale whole-exome sequencing association studies identify rare functional variants influencing serum urate levels	Tin A	Nat Commun	2018	10.1038/s41467-018-06620-4
30255815	Rare variants in the splicing regulatory elements of EXOC3L4 are associated with brain glucose metabolism in Alzheimer's disease	Miller JE	BMC Med Genomics	2018	10.1186/s12920-018-0390-6
30212457	Exome sequencing-based identification of novel type 2 diabetes risk allele loci in the Qatari population	O'Beirne SL	PLoS One	2018	10.1371/journal.pone.0199837

30206357	Exploring rare and low-frequency variants in the Saguenay-Lac-Saint-Jean population identified genes associated with asthma and allergy traits	Morin A	Eur J Hum Genet	2019	10.1038/s41431-018-0266-4
30175238	Meta-analysis of exome array data identifies six novel genetic loci for lung function	Jackson VE	Wellcome Open Res	2018	10.12688/wellcomeopenres.12583.3
30167848	Rare coding variant analysis in a large cohort of Ashkenazi Jewish families with inflammatory bowel disease	Schiff ER	Hum Genet	2018	10.1007/s00439-018-1927-7
30140000	Deep-coverage whole genome sequences and blood lipids among 16,324 individuals	Natarajan P	Nat Commun	2018	10.1038/s41467-018-05747-8
30108311	Whole exome sequencing study identifies novel rare and common Alzheimer's-Associated variants involved in immune response and transcriptional regulation	Bis JC	Mol Psychiatry	2020	10.1038/s41380-018-0112-7
30012220	Exome-chip meta-analysis identifies novel loci associated with cardiac conduction, including ADAMTS6	Prins BP	Genome Biol	2018	10.1186/s13059-018-1457-6
29898891	Germline Variation and Breast Cancer Incidence: A Gene-Based Association Study and Whole-Genome Prediction of Early-Onset Breast Cancer	Scannell Bryan M	Cancer Epidemiol Biomarkers Prev	2018	10.1158/1055-9965.EPI-17-1185
29875488	Genomic atlas of the human plasma proteome	Sun BB	Nature	2018	10.1038/s41586-018-0175-2
29867916	Evaluating the Genetics of Common Variable Immunodeficiency: Monogenetic Model and Beyond	de Valles-Ibáñez G	Front Immunol	2018	10.3389/fimmu.2018.00636
29852030	Association of Genetic Variants With Response to Anti-Vascular Endothelial Growth Factor Therapy in Age-Related Macular Degeneration	Lorés-Motta L	JAMA Ophthalmol	2018	10.1001/jamaophthalmol.2018.2019
29748316	Common and Rare Coding Genetic Variation Underlying the Electrocardiographic PR Interval	Lin H	Circ Genom Precis Med	2018	10.1161/CIRCGEN.117.002037
29486463	Genetic Variation in Genes Underlying Diverse Dementias May Explain a Small Proportion of Cases in the Alzheimer's Disease Sequencing Project	Blue EE	Dement Geriatr Cogn Disord	2018	10.1159/000485503
29455858	A Large-Scale Multi-ancestry Genome-wide Study Accounting for Smoking Behavior Identifies Multiple Significant Loci for Blood Pressure	Sung YJ	Am J Hum Genet	2018	10.1016/j.ajhg.2018.01.015
29441677	A novel susceptibility locus in MST1 and gene-gene interaction network for Crohn's disease in the Chinese population	Wu WKK	J Cell Mol Med	2018	10.1111/jcmm.13530
29411426	Integrating eQTL data with GWAS summary statistics in pathway-based analysis with application to schizophrenia	Wu C	Genet Epidemiol	2018	10.1002/gepi.22110
29378355	A Genome-wide Study of Common and Rare Genetic Variants Associated with Circulating Thrombin Activatable Fibrinolysis Inhibitor	Stanne TM	Thromb Haemost	2018	10.1160/TH17-04-0249
29321365	Genome-based exome sequencing analysis identifies GYG1, DIS3L and DDRGK1 are associated with myocardial infarction in Koreans	Lee JY	J Genet	2017	10.1007/s12041-017-0854-z

29273807	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity	Turcot V	Nat Genet	2018	10.1038/s41588-017-0011-x
29263008	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study	Guerreiro R	Lancet Neurol	2018	10.1016/S1474-4422(17)30400-3
29218897	Codon bias among synonymous rare variants is associated with Alzheimer's disease imaging biomarker	Miller JE	Pac Symp Biocomput	2018	
29190701	A non-threshold region-specific method for detecting rare variants in complex diseases	Hsieh AR	PLoS One	2017	10.1371/journal.pone.0188566
29185836	Exome-chip meta-analysis identifies association between variation in ANKRD26 and platelet aggregation	Chen MH	Platelets	2019	10.1080/09537104.2017.1384538
29155802	Investigation of common, low-frequency and rare genome-wide variation in anorexia nervosa	Huckins LM	Mol Psychiatry	2018	10.1038/mp.2017.88
29093530	Sporadic Hirschsprung Disease: Mutational Spectrum and Novel Candidate Genes Revealed by Next-generation Sequencing	Zhang Z	Sci Rep	2017	10.1038/s41598-017-14835-6
28973304	Exome-wide association study reveals novel psoriasis susceptibility locus at TNFSF15 and rare protective alleles in genes contributing to type I IFN signalling	Dand N	Hum Mol Genet	2017	10.1093/hmg/ddx328
28900119	Analysis of potential protein-modifying variants in 9000 endometriosis patients and 150000 controls of European ancestry	Sapkota Y	Sci Rep	2017	10.1038/s41598-017-10440-9
28879196	Assessment of Whole-Exome Sequence Data in Attempted Suicide within a Bipolar Disorder Cohort	Monson ET	Mol Neuropsychiatry	2017	10.1159/000454773
28867149	Establishing the role of rare coding variants in known Parkinson's disease risk loci	Jansen IE	Neurobiol Aging	2017	10.1016/j.neurobiolaging.2017.07.009
28814775	Mutational profile of rare variants in inflammasome-related genes in Behçet disease: A Next Generation Sequencing approach	Burillo-Sanz S	Sci Rep	2017	10.1038/s41598-017-09164-7
28736464	The Generalized Higher Criticism for Testing SNP-Set Effects in Genetic Association Studies	Barnett I	J Am Stat Assoc	2017	10.1080/01621459.2016.1192039
28642624	Whole Exome Sequencing to Identify Genetic Variants Associated with Raised Atherosclerotic Lesions in Young Persons	Hixson JE	Sci Rep	2017	10.1038/s41598-017-04433-x
28633423	rqtl: an R package for gene-level meta-analysis	Zhbannikov IY	Bioinformatics	2017	10.1093/bioinformatics/btx395
28589856	Association analysis of rare variants near the APOE region with CSF and neuroimaging biomarkers of Alzheimer's disease	Nho K	BMC Med Genomics	2017	10.1186/s12920-017-0267-0
28552196	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits	Tachmazidou I	Am J Hum Genet	2017	10.1016/j.ajhg.2017.04.014

28539126	Knowledge-driven binning approach for rare variant association analysis: application to neuroimaging biomarkers in Alzheimer's disease	Kim D	BMC Med Inform Decis Mak	2017	10.1186/s12911-017-0454-0
28526295	Large-scale exploratory genetic analysis of cognitive impairment in Parkinson's disease	Mata IF	Neurobiol Aging	2017	10.1016/j.neurobiolaging.2017.04.009
28513607	Variants in TTC25 affect autistic trait in patients with autism spectrum disorder and general population	Vojinovic D	Eur J Hum Genet	2017	10.1038/ejhg.2017.82
28480134	Common and rare genetic markers of lipid variation in subjects with type 2 diabetes from the ACCORD clinical trial	Marvel SW	PeerJ	2017	10.7717/peerj.3187
28436151	Whole genome sequence association and ancestry-informed polygenic profile of EEG alpha in a Native American population	Peng Q	Am J Med Genet B Neuropsychiatr Genet	2017	10.1002/ajmg.b.32533
28425186	Association studies of low-frequency coding variants in nonsyndromic cleft lip with or without cleft palate	Leslie EJ	Am J Med Genet A	2017	10.1002/ajmg.a.38210
28417008	Association of gene coding variation and resting metabolic rate in a multi-ethnic sample of children and adults	Hellwege JN	BMC Obes	2017	10.1186/s40608-017-0145-5
28346466	Common and rare exonic MUC5B variants associated with type 2 diabetes in Han Chinese	Chen G	PLoS One	2017	10.1371/journal.pone.0173784
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