Thanh T Hoang

List of Publications by Citations

430 papers	22,276 citations	69 h-index	142 g-index
473 ext. papers	30,242 ext. citations	8.6 avg, IF	6.01 L-index

#	Paper	IF	Citations
430	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer disease. *Nature Genetics*, **2013**, 45, 1452-8	36.3	2714
429	Genetic meta-analysis of diagnosed Alzheimer disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
428	Discovery of the first genome-wide significant risk loci for attention deficit/hyperactivity disorder. *Nature Genetics*, **2019**, 51, 63-75	36.3	826
427	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
426	Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *American Journal of Human Genetics*, **2014**, 94, 677-94	11	635
425	De novo mutations in histone-modifying genes in congenital heart disease. *Nature*, **2013**, 498, 220-3	50.4	591
424	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer disease. *Nature Genetics*, **2017**, 49, 1373-1384	36.3	508
423	Inherited determinants of Crohn disease and ulcerative colitis phenotypes: a genetic association study. *Lancet, The*, **2016**, 387, 156-67	40	449
422	A genome-wide association study identifies KIAA0350 as a type 1 diabetes gene. *Nature*, **2007**, 448, 591-4	50.4	424
421	HMG-coenzyme A reductase inhibition, type 2 diabetes, and bodyweight: evidence from genetic analysis and randomised trials. *Lancet, The*, **2015**, 385, 351-61	40	409
420	Association between alcohol and cardiovascular disease: Mendelian randomisation analysis based on individual participant data. *BMJ, The*, **2014**, 349, g4164	5.9	406
419	A genome-wide association study identifies CDHR3 as a susceptibility locus for early childhood asthma with severe exacerbations. *Nature Genetics*, **2014**, 46, 51-5	36.3	376
418	Multi-ancestry genome-wide association study of 21,000 cases and 95,000 controls identifies new risk loci for atopic dermatitis. *Nature Genetics*, **2015**, 47, 1449-1456	36.3	329
417	Age group and sex differences in performance on a computerized neurocognitive battery in children age 8-21. *Neuropsychology*, **2012**, 26, 251-265	3.8	325
416	Low concordance of multiple variant-calling pipelines: practical implications for exome and genome sequencing. *Genome Medicine*, **2013**, 5, 28	14.4	315
415	Neuroimaging of the Philadelphia neurodevelopmental cohort. *NeuroImage*, **2014**, 86, 544-53	7.9	307
414	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

413	Genome-wide associations for birth weight and correlations with adult disease. *Nature*, **2016**, 538, 248-252	50.4	266
412	PCSK9 genetic variants and risk of type 2 diabetes: a mendelian randomisation study. *Lancet Diabetes and Endocrinology,the*, **2017**, 5, 97-105	18.1	225
411	Linked Sex Differences in Cognition and Functional Connectivity in Youth. *Cerebral Cortex*, **2015**, 25, 2383-94	5.1	209
410	Genetic predisposition to neuroblastoma mediated by a LMO1 super-enhancer polymorphism. *Nature*, **2015**, 528, 418-21	50.4	201
409	137 ancient human genomes from across the Eurasian steppes. *Nature*, **2018**, 557, 369-374	50.4	197
408	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. *Nature Biotechnology*, **2016**, 34, 531-8	44.5	196
407	Distinct features of SARS-CoV-2-specific IgA response in COVID-19 patients. *European Respiratory Journal*, **2020**, 56,	13.6	192
406	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. *Nature Genetics*, **2018**, 50, 26-41	36.3	186
405	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
404	The Philadelphia Neurodevelopmental Cohort: A publicly available resource for the study of normal and abnormal brain development in youth. *NeuroImage*, **2016**, 124, 1115-1119	7.9	173
403	Psychometric properties of the Penn Computerized Neurocognitive Battery. *Neuropsychology*, **2015**, 29, 235-46	3.8	169
402	Neurocognitive growth charting in psychosis spectrum youths. *JAMA Psychiatry*, **2014**, 71, 366-74	14.5	160
401	Increased frequency of de novo copy number variants in congenital heart disease by integrative analysis of single nucleotide polymorphism array and exome sequence data. *Circulation Research*, **2014**, 115, 884-896	15.7	158
400	Causal effects of body mass index on cardiometabolic traits and events: a Mendelian randomization analysis. *American Journal of Human Genetics*, **2014**, 94, 198-208	11	156
399	Age-Related Effects and Sex Differences in Gray Matter Density, Volume, Mass, and Cortical Thickness from Childhood to Young Adulthood. *Journal of Neuroscience*, **2017**, 37, 5065-5073	6.6	152
398	Genetic Evidence for Causal Relationships Between Maternal Obesity-Related Traits and Birth Weight. *JAMA - Journal of the American Medical Association*, **2016**, 315, 1129-40	27.4	149
397	Convergent genetic and expression data implicate immunity in Alzheimer's disease. *Alzheimer's and Dementia*, **2015**, 11, 658-71	1.2	146
396	Meta-analysis of shared genetic architecture across ten pediatric autoimmune diseases. *Nature Medicine*, **2015**, 21, 1018-27	50.5	143

395	The Philadelphia Neurodevelopmental Cohort: constructing a deep phenotyping collaborative. *Journal of Child Psychology and Psychiatry and Allied Disciplines*, **2015**, 56, 1356-1369	7.9	136
394	GWAS identifies four novel eosinophilic esophagitis loci. *Nature Communications*, **2014**, 5, 5593	17.4	135
393	Imaging patterns of brain development and their relationship to cognition. *Cerebral Cortex*, **2015**, 25, 1676-84	5.1	133
392	Gene-centric meta-analysis in 87,736 individuals of European ancestry identifies multiple blood-pressure-related loci. *American Journal of Human Genetics*, **2014**, 94, 349-60	11	131
391	Effects of multiple genetic loci on age at onset in late-onset Alzheimer disease: a genome-wide association study. *JAMA Neurology*, **2014**, 71, 1394-404	17.2	129
390	The impact of quality assurance assessment on diffusion tensor imaging outcomes in a large-scale population-based cohort. *NeuroImage*, **2016**, 125, 903-919	7.9	128
389	The role of TREM2 R47H as a risk factor for Alzheimer's disease, frontotemporal lobar degeneration, amyotrophic lateral sclerosis, and Parkinson's disease. *Alzheimer's and Dementia*, **2015**, 11, 1407-1416	1.2	126
388	Common and Dissociable Mechanisms of Executive System Dysfunction Across Psychiatric Disorders in Youth. *American Journal of Psychiatry*, **2016**, 173, 517-26	11.9	125
387	The diabetes susceptibility gene Clec16a regulates mitophagy. *Cell*, **2014**, 157, 1577-90	56.2	125
386	Impact of puberty on the evolution of cerebral perfusion during adolescence. *Proceedings of the National Academy of Sciences of the United States of America*, **2014**, 111, 8643-8	11.5	122
385	Directional dominance on stature and cognition in diverse human populations. *Nature*, **2015**, 523, 459-462	50.4	119
384	A novel susceptibility locus for type 1 diabetes on Chr12q13 identified by a genome-wide association study. *Diabetes*, **2008**, 57, 1143-6	0.9	118
383	Association of Arrhythmia-Related Genetic Variants With Phenotypes Documented in Electronic Medical Records. *JAMA - Journal of the American Medical Association*, **2016**, 315, 47-57	27.4	115
382	GRIN2D Recurrent De Novo Dominant Mutation Causes a Severe Epileptic Encephalopathy Treatable with NMDA Receptor Channel Blockers. *American Journal of Human Genetics*, **2016**, 99, 802-816	11	106
381	Meta-analysis identifies seven susceptibility loci involved in the atopic march. *Nature Communications*, **2015**, 6, 8804	17.4	105
380	HLA-DRB1*11 and variants of the MHC class II locus are strong risk factors for systemic juvenile idiopathic arthritis. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 15970-5	11.5	103
379	Causal Assessment of Serum Urate Levels in Cardiometabolic Diseases Through a Mendelian Randomization Study. *Journal of the American College of Cardiology*, **2016**, 67, 407-416	15.1	101
378	Investigating the Causal Relationship of C-Reactive Protein with 32 Complex Somatic and Psychiatric Outcomes: A Large-Scale Cross-Consortium Mendelian Randomization Study. *PLoS Medicine*, **2016**, 13, e1001976	11.6	100

377	Genome-wide association study of offspring birth weight in 86 577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. *Human Molecular Genetics*, **2018**, 27, 742-756	5.6	98
376	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nature Neuroscience*, **2017**, 20, 1043-1051	25.5	94
375	Common variants at 12q15 and 12q24 are associated with infant head circumference. *Nature Genetics*, **2012**, 44, 532-538	36.3	94
374	Gene-wide analysis detects two new susceptibility genes for Alzheimer's disease. *PLoS ONE*, **2014**, 9, e94661	3.7	90
373	Comprehensive analysis of gene expression in human retina and supporting tissues. *Human Molecular Genetics*, **2014**, 23, 4001-14	5.6	86
372	Genome-Wide Association Study Identifies African-Specific Susceptibility Loci in African Americans With Inflammatory Bowel Disease. *Gastroenterology*, **2017**, 152, 206-217.e2	13.3	85
371	Structural Brain Abnormalities in Youth With Psychosis Spectrum Symptoms. *JAMA Psychiatry*, **2016**, 73, 515-24	14.5	79
370	A novel common variant in DCST2 is associated with length in early life and height in adulthood. *Human Molecular Genetics*, **2015**, 24, 1155-68	5.6	77
369	A Pleiotropic Missense Variant in SLC39A8 Is Associated With Crohn's Disease and Human Gut Microbiome Composition. *Gastroenterology*, **2016**, 151, 724-32	13.3	77
368	Phenome-wide association studies across large population cohorts support drug target validation. *Nature Communications*, **2018**, 9, 4285	17.4	76
367	Inherited bone marrow failure associated with germline mutation of ACD, the gene encoding telomere protein TPP1. *Blood*, **2014**, 124, 2767-74	2.2	75
366	Genetic correlations among psychiatric and immune-related phenotypes based on genome-wide association data. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 641-657	3.5	75
365	A novel BHLHE41 variant is associated with short sleep and resistance to sleep deprivation in humans. *Sleep*, **2014**, 37, 1327-36	1.1	73
364	Stress and Bronchodilator Response in Children with Asthma. *American Journal of Respiratory and Critical Care Medicine*, **2015**, 192, 47-56	10.2	71
363	Sex differences in the effect of puberty on hippocampal morphology. *Journal of the American Academy of Child and Adolescent Psychiatry*, **2014**, 53, 341-50.e1	7.2	71
362	ARAF recurrent mutation causes central conducting lymphatic anomaly treatable with a MEK inhibitor. *Nature Medicine*, **2019**, 25, 1116-1122	50.5	70
361	Common genetic variants in NEFL influence gene expression and neuroblastoma risk. *Cancer Research*, **2014**, 74, 6913-24	10.1	69
360	Comorbidity of physical and mental disorders in the neurodevelopmental genomics cohort study. *Pediatrics*, **2015**, 135, e927-38	7.4	67

359	Functional neuroimaging abnormalities in youth with psychosis spectrum symptoms. *JAMA Psychiatry*, **2015**, 72, 456-65	14.5	66
358	Whole-genome DNA/RNA sequencing identifies truncating mutations in RBCK1 in a novel Mendelian disease with neuromuscular and cardiac involvement. *Genome Medicine*, **2013**, 5, 67	14.4	66
357	Exome-wide association study reveals novel susceptibility genes to sporadic dilated cardiomyopathy. *PLoS ONE*, **2017**, 12, e0172995	3.7	66
356	Whole-Genome Sequencing of Pharmacogenetic Drug Response in Racially Diverse Children with Asthma. *American Journal of Respiratory and Critical Care Medicine*, **2018**, 197, 1552-1564	10.2	65
355	Within-individual variability in neurocognitive performance: age- and sex-related differences in children and youths from ages 8 to 21. *Neuropsychology*, **2014**, 28, 506-18	3.8	64
354	Elevated levels of the IGF-binding protein protease MMP-1 in asthmatic airway smooth muscle. *American Journal of Respiratory Cell and Molecular Biology*, **1999**, 20, 199-208	5.7	62
353	Discovery and fine-mapping of adiposity loci using high density imputation of genome-wide association studies in individuals of African ancestry: African Ancestry Anthropometry Genetics Consortium. *PLoS Genetics*, **2017**, 13, e1006719	6	60
352	Identification of Four Novel Loci in Asthma in European American and African American Populations. *American Journal of Respiratory and Critical Care Medicine*, **2017**, 195, 456-463	10.2	59
351	Mutations in the ABCC6 gene as a cause of generalized arterial calcification of infancy: genotypic overlap with pseudoxanthoma elasticum. *Journal of Investigative Dermatology*, **2014**, 134, 658-665	4.3	58
350	Whole-genome sequencing in an autism multiplex family. *Molecular Autism*, **2013**, 4, 8	6.5	58
349	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
348	Thymic stromal lymphopoietin-mediated extramedullary hematopoiesis promotes allergic inflammation. *Immunity*, **2013**, 39, 1158-70	32.3	54
347	Association of CLEC16A with human common variable immunodeficiency disorder and role in murine B cells. *Nature Communications*, **2015**, 6, 6804	17.4	53
346	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
345	Relative contribution of type 1 and type 2 diabetes loci to the genetic etiology of adult-onset, non-insulin-requiring autoimmune diabetes. *BMC Medicine*, **2017**, 15, 88	11.4	52
344	GDF15 is a heart-derived hormone that regulates body growth. *EMBO Molecular Medicine*, **2017**, 9, 1150-1164	11.1	51
343	Genes involved in type 1 diabetes: an update. *Genes*, **2013**, 4, 499-521	4.2	49
342	Regulation of second messengers associated with airway smooth muscle contraction and relaxation. *American Journal of Respiratory and Critical Care Medicine*, **1998**, 158, S115-22	10.2	49

341	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. *Human Molecular Genetics*, **2015**, 24, 2297-307	5.6	48
340	CYP3A4 Induction by Rifampin: An Alternative Pathway for Vitamin D Inactivation in Patients With CYP24A1 Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 1440-1446	5.6	47
339	Genetic sharing and heritability of paediatric age of onset autoimmune diseases. *Nature Communications*, **2015**, 6, 8442	17.4	46
338	CYP3A4 mutation causes vitamin D-dependent rickets type 3. *Journal of Clinical Investigation*, **2018**, 128, 1913-1918	15.9	46
337	Fasoracetam in adolescents with ADHD and glutamatergic gene network variants disrupting mGluR neurotransmitter signaling. *Nature Communications*, **2018**, 9, 4	17.4	45
336	The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. *Nature Communications*, **2014**, 5, 4074	17.4	45
335	The Congenital Heart Disease Genetic Network Study: Cohort description. *PLoS ONE*, **2018**, 13, e0191319	3.7	43
334	Pathogenic variant in EPHB4 results in central conducting lymphatic anomaly. *Human Molecular Genetics*, **2018**, 27, 3233-3245	5.6	42
333	Gene expression and genetic variation in human atria. *Heart Rhythm*, **2014**, 11, 266-71	6.7	42
332	A trans-ethnic genome-wide association study identifies gender-specific loci influencing pediatric aBMD and BMC at the distal radius. *Human Molecular Genetics*, **2015**, 24, 5053-9	5.6	40
331	Associations of autozygosity with a broad range of human phenotypes. *Nature Communications*, **2019**, 10, 4957	17.4	40
330	Common variants upstream of MLF1 at 3q25 and within CPZ at 4p16 associated with neuroblastoma. *PLoS Genetics*, **2017**, 13, e1006787	6	40
329	A Frameshift in CSF2RB Predominant Among Ashkenazi Jews Increases Risk for Crohn's Disease and Reduces Monocyte Signaling via GM-CSF. *Gastroenterology*, **2016**, 151, 710-723.e2	13.3	40
328	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. *Science Advances*, **2019**, 5, eaaw3095	14.3	39
327	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. *Genome Medicine*, **2015**, 7, 90	14.4	38
326	CSF1R mosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. *Brain*, **2016**, 139, 1666-72	11.2	38
325	Cross-disorder analysis of schizophrenia and 19 immune-mediated diseases identifies shared genetic risk. *Human Molecular Genetics*, **2019**, 28, 3498-3513	5.6	37
324	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

323	Mutation in IRF2BP2 is responsible for a familial form of common variable immunodeficiency disorder. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 544-550.e4	11.5	37
322	Non-coding RNA dysregulation in the amygdala region of schizophrenia patients contributes to the pathogenesis of the disease. *Translational Psychiatry*, **2018**, 8, 44	8.6	36
321	Electronic Health Record Based Algorithm to Identify Patients with Autism Spectrum Disorder. *PLoS ONE*, **2016**, 11, e0159621	3.7	36
320	Practical considerations in genomic decision support: The eMERGE experience. *Journal of Pathology Informatics*, **2015**, 6, 50	4.4	36
319	Burden of potentially pathologic copy number variants is higher in children with isolated congenital heart disease and significantly impairs covariate-adjusted transplant-free survival. *Journal of Thoracic and Cardiovascular Surgery*, **2016**, 151, 1147-51.e4	1.5	35
318	Genome-wide mapping of IBD segments in an Ashkenazi PD cohort identifies associated haplotypes. *Human Molecular Genetics*, **2014**, 23, 4693-702	5.6	35
317	Mapping genomic loci implicates genes and synaptic biology in schizophrenia.. *Nature*, **2022**,	50.4	35
316	The Role of ARF6 in Biliary Atresia. *PLoS ONE*, **2015**, 10, e0138381	3.7	34
315	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. *Human Molecular Genetics*, **2018**, 27, 3305-3312	5.6	34
314	Association Between Mitochondrial DNA Haplogroup Variation and Autism Spectrum Disorders. *JAMA Psychiatry*, **2017**, 74, 1161-1168	14.5	33
313	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. *American Journal of Human Genetics*, **2016**, 98, 782-8	11	33
312	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants. *Genetics in Medicine*, **2019**, 21, 850-860	8.1	33
311	The eMERGE genotype set of 83,717 subjects imputed to ~40 million variants genome wide and association with the herpes zoster medical record phenotype. *Genetic Epidemiology*, **2019**, 43, 63-81	2.6	32
310	Rarity of the Alzheimer disease-protective APP A673T variant in the United States. *JAMA Neurology* , **2015**, 72, 209-16	17.2	31
309	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. *American Journal of Human Genetics*, **2020**, 107, 432-444	11	31
308	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2019**, 142, 50-58	11.2	31
307	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
306	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. *Clinical Epigenetics*, **2019**, 11, 64	7.7	29

305	Autocrine regulation of airway smooth muscle responsiveness. *Respiratory Physiology and Neurobiology*, **2003**, 137, 263-76	2.8	29
304	Exome Sequencing Reveals Mutations in AIRE as a Cause of Isolated Hypoparathyroidism. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 1726-1733	5.6	28
303	Identification of rare DNA sequence variants in high-risk autism families and their prevalence in a large case/control population. *Molecular Autism*, **2014**, 5, 5	6.5	28
302	Copy number variation meta-analysis reveals a novel duplication at 9p24 associated with multiple neurodevelopmental disorders. *Genome Medicine*, **2017**, 9, 106	14.4	27
301	Variants of the ACTG2 gene correlate with degree of severity and presence of megacystis in chronic intestinal pseudo-obstruction. *European Journal of Human Genetics*, **2016**, 24, 1211-5	5.3	27
300	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. *American Journal of Human Genetics*, **2017**, 101, 985-994	11	27
299	Fraction of exhaled nitric oxide values in childhood are associated with 17q11.2-q12 and 17q12-q21 variants. *Journal of Allergy and Clinical Immunology*, **2014**, 134, 46-55	11.5	27
298	Genome-wide association study for acute otitis media in children identifies FNDC1 as disease contributing gene. *Nature Communications*, **2016**, 7, 12792	17.4	26
297	Mutations in SPECC1L, encoding sperm antigen with calponin homology and coiled-coil domains 1-like, are found in some cases of autosomal dominant Opitz G/BBB syndrome. *Journal of Medical Genetics*, **2015**, 52, 104-10	5.8	26
296	Genome-wide association studies (GWAS): impact on elucidating the aetiology of diabetes. *Diabetes/Metabolism Research and Reviews*, **2011**, 27, 685-96	7.5	26
295	Autocrine cytokine signaling mediates effects of rhinovirus on airway responsiveness. *American Journal of Physiology - Lung Cellular and Molecular Physiology*, **2000**, 278, L1146-53	5.8	26
294	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. *PLoS Genetics*, **2020**, 16, e1008718	6	25
293	A Genomewide Association Study Identifies Two Sex-Specific Loci, at SPTB and IZUMO3, Influencing Pediatric Bone Mineral Density at Multiple Skeletal Sites. *Journal of Bone and Mineral Research*, **2017**, 32, 1274-1281	6.3	24
292	A genome-wide association meta-analysis of diarrhoeal disease in young children identifies FUT2 locus and provides plausible biological pathways. *Human Molecular Genetics*, **2016**, 25, 4127-4142	5.6	24
291	Mutations in topoisomerase IIresult in a B cell immunodeficiency. *Nature Communications*, **2019**, 10, 3644	17.4	24
290	The power of genetic diversity in genome-wide association studies of lipids. *Nature*, **2021**,	50.4	24
289	Genetic Architecture of Abdominal Aortic Aneurysm in the Million Veteran Program. *Circulation*, **2020**, 142, 1633-1646	16.7	24
288	A genome-wide association study identifies a susceptibility locus for biliary atresia on 2p16.1 within the gene EFEMP1. *PLoS Genetics*, **2018**, 14, e1007532	6	23

287	The Genetic Contribution to Type 1 Diabetes. *Current Diabetes Reports*, **2019**, 19, 116	5.6	23
286	Genetic variation in genes encoding airway epithelial potassium channels is associated with chronic rhinosinusitis in a pediatric population. *PLoS ONE*, **2014**, 9, e89329	3.7	23
285	Genome-Wide Association Studies and Meta-Analyses for Congenital Heart Defects. *Circulation: Cardiovascular Genetics*, **2017**, 10, e001449		22
284	Late-onset hereditary hypophosphatemic rickets with hypercalciuria (HHRH) due to mutation of SLC34A3/NPT2c. *Bone*, **2017**, 97, 15-19	4.7	22
283	DeepPolyA: A Convolutional Neural Network Approach for Polyadenylation Site Prediction. *IEEE Access*, **2018**, 6, 24340-24349	3.5	22
282	Patient genotypes impact survival after surgery for isolated congenital heart disease. *Annals of Thoracic Surgery*, **2014**, 98, 104-10; discussion 110-1	2.7	22
281	Variants in the fetal genome near pro-inflammatory cytokine genes on 2q13 associate with gestational duration. *Nature Communications*, **2019**, 10, 3927	17.4	21
280	Expanding the SPECC1L mutation phenotypic spectrum to include Teebi hypertelorism syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2497-502	2.5	21
279	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy. *Orphanet Journal of Rare Diseases*, **2014**, 9, 190	4.2	21
278	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
277	The Role of mGluR Copy Number Variation in Genetic and Environmental Forms of Syndromic Autism Spectrum Disorder. *Scientific Reports*, **2016**, 6, 19372	4.9	20
276	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. *American Journal of Human Genetics*, **2019**, 105, 283-301	11	20
275	Genome-wide association study of maternal and inherited loci for conotruncal heart defects. *PLoS ONE*, **2014**, 9, e96057	3.7	20
274	Association of Mutations in SLC12A1 Encoding the NKCC2 Cotransporter With Neonatal Primary Hyperparathyroidism. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 2196-200	5.6	20
273	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
272	Gene-Centric Analysis of Preeclampsia Identifies Maternal Association at. *Hypertension*, **2018**, 72, 408-418	8.5	20
271	Pathway analysis supports association of nonsyndromic cryptorchidism with genetic loci linked to cytoskeleton-dependent functions. *Human Reproduction*, **2015**, 30, 2439-51	5.7	19
270	Common variants in MMP20 at 11q22.2 predispose to 11q deletion and neuroblastoma risk. *Nature Communications*, **2017**, 8, 569	17.4	19

269	Complex management of a patient with a contiguous Xp11.4 gene deletion involving ornithine transcarbamylase: a role for detailed molecular analysis in complex presentations of classical diseases. *Molecular Genetics and Metabolism*, **2008**, 94, 498-502	3.7	19
268	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. *Brain*, **2020**, 143, 112-130	11.2	19
267	Common and Rare Genetic Risk Factors Converge in Protein Interaction Networks Underlying Schizophrenia. *Frontiers in Genetics*, **2018**, 9, 434	4.5	19
266	Genome-wide association study of maternal and inherited effects on left-sided cardiac malformations. *Human Molecular Genetics*, **2015**, 24, 265-73	5.6	18
265	A novel Rab27a mutation binds melanophilin, but not Munc13-4, causing immunodeficiency without albinism. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 599-601.e3	11.5	18
264	The Long Noncoding RNA Landscape in Amygdala Tissues from Schizophrenia Patients. *EBioMedicine*, **2018**, 34, 171-181	8.8	18
263	Mutations in cause autosomal recessive retinitis pigmentosa with intellectual disability. *Journal of Medical Genetics*, **2017**, 54, 698-704	5.8	18
262	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. *Nature Communications*, **2020**, 11, 255	17.4	17
261	Food allergen triggers are increased in children with the TSLP risk allele and eosinophilic esophagitis. *Clinical and Translational Gastroenterology*, **2018**, 9, 139	4.2	17
260	Combining targeted panel-based resequencing and copy-number variation analysis for the diagnosis of inherited syndromic retinopathies and associated ciliopathies. *Scientific Reports*, **2018**, 8, 5285	4.9	17
259	Mendelian randomization analysis demonstrates that low vitamin D is unlikely causative for pediatric asthma. *Journal of Allergy and Clinical Immunology*, **2016**, 138, 1747-1749.e4	11.5	17
258	Rare variants at 16p11.2 are associated with common variable immunodeficiency. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1569-77	11.5	17
257	The role of leptin, melanocortin, and neurotrophin system genes on body weight in anorexia nervosa and bulimia nervosa. *Journal of Psychiatric Research*, **2014**, 55, 77-86	5.2	17
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