Hakon H Hakonarson

List of Publications by Year in Descending Order

648 papers	75,835 citations	128 h-index	267 g-index
700 ext. papers	92,705 ext. citations	10.3 avg, IF	7.29 L-index

#	Paper	IF	Citations
648	Under-specification as the source of ambiguity and vagueness in narrative phenotype algorithm definitions.. *BMC Medical Informatics and Decision Making*, **2022**, 22, 23	3.6	0
647	Genetic association of primary nonresponse to anti-TNF therapy in patients with inflammatory bowel disease. *Pharmacogenetics and Genomics*, **2022**, 32, 1-9	1.9	0
646	The reckoning: The return of genomic results to 1444 participants across the eMERGE3 Network.. *Genetics in Medicine*, **2022**,	8.1	2
645	A novel unbalanced translocation between chromosomes 5p and 18q leading to dysmorphology and global developmental delay.. *Molecular Genetics & Genomic Medicine*, **2022**, e1900	2.3	1
644	Expansion of Schizophrenia Gene Network Knowledge Using Machine Learning Selected Signals From Dorsolateral Prefrontal Cortex and Amygdala RNA-seq Data.. *Frontiers in Psychiatry*, **2022**, 13, 797329	5	0
643	CSF-1 maintains pathogenic but not homeostatic myeloid cells in the central nervous system during autoimmune neuroinflammation.. *Proceedings of the National Academy of Sciences of the United States of America*, **2022**, 119, e2111804119	11.5	0
642	Distinct diagnostic trajectories in NBAS-associated acute liver failure highlights the need for timely functional studies.. *JIMD Reports*, **2022**, 63, 240-249	1.9	
641	Ancestral diversity improves discovery and fine-mapping of genetic loci for anthropometric traits-The Hispanic/Latino Anthropometry Consortium.. *Human Genetics and Genomics Advances*, **2022**, 3, 100099	0.8	0
640	Maternal effect genes as risk factors for congenital heart defects.. *Human Genetics and Genomics Advances*, **2022**, 3, 100098	0.8	0
639	Manifestations of Alzheimer's disease genetic risk in the blood are evident in a multiomic analysis in healthy adults aged 18 to 90.. *Scientific Reports*, **2022**, 12, 6117	4.9	1
638	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases.. *JAMA Oncology*, **2022**,	13.4	2
637	Burden of rare coding variants reveals genetic heterogeneity between obese and non-obese asthma patients in the African American population.. *Respiratory Research*, **2022**, 23, 116	7.3	0
636	Saudi Arabian CML patient with a novel four-way translocation at t(9;22;5;2)(q34;q11.2;p13;q44).. *Molecular Genetics & Genomic Medicine*, **2022**, e1865	2.3	
635	Mendelian randomization analysis of plasma levels of CD209 and MICB proteins and the risk of varicose veins of lower extremities.. *PLoS ONE*, **2022**, 17, e0268725	3.7	
634	The power of genetic diversity in genome-wide association studies of lipids. *Nature*, **2021**,	50.4	24
633	Using common genetic variants to find drugs for common epilepsies.. *Brain Communications*, **2021**, 3, fcab287	4.5	0
632	Rare variant analysis in eczema identifies exonic variants in DUSP1, NOTCH4 and SLC9A4. *Nature Communications*, **2021**, 12, 6618	17.4	2

631	Rare neurological manifestations in a Saudi Arabian patient with Ehlers-Danlos syndrome and a novel homozygous variant in the TNXB gene. *American Journal of Medical Genetics, Part A*, **2021**, 188, 618	2.5	
630	NAC blocks Cystatin C amyloid complex aggregation in a cell system and in skin of HCCAA patients. *Nature Communications*, **2021**, 12, 1827	17.4	1
629	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1649-1665	2.5	0
628	Response to Li and Hopper. *American Journal of Human Genetics*, **2021**, 108, 527-529	11	1
627	Macrophages in SHH subgroup medulloblastoma display dynamic heterogeneity that varies with treatment modality. *Cell Reports*, **2021**, 34, 108917	10.6	7
626	Machine Learning Reduced Gene/Non-Coding RNA Features That Classify Schizophrenia Patients Accurately and Highlight Insightful Gene Clusters. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
625	Genome-wide association analysis in dilated cardiomyopathy reveals two new players in systolic heart failure on chromosomes 3p25.1 and 22q11.23. *European Heart Journal*, **2021**, 42, 2000-2011	9.5	14
624	Model-based deep embedding for constrained clustering analysis of single cell RNA-seq data. *Nature Communications*, **2021**, 12, 1873	17.4	7
623	Common Variation in Cytoskeletal Genes is Associated with Conotruncal Heart Defects. *Genes*, **2021**, 12,	4.2	1
622	Discovery and fine-mapping of height loci via high-density imputation of GWASs in individuals of African ancestry. *American Journal of Human Genetics*, **2021**, 108, 564-582	11	7
621	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. *Human Genetics*, **2021**, 140, 1061-1076	6.3	1
620	Inducible knockout of Clec16a in mice results in sensory neurodegeneration. *Scientific Reports*, **2021**, 11, 9319	4.9	2
619	JAK/STAT inhibitor therapy partially rescues the lipodystrophic autoimmune phenotype in Clec16a KO mice. *Scientific Reports*, **2021**, 11, 7372	4.9	1
618	New insights into hallux valgus by whole exome sequencing study. *Experimental Biology and Medicine*, **2021**, 246, 1607-1616	3.7	0
617	A new syndrome of moyamoya disease, kidney dysplasia, aminotransferase elevation, and skin disease associated with de novo variants in RNF213. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2168-2174	2.5	2
616	Elevation of Circulating LIGHT (TNFSF14) and Interleukin-18 Levels in Sepsis-Induced Multi-Organ Injuries **2021**,		1
615	RUNX-1 haploinsufficiency causes a marked deficiency of megakaryocyte-biased hematopoietic progenitor cells. *Blood*, **2021**, 137, 2662-2675	2.2	6
614	Serum levels of the IgA isotype switch factor TGF-β are elevated in patients with COVID-19. *FEBS Letters*, **2021**, 595, 1819-1824	3.8	4

613	ANKRD11 variants: KBG syndrome and beyond. *Clinical Genetics*, **2021**, 100, 187-200	4	4
612	Large trans-ethnic meta-analysis identifies AKR1C4 as a novel gene associated with age at menarche. *Human Reproduction*, **2021**, 36, 1999-2010	5.7	1
611	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. *Journal of Experimental Medicine*, **2021**, 218,	16.6	6
610	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. *American Journal of Human Genetics*, **2021**, 108, 965-982	11	6
609	Novel EDGE encoding method enhances ability to identify genetic interactions. *PLoS Genetics*, **2021** , 17, e1009534	6	0
608	Combined application of genetic and polygenic risk scores for type 1 diabetes risk prediction. *Diabetes, Obesity and Metabolism*, **2021**, 23, 2001-2003	6.7	
607	Genomic considerations for FHIR ; eMERGE implementation lessons. *Journal of Biomedical Informatics*, **2021**, 118, 103795	10.2	5
606	A Mendelian Randomization Approach Using 3-HMG-Coenzyme-A Reductase Gene Variation to Evaluate the Association of Statin-Induced Low-Density Lipoprotein Cholesterol Lowering With Noncardiovascular Disease Phenotypes. *JAMA Network Open*, **2021**, 4, e2112820	10.4	5
605	A unified framework identifies new links between plasma lipids and diseases from electronic medical records across large-scale cohorts. *Nature Genetics*, **2021**, 53, 972-981	36.3	2
604	Expanding the genetic landscape of oral-facial-digital syndrome with two novel genes. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2409-2416	2.5	1
603	Cleft palate morphology, genetic etiology, and risk of mortality in infants with Robin sequence. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3694-3700	2.5	
602	Deep learning prediction of attention-deficit hyperactivity disorder in African Americans by copy number variation. *Experimental Biology and Medicine*, **2021**, 246, 2317-2323	3.7	2
601	Neptune: an environment for the delivery of genomic medicine. *Genetics in Medicine*, **2021**, 23, 1838-1848	8.1	1
600	Shared genetic risk between eating disorder- and substance-use-related phenotypes: Evidence from genome-wide association studies. *Addiction Biology*, **2021**, 26, e12880	4.6	12
599	Mapping the 17q12-21.1 Locus for Variants Associated with Early-Onset Asthma in African Americans. *American Journal of Respiratory and Critical Care Medicine*, **2021**, 203, 424-436	10.2	5
598	Association of DLL1 with type 1 diabetes in patients characterized by low polygenic risk score. *Metabolism: Clinical and Experimental*, **2021**, 114, 154418	12.7	4
597	Risk of pre-eclampsia in patients with a maternal genetic predisposition to common medical conditions: a case-control study. *BJOG: an International Journal of Obstetrics and Gynaecology*, **2021**, 128, 55-65	3.7	3
596	Unsupervised modeling and genome-wide association identify novel features of allergic march trajectories. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 677-685.e10	11.5	6

595	De novo loss-of-function variants in X-linked MED12 are associated with Hardikar syndrome in females. *Genetics in Medicine*, **2021**, 23, 637-644	8.1	7
594	Novel Alzheimer Disease Risk Loci and Pathways in African American Individuals Using the African Genome Resources Panel: A Meta-analysis. *JAMA Neurology*, **2021**, 78, 102-113	17.2	32
593	Evaluation of the MC4R gene across eMERGE network identifies many unreported obesity-associated variants. *International Journal of Obesity*, **2021**, 45, 155-169	5.5	5
592	Loci identified by a genome-wide association study of carotid artery stenosis in the eMERGE network. *Genetic Epidemiology*, **2021**, 45, 4-15	2.6	5
591	Lossless integration of multiple electronic health records for identifying pleiotropy using summary statistics. *Nature Communications*, **2021**, 12, 168	17.4	1
590	Association of novel rare coding variants with juvenile idiopathic arthritis. *Annals of the Rheumatic Diseases*, **2021**, 80, 626-631	2.4	2
589	Genome-wide association study implicates novel loci and reveals candidate effector genes for longitudinal pediatric bone accrual. *Genome Biology*, **2021**, 22, 1	18.3	58
588	Expanded phenotypic spectrum of JAG1-associated diseases: Central conducting lymphatic anomaly with a pathogenic variant in JAG1. *Clinical Genetics*, **2021**, 99, 742-743	4	2
587	Integrative analysis of genome-wide association studies identifies novel loci associated with neuropsychiatric disorders. *Translational Psychiatry*, **2021**, 11, 69	8.6	5
586	Epigenetics in child psychiatry **2021**, 553-573		
585	A novel heterotaxy gene: Expansion of the phenotype of TTC21B-spectrum disease. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1266-1269	2.5	1
584	Rare Recurrent Variants in Noncoding Regions Impact Attention-Deficit Hyperactivity Disorder (ADHD) Gene Networks in Children of both African American and European American Ancestry. *Genes*, **2021**, 12,	4.2	2
583	Genome-wide association studies of low back pain and lumbar spinal disorders using electronic health record data identify a locus associated with lumbar spinal stenosis. *Pain*, **2021**, 162, 2263-2272	8	2
582	MONTAGE: a new tool for high-throughput detection of mosaic copy number variation. *BMC Genomics*, **2021**, 22, 133	4.5	0
581	Copy Number Variant Analysis and Genome-wide Association Study Identify Loci with Large Effect for Vesicoureteral Reflux. *Journal of the American Society of Nephrology: JASN*, **2021**,	12.7	5
580	Performance of model-based multifactor dimensionality reduction methods for epistasis detection by controlling population structure. *BioData Mining*, **2021**, 14, 16	4.3	1
579	Genetic architecture of type 1 diabetes with low genetic risk score informed by 41 unreported loci. *Communications Biology*, **2021**, 4, 908	6.7	1
578	Insights into non-autoimmune type 1 diabetes with 13 novel loci in low polygenic risk score patients. *Scientific Reports*, **2021**, 11, 16013	4.9	1

577	Common Genetic Variation And Age at Onset Of Anorexia Nervosa. *Biological Psychiatry Global Open Science*, **2021**,		3
576	Effect of micro-osteoperforations on the gene expression profile of the periodontal ligament of orthodontically moved human teeth. *Clinical Oral Investigations*, **2021**, 1	4.2	1
575	A genome-wide association meta-analysis identifies new eosinophilic esophagitis loci. *Journal of Allergy and Clinical Immunology*, **2021**,	11.5	1
574	HIF-1 Pulmonary Phenotype Wide Association Study Unveils a Link to Inflammatory Airway Conditions. *Frontiers in Genetics*, **2021**, 12, 756645	4.5	3
573	DeepCNV: a deep learning approach for authenticating copy number variations. *Briefings in Bioinformatics*, **2021**, 22,	13.4	1
572	Ciliopathies: Coloring outside of the lines. *American Journal of Medical Genetics, Part A*, **2021**, 185, 687-694	2.5	2
571	Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study.. *Circulation*, **2021**,	16.7	2
570	Mitochondrial DNA haplogroups and risk of attention deficit and hyperactivity disorder in European Americans. *Translational Psychiatry*, **2020**, 10, 370	8.6	0
569	COVID-19: Look to the Future, Learn from the Past. *Viruses*, **2020**, 12,	6.2	5
568	Lung Function in African American Children with Asthma Is Associated with Novel Regulatory Variants of the KIT Ligand and Gene-By-Air-Pollution Interaction. *Genetics*, **2020**, 215, 869-886	4	3
567	Variants in the Kisspeptin-GnRH Pathway Modulate the Hormonal Profile and Reproductive Outcomes. *DNA and Cell Biology*, **2020**, 39, 1012-1022	3.6	2
566	Distinct features of SARS-CoV-2-specific IgA response in COVID-19 patients. *European Respiratory Journal*, **2020**, 56,	13.6	192
565	The polygenic architecture of left ventricular mass mirrors the clinical epidemiology. *Scientific Reports*, **2020**, 10, 7561	4.9	3
564	Frequency of genomic secondary findings among 21,915 eMERGE network participants. *Genetics in Medicine*, **2020**, 22, 1470-1477	8.1	23
563	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17 458 subjects. *Brain*, **2020**, 143, 2106-2118	11.2	14
562	Predictive Utility of Polygenic Risk Scores for Coronary Heart Disease in Three Major Racial and Ethnic Groups. *American Journal of Human Genetics*, **2020**, 106, 707-716	11	37
561	Activating variants in PDGFRB result in a spectrum of disorders responsive to imatinib monotherapy. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1576-1591	2.5	11
560	Application of exome sequencing to diagnose a novel presentation of the Cornelia de Lange syndrome in an Afro-Caribbean family. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1318	2.3	2

559	Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects. *PLoS ONE*, **2020**, 15, e0234357	3.7	5
558	Neuroinflammation and EIF2 Signaling Persist despite Antiretroviral Treatment in an hiPSC Tri-culture Model of HIV Infection. *Stem Cell Reports*, **2020**, 14, 703-716	8	14
557	A homozygous truncating NALCN variant in two Afro-Caribbean siblings with hypotonia and dolichocephaly. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1877-1880	2.5	1
556	Detection of maternal X chromosome abnormalities using single nucleotide polymorphism-based noninvasive prenatal testing. *American Journal of Obstetrics & Gynecology MFM*, **2020**, 2, 100152	7.4	3
555	Mitochondrial DNA Haplogroups and Susceptibility to Neuroblastoma. *Journal of the National Cancer Institute*, **2020**, 112, 1259-1266	9.7	5
554	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. *Molecular Genetics and Metabolism*, **2020**, 130, 49-57	3.7	11
553	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. *Nature Communications*, **2020**, 11, 255	17.4	17
552	Type I IFN response associated with mTOR activation in the TAFRO subtype of idiopathic multicentric Castleman disease. *JCI Insight*, **2020**, 5,	9.9	13
551	Asthma and its relationship to mitochondrial copy number: Results from the Asthma Translational Genomics Collaborative (ATGC) of the Trans-Omics for Precision Medicine (TOPMed) program. *PLoS ONE*, **2020**, 15, e0242364	3.7	2
550	Kaposiform lymphangiomatosis effectively treated with MEK inhibition. *EMBO Molecular Medicine*, **2020**, 12, e12324	12	16
549	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. *Molecular Vision*, **2020**, 26, 216-225	2.3	2
548	Rare Genetic Variants of Large Effect Influence Risk of Type 1 Diabetes. *Diabetes*, **2020**, 69, 784-795	0.9	14
547	Genetic architecture study of rheumatoid arthritis and juvenile idiopathic arthritis. *PeerJ*, **2020**, 8, e8234	3.1	2
546	Intragenic Deletions of GNAS in Pseudohypoparathyroidism Type 1A Identify a New Region Affecting Methylation of Exon A/B. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	3
545	The Infection Rate of COVID-19 in Wuhan, China: Combined Analysis of Population Samples. *Journal of Medical Internet Research*, **2020**, 22, e20914	7.6	0
544	Interpretation of Maturity-Onset Diabetes of the Young Genetic Variants Based on American College of Medical Genetics and Genomics Criteria: Machine-Learning Model Development. *JMIR Biomedical Engineering*, **2020**, 5, e20506	1.3	0
543	TNFAIP8 controls murine intestinal stem cell homeostasis and regeneration by regulating microbiome-induced Akt signaling. *Nature Communications*, **2020**, 11, 2591	17.4	7
542	Genetic Underpinnings of Asthma and Related Traits **2020**, 341-360		

541	Evaluating sequence data quality from the Swift Accel-Amplicon CFTR Panel. *Scientific Data*, **2020**, 7, 8	8.2	0
540	A Polygenic and Phenotypic Risk Prediction for Polycystic Ovary Syndrome Evaluated by Phenome-Wide Association Studies. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	17
539	Are serum brain-derived neurotrophic factor concentrations related to brain structure and psychopathology in late childhood and early adolescence?. *CNS Spectrums*, **2020**, 25, 790-796	1.8	1
538	X-chromosome association studies of congenital heart defects. *American Journal of Medical Genetics, Part A*, **2020**, 182, 250-254	2.5	0
537	Genetic Discrimination Between LADA and Childhood-Onset Type 1 Diabetes Within the MHC. *Diabetes Care*, **2020**, 43, 418-425	14.6	15
536	Genetic Architecture of Abdominal Aortic Aneurysm in the Million Veteran Program. *Circulation*, **2020**, 142, 1633-1646	16.7	24
535	The Multi-Omics Architecture of Juvenile Idiopathic Arthritis. *Cells*, **2020**, 9,	7.9	7
534	A distinct GM-CSF T helper cell subset requires T-bet to adopt a T1 phenotype and promote neuroinflammation. *Science Immunology*, **2020**, 5,	28	7
533	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. *PLoS Genetics*, **2020**, 16, e1008718	6	25
532	European genetic ancestry associated with risk of childhood ependymoma. *Neuro-Oncology*, **2020**, 22, 1637-1646	1	5
531	Severe Lymphatic Disorder Resolved With MEK Inhibition in a Patient With Noonan Syndrome and SOS1 Mutation. *Pediatrics*, **2020**, 146,	7.4	11
530	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
529	Biliary-Atresia-Associated Mannosidase-1-Alpha-2 Gene Regulates Biliary and Ciliary Morphogenesis and Laterality. *Frontiers in Physiology*, **2020**, 11, 538701	4.6	4
528	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. *American Journal of Human Genetics*, **2020**, 107, 432-444	11	31
527	Participant choices for return of genomic results in the eMERGE Network. *Genetics in Medicine*, **2020**, 22, 1821-1829	8.1	14
526	Genetic predisposition to longer telomere length and risk of childhood, adolescent and adult-onset ependymoma. *Acta Neuropathologica Communications*, **2020**, 8, 173	7.3	9
525	Non-coding structural variation differentially impacts attention-deficit hyperactivity disorder (ADHD) gene networks in African American vs Caucasian children. *Scientific Reports*, **2020**, 10, 15252	4.9	4
524	Genomic risk scores for juvenile idiopathic arthritis and its subtypes. *Annals of the Rheumatic Diseases*, **2020**, 79, 1572-1579	2.4	3

523	Genetic Studies of Leptin Concentrations Implicate Leptin in the Regulation of Early Adiposity. *Diabetes*, **2020**, 69, 2806-2818	0.9	10
522	Neural crest-derived tumor neuroblastoma and melanoma share 1p13.2 as susceptibility locus that shows a long-range interaction with the SLC16A1 gene. *Carcinogenesis*, **2020**, 41, 284-295	4.6	11
521	Association of Genetic Risk of Obesity with Postoperative Complications Using Mendelian Randomization. *World Journal of Surgery*, **2020**, 44, 84-94	3.3	1
520	CNV Association of Diverse Clinical Phenotypes from eMERGE reveals novel disease biology underlying cardiovascular disease. *International Journal of Cardiology*, **2020**, 298, 107-113	3.2	2
519	A genome-wide association study of polycystic ovary syndrome identified from electronic health records. *American Journal of Obstetrics and Gynecology*, **2020**, 223, 559.e1-559.e21	6.4	20
518	Role of the ADCY9 gene in cardiac abnormalities of the Rubinstein-Taybi syndrome. *Orphanet Journal of Rare Diseases*, **2020**, 15, 101	4.2	1
517	Target Genes of Autism Risk Loci in Brain Frontal Cortex. *Frontiers in Genetics*, **2019**, 10, 707	4.5	8
516	Application of ACMG criteria to classify variants in the human gene mutation database. *Journal of Human Genetics*, **2019**, 64, 1091-1095	4.3	7
515	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. *Science Advances*, **2019**, 5, eaaw3095	14.3	39
514	Germline 16p11.2 Microdeletion Predisposes to Neuroblastoma. *American Journal of Human Genetics*, **2019**, 105, 658-668	11	12
513	Variants in the fetal genome near pro-inflammatory cytokine genes on 2q13 associate with gestational duration. *Nature Communications*, **2019**, 10, 3927	17.4	21
512	Association of Rare Recurrent Copy Number Variants With Congenital Heart Defects Based on Next-Generation Sequencing Data From Family Trios. *Frontiers in Genetics*, **2019**, 10, 819	4.5	6
511	Harmonizing Clinical Sequencing and Interpretation for the eMERGE III Network. *American Journal of Human Genetics*, **2019**, 105, 588-605	11	63
510	Multiple sclerosis genomic map implicates peripheral immune cells and microglia in susceptibility. *Science*, **2019**, 365,	33.3	309
509	The Autoimmune Disorder Susceptibility Gene Restrains NK Cell Function in YTS NK Cell Line and Knockout Mice. *Frontiers in Immunology*, **2019**, 10, 68	8.4	11
508	Association of Thyroid Function Genetic Predictors With Atrial Fibrillation: A Phenome-Wide Association Study and Inverse-Variance Weighted Average Meta-analysis. *JAMA Cardiology*, **2019**, 4, 136-143	16.3	20
507	Cross-disorder analysis of schizophrenia and 19 immune-mediated diseases identifies shared genetic risk. *Human Molecular Genetics*, **2019**, 28, 3498-3513	5.6	37
506	Copy number variations in individuals with conotruncal heart defects reveal some shared developmental pathways irrespective of 22q11.2 deletion status. *Birth Defects Research*, **2019**, 111, 888-905	2.8	2

505	Damaging Variants in Proangiogenic Genes Impair Growth in Fetuses with Cardiac Defects. *Journal of Pediatrics*, **2019**, 213, 103-109	3.6	11
504	Characterization of Rare Variants in MC4R in African American and Latino Children With Severe Early-Onset Obesity. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 2961-2970	5.6	12
503	Drug-resistant epilepsy classified by a phenotyping algorithm associates with NTRK2. *Acta Neurologica Scandinavica*, **2019**, 140, 169-176	3.8	4
502	Associations Between Attention-Deficit/Hyperactivity Disorder and Various Eating Disorders: A Swedish Nationwide Population Study Using Multiple Genetically Informative Approaches. *Biological Psychiatry*, **2019**, 86, 577-586	7.9	24
501	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. *Nature Genetics*, **2019**, 51, 804-814	36.3	181
500	Heritability and genome-wide association study of benign prostatic hyperplasia (BPH) in the eMERGE network. *Scientific Reports*, **2019**, 9, 6077	4.9	11
499	Multiple Epistasis Interactions Within MHC Are Associated With Ulcerative Colitis. *Frontiers in Genetics*, **2019**, 10, 257	4.5	6
498	Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. *Genetics in Medicine*, **2019**, 21, 2135-2144	8.1	13
497	Identification of Target Genes at Juvenile Idiopathic Arthritis GWAS Loci in Human Neutrophils. *Frontiers in Genetics*, **2019**, 10, 181	4.5	3
496	Microduplications at the 15q11.2 BP1-BP2 locus are enriched in patients with anorexia nervosa. *Journal of Psychiatric Research*, **2019**, 113, 34-38	5.2	6
495	Mutations in topoisomerase II result in a B cell immunodeficiency. *Nature Communications*, **2019**, 10, 3644	17.4	24
494	Effect of parental origin of damaging variants in pro-angiogenic genes on fetal growth in patients with congenital heart defects: Data and analyses. *Data in Brief*, **2019**, 25, 104311	1.2	2
493	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. *American Journal of Human Genetics*, **2019**, 105, 267-282	11	104
492	GWAS and enrichment analyses of non-alcoholic fatty liver disease identify new trait-associated genes and pathways across eMERGE Network. *BMC Medicine*, **2019**, 17, 135	11.4	53
491	Gene-based genome-wide association studies and meta-analyses of conotruncal heart defects. *PLoS ONE*, **2019**, 14, e0219926	3.7	5
490	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. *Nature Genetics*, **2019**, 51, 1207-1214	36.3	303
489	Genetic risk for Alzheimer's disease and functional brain connectivity in children and adolescents. *Neurobiology of Aging*, **2019**, 82, 10-17	5.6	7
488	A trans-ancestral meta-analysis of genome-wide association studies reveals loci associated with childhood obesity. *Human Molecular Genetics*, **2019**, 28, 3327-3338	5.6	30

487	Genetic comparison of sickle cell anaemia cohorts from Brazil and the United States reveals high levels of divergence. *Scientific Reports*, **2019**, 9, 10896	4.9	4
486	ARAF recurrent mutation causes central conducting lymphatic anomaly treatable with a MEK inhibitor. *Nature Medicine*, **2019**, 25, 1116-1122	50.5	70
485	Epistasis Detection in Genome-Wide Screening for Complex Human Diseases in Structured Populations. *Systems Medicine (New Rochelle, N Y)*, **2019**, 2, 19-27	1.6	5
484	Phenome-wide association analysis of LDL-cholesterol lowering genetic variants in PCSK9. *BMC Cardiovascular Disorders*, **2019**, 19, 240	2.3	8
483	The Genetic Contribution to Type 1 Diabetes. *Current Diabetes Reports*, **2019**, 19, 116	5.6	23
482	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
481	Detecting multiple differentially methylated CpG sites and regions related to dimensional psychopathology in youths. *Clinical Epigenetics*, **2019**, 11, 146	7.7	8
480	Detecting potential pleiotropy across cardiovascular and neurological diseases using univariate, bivariate, and multivariate methods on 43,870 individuals from the eMERGE network. *Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing*, **2019**, 24, 272-283	1.3	6
479	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
478	Candidate gene analyses for acute pain and morphine analgesia after pediatric day surgery: African American versus European Caucasian ancestry and dose prediction limits. *Pharmacogenomics Journal*, **2019**, 19, 570-581	3.5	9
477	Expanding the Genetic Landscape of Usher-Like Phenotypes **2019**, 60, 4701-4710		5
476	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. *Cell*, **2019**, 179, 1469-1482.e11	56.2	402
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474	Cryptic intronic NBAS variant reveals the genetic basis of recurrent liver failure in a child. *Molecular Genetics and Metabolism*, **2019**, 126, 77-82	3.7	11
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9	Discovery of the first genome-wide significant risk loci for ADHD		62
8	Genome-wide association study implicates novel loci and reveals candidate effector genes for longitudinal pediatric bone accrual through variant-to-gene mapping		5
7	Genome wide association analysis in dilated cardiomyopathy reveals two new key players in systolic heart failure on chromosome 3p25.1 and 22q11.23		1
6	Integrative Genetics Analysis of Juvenile Idiopathic Arthritis Identifies Novel Loci		1
5	Genetics of Low Polygenic Risk Score Type 1 Diabetes Patients: rare variants in 22 novel loci		1
4	Phenome-wide association studies (PheWAS) across large real-world data population cohorts support drug target validation		5
3	Identifying tissues implicated in Anorexia Nervosa using Transcriptomic Imputation		1
2	Phenome-wide association analysis of LDL-cholesterol lowering genetic variants in PCSK9		1

1 A cross-disorder dosage sensitivity map of the human genome 5