Thanh T Hoang

List of Publications by Year
in descending order

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456 papers	36,150 citations	6606 79 h-index	5249 165 g-index
473 all docs	473 docs citations	473 times ranked	55257 citing authors

#	Article	IF	Citations
1	Meta-analysis of 74,046 individuals identifies 11 new susceptibility loci for Alzheimer's disease. Nature Genetics, 2013, 45, 1452-1458.	9.4	3,741
2	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates Aî², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	9.4	1,962
3	Discovery of the first genome-wide significant risk loci for attention deficit/hyperactivity disorder. Nature Genetics, 2019, 51, 63-75.	9.4	1,594
4	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
5	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	13.7	929
6	Convergence of Genes and Cellular Pathways Dysregulated in Autism Spectrum Disorders. American Journal of Human Genetics, 2014, 94, 677-694.	2.6	819
7	De novo mutations in histone-modifying genes in congenital heart disease. Nature, 2013, 498, 220-223.	13.7	798
8	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	9.4	783
9	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214.	9.4	641
10	Inherited determinants of Crohn's disease and ulcerative colitis phenotypes: a genetic association study. Lancet, The, 2016, 387, 156-167.	6.3	607
11	HMG-coenzyme A reductase inhibition, type 2 diabetes, and bodyweight: evidence from genetic analysis and randomised trials. Lancet, The, 2015, 385, 351-361.	6.3	562
12	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.	9.4	529
13	Association between alcohol and cardiovascular disease: Mendelian randomisation analysis based on individual participant data. BMJ, The, 2014, 349, g4164-g4164.	3.0	528
14	A genome-wide association study identifies KIAA0350 as a type 1 diabetes gene. Nature, 2007, 448, 591-594.	13.7	497
15	A genome-wide association study identifies CDHR3 as a susceptibility locus for early childhood asthma with severe exacerbations. Nature Genetics, 2014, 46, 51-55.	9.4	497
16	Neuroimaging of the Philadelphia Neurodevelopmental Cohort. NeuroImage, 2014, 86, 544-553.	2.1	452
17	Age group and sex differences in performance on a computerized neurocognitive battery in children age 8â˜21.. Neuropsychology, 2012, 26, 251-265.	1.0	432
18	Genome-wide associations for birth weight and correlations with adult disease. Nature, 2016, 538, 248-252.	13.7	406

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19	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. Nature Genetics, 2019, 51, 804-814.	9.4	402
20	Low concordance of multiple variant-calling pipelines: practical implications for exome and genome sequencing. Genome Medicine, 2013, 5, 28.	3.6	381
21	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	13.7	353
22	137 ancient human genomes from across the Eurasian steppes. Nature, 2018, 557, 369-374.	13.7	325
23	Linked Sex Differences in Cognition and Functional Connectivity in Youth. Cerebral Cortex, 2015, 25, 2383-2394.	1.6	302
24	PCSK9 genetic variants and risk of type 2 diabetes: a mendelian randomisation study. Lancet Diabetes and Endocrinology,the, 2017, 5, 97-105.	5.5	298
25	Distinct features of SARS-CoV-2-specific IgA response in COVID-19 patients. European Respiratory Journal, 2020, 56, 2001526.	3.1	292
26	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	9.4	286
27	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. Nature Biotechnology, 2016, 34, 531-538.	9.4	273
28	Psychometric properties of the Penn Computerized Neurocognitive Battery.. Neuropsychology, 2015, 29, 235-246.	1.0	272
29	The Philadelphia Neurodevelopmental Cohort: A publicly available resource for the study of normal and abnormal brain development in youth. NeuroImage, 2016, 124, 1115-1119.	2.1	268
30	Genetic predisposition to neuroblastoma mediated by a LMO1 super-enhancer polymorphism. Nature, 2015, 528, 418-421.	13.7	263
31	Genome-wide association study implicates novel loci and reveals candidate effector genes for longitudinal pediatric bone accrual. Genome Biology, 2021, 22, 1.	3.8	239
32	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.	1.7	235
33	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.	2.0	229
34	Genetic Evidence for Causal Relationships Between Maternal Obesity-Related Traits and Birth Weight. JAMA - Journal of the American Medical Association, 2016, 315, 1129.	3.8	220
35	Meta-analysis of shared genetic architecture across ten pediatric autoimmune diseases. Nature Medicine, 2015, 21, 1018-1027.	15.2	212
36	The Philadelphia Neurodevelopmental Cohort: constructing a deep phenotyping collaborative. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2015, 56, 1356-1369.	3.1	208

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37	Neurocognitive Growth Charting in Psychosis Spectrum Youths. JAMA Psychiatry, 2014, 71, 366.	6.0	206
38	The impact of quality assurance assessment on diffusion tensor imaging outcomes in a large-scale population-based cohort. NeuroImage, 2016, 125, 903-919.	2.1	202
39	Causal Effects of Body Mass Index on Cardiometabolic Traits and Events: A Mendelian Randomization Analysis. American Journal of Human Genetics, 2014, 94, 198-208.	2.6	199
40	Imaging Patterns of Brain Development and their Relationship to Cognition. Cerebral Cortex, 2015, 25, 1676-1684.	1.6	196
41	Common and Dissociable Mechanisms of Executive System Dysfunction Across Psychiatric Disorders in Youth. American Journal of Psychiatry, 2016, 173, 517-526.	4.0	191
42	GWAS identifies four novel eosinophilic esophagitis loci. Nature Communications, 2014, 5, 5593.	5.8	181
43	Directional dominance on stature and cognition inÂdiverse human populations. Nature, 2015, 523, 459-462.	13.7	173
44	Convergent genetic and expression data implicate immunity in Alzheimer's disease. Alzheimer's and Dementia, 2015, 11, 658-671.	0.4	173
45	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, 8643-8648.	3.3	169
46	Effects of Multiple Genetic Loci on Age at Onset in Late-Onset Alzheimer Disease. JAMA Neurology, 2014, 71, 1394.	4.5	166
47	The Diabetes Susceptibility Gene Clec16a Regulates Mitophagy. Cell, 2014, 157, 1577-1590.	13.5	166
48	Gene-centric Meta-analysis in 87,736 Individuals of European Ancestry Identifies Multiple Blood-Pressure-Related Loci. American Journal of Human Genetics, 2014, 94, 349-360.	2.6	158
49	Genetic correlations among psychiatric and immuneâ€related phenotypes based on genomeâ€wide association data. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 641-657.	1.1	158
50	Genome-wide association study of offspring birth weight in 86â€‰577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. Human Molecular Genetics, 2018, 27, 742-756.	1.4	156
51	Gene-Wide Analysis Detects Two New Susceptibility Genes for Alzheimer's Disease. PLoS ONE, 2014, 9, e94661.	1.1	155
52	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.	0.4	152
53	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	7.1	152
54	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.	3.9	150

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55	Meta-analysis identifies seven susceptibility loci involved in the atopic march. Nature Communications, 2015, 6, 8804.	5.8	148
56	Association of Arrhythmia-Related Genetic Variants With Phenotypes Documented in Electronic Medical Records. JAMA - Journal of the American Medical Association, 2016, 315, 47.	3.8	148
57	HLA-DRB1*11and 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, 15970-15975.	3.3	139
58	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.	2.6	138
59	Causal Assessment of Serum Urate Levels inÂCardiometabolic Diseases Through a Mendelian Randomization Study. Journal of the American College of Cardiology, 2016, 67, 407-416.	1.2	138
60	A Novel Susceptibility Locus for Type 1 Diabetes on Chr12q13 Identified by a Genome-Wide Association Study. Diabetes, 2008, 57, 1143-1146.	0.3	137
61	ARAF recurrent mutation causes central conducting lymphatic anomaly treatable with a MEK inhibitor. Nature Medicine, 2019, 25, 1116-1122.	15.2	136
62	Phenome-wide association studies across large population cohorts support drug target validation. Nature Communications, 2018, 9, 4285.	5.8	134
63	Common variants at 12q15 and 12q24 are associated with infant head circumference. Nature Genetics, 2012, 44, 532-538.	9.4	130
64	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. American Journal of Human Genetics, 2020, 107, 432-444.	2.6	124
65	Genome-Wide Association Study Identifies African-Specific Susceptibility Loci in African Americans With Inflammatory Bowel Disease. Gastroenterology, 2017, 152, 206-217.e2.	0.6	120
66	Structural Brain Abnormalities in Youth With Psychosis Spectrum Symptoms. JAMA Psychiatry, 2016, 73, 515.	6.0	116
67	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	0.7	114
68	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.	2.3	110
69	Comprehensive analysis of gene expression in human retina and supporting tissues. Human Molecular Genetics, 2014, 23, 4001-4014.	1.4	109
70	A novel common variant in DCST2 is associated with length in early life and height in adulthood. Human Molecular Genetics, 2015, 24, 1155-1168.	1.4	109
71	A Pleiotropic Missense Variant in SLC39A8 Is Associated With Crohnâ€™s Disease and Human Gut Microbiome Composition. Gastroenterology, 2016, 151, 724-732.	0.6	109
72	A NovelBHLHE41Variant is Associated with Short Sleep and Resistance to Sleep Deprivation in Humans. Sleep, 2014, 37, 1327-1336.	0.6	104

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73	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.	2.5	102
74	Functional Neuroimaging Abnormalities in Youth With Psychosis Spectrum Symptoms. JAMA Psychiatry, 2015, 72, 456.	6.0	100
75	Stress and Bronchodilator Response in Children with Asthma. American Journal of Respiratory and Critical Care Medicine, 2015, 192, 47-56.	2.5	99
76	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.	1.5	98
77	Inherited bone marrow failure associated with germline mutation of ACD, the gene encoding telomere protein TPP1. Blood, 2014, 124, 2767-2774.	0.6	97
78	Comorbidity of Physical and Mental Disorders in the Neurodevelopmental Genomics Cohort Study. Pediatrics, 2015, 135, e927-e938.	1.0	96
79	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. PLoS Genetics, 2020, 16, e1008718.	1.5	95
80	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.	2.6	93
81	Exome-wide association study reveals novel susceptibility genes to sporadic dilated cardiomyopathy. PLoS ONE, 2017, 12, e0172995.	1.1	92
82	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.	2.5	91
83	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.	3.6	87
84	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. Science Advances, 2019, 5, eaaw3095.	4.7	86
85	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	5.8	84
86	Sex Differences in the Effect of Puberty on Hippocampal Morphology. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 341-350.e1.	0.3	83
87	Within-individual variability in neurocognitive performance: Age- and sex-related differences in children and youths from ages 8 to 21.. Neuropsychology, 2014, 28, 506-518.	1.0	82
88	The Congenital Heart Disease Genetic Network Study: Cohort description. PLoS ONE, 2018, 13, e0191319.	1.1	82
89	Genetic Architecture of Abdominal Aortic Aneurysm in the Million Veteran Program. Circulation, 2020, 142, 1633-1646.	1.6	78
90	CYP3A4 mutation causes vitamin Dâ€“dependent rickets type 3. Journal of Clinical Investigation, 2018, 128, 1913-1918.	3.9	77

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91	Whole-genome sequencing in an autism multiplex family. Molecular Autism, 2013, 4, 8.	2.6	76
92	A trans-ancestral meta-analysis of genome-wide association studies reveals loci associated with childhood obesity. Human Molecular Genetics, 2019, 28, 3327-3338.	1.4	76
93	Common Genetic Variants in NEFL Influence Gene Expression and Neuroblastoma Risk. Cancer Research, 2014, 74, 6913-6924.	0.4	74
94	Fasoracetam in adolescents with ADHD and glutamatergic gene network variants disrupting mGluR neurotransmitter signaling. Nature Communications, 2018, 9, 4.	5.8	74
95	Pathogenic variant in EPHB4 results in central conducting lymphatic anomaly. Human Molecular Genetics, 2018, 27, 3233-3245.	1.4	73
96	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.	1.8	72
97	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	1.8	71
98	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.	0.3	70
99	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.	1.4	69
100	<scp>GDF</scp>15 is a heartâ€derived hormone that regulates body growth. EMBO Molecular Medicine, 2017, 9, 1150-1164.	3.3	69
101	Rare Genetic Variants of Large Effect Influence Risk of Type 1 Diabetes. Diabetes, 2020, 69, 784-795.	0.3	69
102	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	1.1	68
103	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.	2.3	67
104	The Role of ARF6 in Biliary Atresia. PLoS ONE, 2015, 10, e0138381.	1.1	66
105	Cross-disorder analysis of schizophrenia and 19 immune-mediated diseases identifies shared genetic risk. Human Molecular Genetics, 2019, 28, 3498-3513.	1.4	65
106	Thymic Stromal Lymphopoietin-Mediated Extramedullary Hematopoiesis Promotes Allergic Inflammation. Immunity, 2013, 39, 1158-1170.	6.6	64
107	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. Human Molecular Genetics, 2015, 24, 2297-2307.	1.4	64
108	Association of CLEC16A with human common variable immunodeficiency disorder and role in murine B cells. Nature Communications, 2015, 6, 6804.	5.8	63

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109	The eMERGE genotype set of 83,717 subjects imputed to ~40â€‰million variants genome wide and association with the herpes zoster medical record phenotype. Genetic Epidemiology, 2019, 43, 63-81.	0.6	63
110	Common variants upstream of MLF1 at 3q25 and within CPZ at 4p16 associated with neuroblastoma. PLoS Genetics, 2017, 13, e1006787.	1.5	62
111	Genes Involved in Type 1 Diabetes: An Update. Genes, 2013, 4, 499-521.	1.0	61
112	Electronic Health Record Based Algorithm to Identify Patients with Autism Spectrum Disorder. PLoS ONE, 2016, 11, e0159621.	1.1	59
113	Regulation of Second Messengers Associated with Airway Smooth Muscle Contraction and Relaxation. American Journal of Respiratory and Critical Care Medicine, 1998, 158, S115-S122.	2.5	58
114	Genetic sharing and heritability of paediatric age of onset autoimmune diseases. Nature Communications, 2015, 6, 8442.	5.8	58
115	Association Between Mitochondrial DNA Haplogroup Variation and Autism Spectrum Disorders. JAMA Psychiatry, 2017, 74, 1161.	6.0	57
116	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, 1147-1151.e4.	0.4	55
117	Non-coding RNA dysregulation in the amygdala region of schizophrenia patients contributes to the pathogenesis of the disease. Translational Psychiatry, 2018, 8, 44.	2.4	55
118	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.	1.5	54
119	CSF1Rmosaicism in a family with hereditary diffuse leukoencephalopathy with spheroids. Brain, 2016, 139, 1666-1672.	3.7	53
120	The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. Nature Communications, 2014, 5, 4074.	5.8	52
121	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.	0.6	51
122	A genome-wide association study identifies a susceptibility locus for biliary atresia on 2p16.1 within the gene EFEMP1. PLoS Genetics, 2018, 14, e1007532.	1.5	51
123	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	3.7	51
124	Kaposiform lymphangiomatosis effectively treated with <scp>MEK</scp> inhibition. EMBO Molecular Medicine, 2020, 12, e12324.	3.3	51
125	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, 782-788.	2.6	50
126	Genome-wide association study for acute otitis media in children identifies FNDC1 as disease contributing gene. Nature Communications, 2016, 7, 12792.	5.8	50

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127	Association of Neighborhood Deprivation With Epigenetic Aging Using 4 Clock Metrics. JAMA Network Open, 2020, 3, e2024329.	2.8	50
128	Genome-wide mapping of IBD segments in an Ashkenazi PD cohort identifies associated haplotypes. Human Molecular Genetics, 2014, 23, 4693-4702.	1.4	49
129	Concept and design of a genome-wide association genotyping array tailored for transplantation-specific studies. Genome Medicine, 2015, 7, 90.	3.6	49
130	Variants in the fetal genome near pro-inflammatory cytokine genes on 2q13 associate with gestational duration. Nature Communications, 2019, 10, 3927.	5.8	49
131	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.	0.7	49
132	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.	1.0	49
133	Gene expression and genetic variation in human atria. Heart Rhythm, 2014, 11, 266-271.	0.3	48
134	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, 5053-5059.	1.4	48
135	The Genetic Contribution to Type 1 Diabetes. Current Diabetes Reports, 2019, 19, 116.	1.7	48
136	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. Nature Communications, 2020, 11, 255.	5.8	48
137	Genome-Wide Association Studies and Meta-Analyses for Congenital Heart Defects. Circulation: Cardiovascular Genetics, 2017, 10, e001449.	5.1	47
138	Gene-Centric Analysis of Preeclampsia Identifies Maternal Association at PLEKHG1. Hypertension, 2018, 72, 408-416.	1.3	46
139	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	2.6	46
140	Model-based deep embedding for constrained clustering analysis of single cell RNA-seq data. Nature Communications, 2021, 12, 1873.	5.8	46
141	USMG5 Ashkenazi Jewish founder mutation impairs mitochondrial complex V dimerization and ATP synthesis. Human Molecular Genetics, 2018, 27, 3305-3312.	1.4	45
142	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.	2.6	44
143	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, 1211-1215.	1.4	43
144	Neuroinflammation and EIF2 Signaling Persist despite Antiretroviral Treatment in an hiPSC Tri-culture Model of HIV Infection. Stem Cell Reports, 2020, 14, 703-716.	2.3	42

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145	Practical considerations in genomic decision support: The eMERGE experience. Journal of Pathology Informatics, 2015, 6, 50.	0.8	42
146	Rarity of the Alzheimer Diseaseâ€“ProtectiveAPPA673T Variant in the United States. JAMA Neurology, 2015, 72, 209.	4.5	41
147	Copy number variation meta-analysis reveals a novel duplication at 9p24 associated with multiple neurodevelopmental disorders. Genome Medicine, 2017, 9, 106.	3.6	41
148	Mutations inSPECC1L, 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, 104-110.	1.5	40
149	A Polygenic and Phenotypic Risk Prediction for Polycystic Ovary Syndrome Evaluated by Phenome-Wide Association Studies. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1918-1936.	1.8	40
150	Epigenome-wide association study of DNA methylation and adult asthma in the Agricultural Lung Health Study. European Respiratory Journal, 2020, 56, 2000217.	3.1	40
151	Integrative analysis of genome-wide association studies identifies novel loci associated with neuropsychiatric disorders. Translational Psychiatry, 2021, 11, 69.	2.4	39
152	Mutations in topoisomerase IIÎ² result in a B cell immunodeficiency. Nature Communications, 2019, 10, 3644.	5.8	37
153	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.	2.6	36
154	Autocrine cytokine signaling mediates effects of rhinovirus on airway responsiveness. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2000, 278, L1146-L1153.	1.3	35
155	A genome-wide association meta-analysis of diarrhoeal disease in young children identifiesFUT2locus and provides plausible biological pathways. Human Molecular Genetics, 2016, 25, 4127-4142.	1.4	35
156	Exome Sequencing Reveals Mutations in AIRE as a Cause of Isolated Hypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 1726-1733.	1.8	35
157	Type I IFN response associated with mTOR activation in the TAFRO subtype of idiopathic multicentric Castleman disease. JCI Insight, 2020, 5, .	2.3	35
158	Cell-free DNA screening for prenatal detection of 22q11.2 deletion syndrome. American Journal of Obstetrics and Gynecology, 2022, 227, 79.e1-79.e11.	0.7	35
159	Autocrine regulation of airway smooth muscle responsiveness. Respiratory Physiology and Neurobiology, 2003, 137, 263-276.	0.7	34
160	DeepPolyA: A Convolutional Neural Network Approach for Polyadenylation Site Prediction. IEEE Access, 2018, 6, 24340-24349.	2.6	34
161	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	0.7	34
162	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.	1.5	33

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163	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	3.7	33
164	A distinct GM-CSF ⁺ T helper cell subset requires T-bet to adopt a T _H 1 phenotype and promote neuroinflammation. Science Immunology, 2020, 5, .	5.6	33
165	The Long Noncoding RNA Landscape in Amygdala Tissues from Schizophrenia Patients. EBioMedicine, 2018, 34, 171-181.	2.7	32
166	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy. Orphanet Journal of Rare Diseases, 2014, 9, 190.	1.2	31
167	Germline 16p11.2 Microdeletion Predisposes to Neuroblastoma. American Journal of Human Genetics, 2019, 105, 658-668.	2.6	31
168	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. Journal of Experimental Medicine, 2021, 218, .	4.2	31
169	Patient Genotypes Impact Survival After Surgery for Isolated Congenital Heart Disease. Annals of Thoracic Surgery, 2014, 98, 104-111.	0.7	30
170	A Genomewide Association Study Identifies Two Sexâ€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.	3.1	30
171	Late-onset hereditary hypophosphatemic rickets with hypercalciuria (HHRH) due to mutation of SLC34A3/NPT2c. Bone, 2017, 97, 15-19.	1.4	30
172	Cell-free DNA screening for trisomies 21, 18, and 13 in pregnancies at low and high risk for aneuploidy with genetic confirmation. American Journal of Obstetrics and Gynecology, 2022, 227, 259.e1-259.e14.	0.7	30
173	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.	1.5	28
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