Thanh T Hoang

List of Publications by Year
in descending order

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Version: 2024-02-01

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	Treatment of severe Kaposiform lymphangiomatosis positive for NRAS mutation by MEK inhibition. Pediatric Research, 2023, 94, 1911-1915.	1.1	16
2	Genetic association of primary nonresponse to anti-TNFÎ± therapy in patients with inflammatory bowel disease. Pharmacogenetics and Genomics, 2022, 32, 1-9.	0.7	2
3	Effect of micro-osteoperforations on the gene expression profile of the periodontal ligament of orthodontically moved human teeth. Clinical Oral Investigations, 2022, 26, 1985-1996.	1.4	4
4	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. Biological Psychiatry, 2022, 91, 313-327.	0.7	114
5	A genome-wide association meta-analysis identifies new eosinophilic esophagitis loci. Journal of Allergy and Clinical Immunology, 2022, 149, 988-998.	1.5	19
6	Rare neurological manifestations in a Saudi Arabian patient with <scp>Ehlersâ€“Danlos</scp> syndrome and a novel homozygous variant in the <scp>TNXB</scp> gene. American Journal of Medical Genetics, Part A, 2022, 188, 618-623.	0.7	1
7	A novel <scp>MBTPS2</scp> variant associated with <scp>BRESHECK</scp> syndrome impairs <scp>sterolâ€regulated</scp> transcription and the endoplasmic reticulum stress response. American Journal of Medical Genetics, Part A, 2022, 188, 463-472.	0.7	4
8	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	1.1	0
9	Variants in ADD1 cause intellectual disability, corpus callosum dysgenesis, and ventriculomegaly in humans. Genetics in Medicine, 2022, 24, 319-331.	1.1	6
10	Application of deep learning algorithm on whole genome sequencing data uncovers structural variants associated with multiple mental disorders in African American patients. Molecular Psychiatry, 2022, 27, 1469-1478.	4.1	13
11	Identification of Mitochondrial DNA Variants Associated With Risk of Neuroblastoma. Journal of the National Cancer Institute, 2022, 114, 910-913.	3.0	4
12	Under-specification as the source of ambiguity and vagueness in narrative phenotype algorithm definitions. BMC Medical Informatics and Decision Making, 2022, 22, 23.	1.5	1
13	Circulating LIGHT (TNFSF14) and Interleukin-18 Levels in Sepsis-Induced Multi-Organ Injuries. Biomedicines, 2022, 10, 264.	1.4	7
14	Mendelian randomization study of obesity and type 2 diabetes in hospitalized COVID-19 patients. Metabolism: Clinical and Experimental, 2022, 129, 155156.	1.5	17
15	Improved genetic risk scoring algorithm for type 1 diabetes prediction. Pediatric Diabetes, 2022, 23, 320-323.	1.2	11
16	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
17	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
18	Genetic analysis in African American children supports ancestry specific neuroblastoma susceptibility. Cancer Epidemiology Biomarkers and Prevention, 2022, , cebp.EPI-21-0782-A.2021.	1.1	1

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19	The reckoning: The return of genomic results to 1444 participants across the eMERGE3 Network. Genetics in Medicine, 2022, 24, 1130-1138.	1.1	12
20	A novel unbalanced translocation between chromosomes 5p and 18q leading to dysmorphology and global developmental delay. Molecular Genetics & Genomic Medicine, 2022, , e1900.	0.6	1
21	Genetic analysis for type 1 diabetes genes in juvenile dermatomyositis unveils genetic disease overlap. Rheumatology, 2022, , .	0.9	2
22	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.0	5
23	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.	1.3	9
24	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	1.1	5
25	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.	3.3	10
26	Identification of Novel Loci Shared by Juvenile Idiopathic Arthritis Subtypes Through Integrative Genetic Analysis. Arthritis and Rheumatology, 2022, 74, 1420-1429.	2.9	4
27	Exome and <scp>RNAâ€Seq</scp> analyses of an incomplete penetrance variant in <scp> USP9X </scp> in femaleâ€specific syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2022, , .	0.7	1
28	Distinct diagnostic trajectories in <scp>NBAS</scp>â€associated acute liver failure highlights the need for timely functional studies. JIMD Reports, 2022, 63, 240-249.	0.7	2
29	Ancestral diversity improves discovery and fine-mapping of genetic loci for anthropometric traitsâ€"The Hispanic/Latino Anthropometry Consortium. Human Genetics and Genomics Advances, 2022, 3, 100099.	1.0	3
30	Maternal effect genes as risk factors for congenital heart defects. Human Genetics and Genomics Advances, 2022, 3, 100098.	1.0	2
31	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	13.7	929
32	Further supporting <scp>SMARCC2</scp>â€related neurodevelopmental disorder through exome analysis and reanalysis in two patients. American Journal of Medical Genetics, Part A, 2022, 188, 878-882.	0.7	3
33	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncology, 2022, 8, 835.	3.4	25
34	Pulmonary Function and Blood DNA Methylation: A Multiancestry Epigenome-Wide Association Meta-analysis. American Journal of Respiratory and Critical Care Medicine, 2022, 206, 321-336.	2.5	15
35	Copy Number Variant Risk Scores Associated With Cognition, Psychopathology, and Brain Structure in Youths in the Philadelphia Neurodevelopmental Cohort. JAMA Psychiatry, 2022, 79, 699.	6.0	8
36	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.	1.4	1

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37	Saudi Arabian CML patient with a novel fourâ€way translocation at t(9;22;5;2)(q34;q11.2;p13;q44). Molecular Genetics & Genomic Medicine, 2022, , e1865.	0.6	1
38	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.	1.1	1
39	Genetics etiologies and genotype phenotype correlations in a cohort of individuals with central conducting lymphatic anomaly. European Journal of Human Genetics, 2022, 30, 1022-1028.	1.4	9
40	Mutation burden analysis of six common mental disorders in African Americans by whole genome sequencing. Human Molecular Genetics, 2022, 31, 3769-3776.	1.4	4
41	Shared genetic risk between eating disorderâ€and substanceâ€useâ€related phenotypes: Evidence from genomeâ€wide association studies. Addiction Biology, 2021, 26, e12880.	1.4	28
42	Mapping the 17q12â€“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.	2.5	16
43	Association of DLL1 with type 1 diabetes in patients characterized by low polygenic risk score. Metabolism: Clinical and Experimental, 2021, 114, 154418.	1.5	6
44	Unsupervised modeling and genome-wide association identify novel features of allergic march trajectories. Journal of Allergy and Clinical Immunology, 2021, 147, 677-685.e10.	1.5	19
45	De novo loss-of-function variants in X-linked MED12 are associated with Hardikar syndrome in females. Genetics in Medicine, 2021, 23, 637-644.	1.1	16
46	Evaluation of the MC4R gene across eMERGE network identifies many unreported obesity-associated variants. International Journal of Obesity, 2021, 45, 155-169.	1.6	19
47	Loci identified by a genomeâ€wide association study of carotid artery stenosis in the eMERGE network. Genetic Epidemiology, 2021, 45, 4-15.	0.6	6
48	Lossless integration of multiple electronic health records for identifying pleiotropy using summary statistics. Nature Communications, 2021, 12, 168.	5.8	2
49	<i>FLNC</i> and <i>MYLK2</i> Gene Mutations in a Chinese Family with Different Phenotypes of Cardiomyopathy. International Heart Journal, 2021, 62, 127-134.	0.5	6
50	Association of novel rare coding variants with juvenile idiopathic arthritis. Annals of the Rheumatic Diseases, 2021, 80, 626-631.	0.5	6
51	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
52	Association between triglycerides, known risk SNVs and conserved rare variation in SLC25A40 in a multi-ancestry cohort. BMC Medical Genomics, 2021, 14, 11.	0.7	4
53	Expanded phenotypic spectrum of <scp>JAG1</scp>â€associated diseases: Central conducting lymphatic anomaly with a pathogenic variant in <scp>JAG1</scp>. Clinical Genetics, 2021, 99, 742-743.	1.0	7
54	Integrative analysis of genome-wide association studies identifies novel loci associated with neuropsychiatric disorders. Translational Psychiatry, 2021, 11, 69.	2.4	39

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55	A Transparent Approach to Calculate Detection Rate and Residual Risk for Carrier Screening. Journal of Molecular Diagnostics, 2021, 23, 91-102.	1.2	2
56	A novel heterotaxy gene: Expansion of the phenotype of TTC21B â€spectrum disease. American Journal of Medical Genetics, Part A, 2021, 185, 1266-1269.	0.7	5
57	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, 310.	1.0	10
58	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.	2.0	17
59	MONTAGE: a new tool for high-throughput detection of mosaic copy number variation. BMC Genomics, 2021, 22, 133.	1.2	4
60	Performance of model-based multifactor dimensionality reduction methods for epistasis detection by controlling population structure. BioData Mining, 2021, 14, 16.	2.2	2
61	NAC blocks Cystatin C amyloid complex aggregation in a cell system and in skin of HCCAA patients. Nature Communications, 2021, 12, 1827.	5.8	5
62	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
63	Response to Li and Hopper. American Journal of Human Genetics, 2021, 108, 527-529.	2.6	5
64	Macrophages in SHH subgroup medulloblastoma display dynamic heterogeneity that varies with treatment modality. Cell Reports, 2021, 34, 108917.	2.9	27
65	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, 3364.	1.8	4
66	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
67	Model-based deep embedding for constrained clustering analysis of single cell RNA-seq data. Nature Communications, 2021, 12, 1873.	5.8	46
68	Common Variation in Cytoskeletal Genes Is Associated with Conotruncal Heart Defects. Genes, 2021, 12, 655.	1.0	2
69	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.	2.6	18
70	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. Human Genetics, 2021, 140, 1061-1076.	1.8	4
71	Inducible knockout of Clec16a in mice results in sensory neurodegeneration. Scientific Reports, 2021, 11, 9319.	1.6	7
72	JAK/STAT inhibitor therapy partially rescues the lipodystrophic autoimmune phenotype in Clec16a KO mice. Scientific Reports, 2021, 11, 7372.	1.6	6

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73	New insights into hallux valgus by whole exome sequencing study. Experimental Biology and Medicine, 2021, 246, 1607-1616.	1.1	2
74	A new syndrome of moyamoya disease, kidney dysplasia, aminotransferase elevation, and skin disease associated withÂde novo variants in <scp>RNF213</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 2168-2174.	0.7	8
75	RUNX-1 haploinsufficiency causes a marked deficiency of megakaryocyte-biased hematopoietic progenitor cells. Blood, 2021, 137, 2662-2675.	0.6	16
76	Serum levels of the IgA isotype switch factor TGFâ€î²1 are elevated in patients with COVIDâ€19. FEBS Letters, 2021, 595, 1819-1824.	1.3	16
77	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	1.0	21
78	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	4.7	17
79	Metabolomic profiling of anaerobic and aerobic energy metabolic pathways in chronic obstructive pulmonary disease. Experimental Biology and Medicine, 2021, 246, 1586-1596.	1.1	5
80	Genetic correlations between COVID-19 and a variety of traits and diseases. Innovation(China), 2021, 2, 100112.	5.2	7
81	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. Journal of Experimental Medicine, 2021, 218, .	4.2	31
82	Interaction between Genetic Risk Scores for reduced pulmonary function and smoking, asthma and endotoxin. Thorax, 2021, 76, 1219-1226.	2.7	7
83	Combined application of genetic and polygenic risk scores for type 1 diabetes risk prediction. Diabetes, Obesity and Metabolism, 2021, 23, 2001-2003.	2.2	2
84	Genomic considerations for FHIRÂ®; eMERGE implementation lessons. Journal of Biomedical Informatics, 2021, 118, 103795.	2.5	15
85	Using primary teeth and archived dried spots for exposomic studies in children: Exploring new paths in the environmental epidemiology of pediatric cancer. BioEssays, 2021, 43, e2100030.	1.2	6
86	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.	2.8	16
87	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.	9.4	17
88	Association Between a Common, Benign Genotype and Unnecessary Bone Marrow Biopsies Among African American Patients. JAMA Internal Medicine, 2021, 181, 1100.	2.6	18
89	Expanding the genetic landscape of oralâ€facialâ€digital syndrome with two novel genes. American Journal of Medical Genetics, Part A, 2021, 185, 2409-2416.	0.7	9
90	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.	0.7	1

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91	Deep learning prediction of attention-deficit hyperactivity disorder in African Americans by copy number variation. Experimental Biology and Medicine, 2021, 246, 2317-2323.	1.1	8
92	Genome-Wide Association Studies of Conotruncal Heart Defects with Normally Related Great Vessels in the United States. Genes, 2021, 12, 1030.	1.0	1
93	Genetic architecture of type 1 diabetes with low genetic risk score informed by 41 unreported loci. Communications Biology, 2021, 4, 908.	2.0	9
94	Neighborhood deprivation and epigenetic aging. ISEE Conference Abstracts, 2021, 2021, .	0.0	0
95	Discovery of Novel Host Molecular Factors Underlying HBV/HCV Infection. Frontiers in Cell and Developmental Biology, 2021, 9, 690882.	1.8	0
96	Newborn DNA Methylation Signatures Related to Prenatal Smoking Exposures in the PACE Consortium. ISEE Conference Abstracts, 2021, 2021, .	0.0	0
97	Insights into non-autoimmune type 1 diabetes with 13 novel loci in low polygenic risk score patients. Scientific Reports, 2021, 11, 16013.	1.6	7
98	Genetic Variation in PADI6-PADI4 on 1p36.13 Is Associated with Common Forms of Human Generalized Epilepsy. Genes, 2021, 12, 1441.	1.0	7
99	Epigenome-Wide DNA Methylation and Pesticide Use in the Agricultural Lung Health Study. Environmental Health Perspectives, 2021, 129, 97008.	2.8	20
100	HIF-1Î± Pulmonary Phenotype Wide Association Study Unveils a Link to Inflammatory Airway Conditions. Frontiers in Genetics, 2021, 12, 756645.	1.1	6
101	DeepCNV: a deep learning approach for authenticating copy number variations. Briefings in Bioinformatics, 2021, 22, .	3.2	15
102	Ciliopathies: Coloring outside of the lines. American Journal of Medical Genetics, Part A, 2021, 185, 687-694.	0.7	7
103	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	2.6	11
104	Rare variant analysis in eczema identifies exonic variants in DUSP1, NOTCH4 and SLC9A4. Nature Communications, 2021, 12, 6618.	5.8	17
105	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	13.7	353
106	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.	1.3	18
107	Association of Genetic Risk of Obesity with Postoperative Complications Using Mendelian Randomization. World Journal of Surgery, 2020, 44, 84-94.	0.8	4
108	CNV Association of Diverse Clinical Phenotypes from eMERGE reveals novel disease biology underlying cardiovascular disease. International Journal of Cardiology, 2020, 298, 107-113.	0.8	7

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109	Evaluating sequence data quality from the Swift Accel-Amplicon CFTR Panel. Scientific Data, 2020, 7, 8.	2.4	2
110	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
111	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.	0.7	1
112	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	3.7	33
113	Xâ€chromosome association studies of congenital heart defects. American Journal of Medical Genetics, Part A, 2020, 182, 250-254.	0.7	1
114	Genetic Discrimination Between LADA and Childhood-Onset Type 1 Diabetes Within the MHC. Diabetes Care, 2020, 43, 418-425.	4.3	23
115	Genetic Architecture of Abdominal Aortic Aneurysm in the Million Veteran Program. Circulation, 2020, 142, 1633-1646.	1.6	78
116	The Multi-Omics Architecture of Juvenile Idiopathic Arthritis. Cells, 2020, 9, 2301.	1.8	18
117	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
118	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. PLoS Genetics, 2020, 16, e1008718.	1.5	95
119	Genomeâ€wide association analysis of psoriasis patients treated with antiâ€TNF drugs. Experimental Dermatology, 2020, 29, 1225-1232.	1.4	11
120	European genetic ancestry associated with risk of childhood ependymoma. Neuro-Oncology, 2020, 22, 1637-1646.	0.6	16
121	Biliary-Atresia-Associated Mannosidase-1-Alpha-2 Gene Regulates Biliary and Ciliary Morphogenesis and Laterality. Frontiers in Physiology, 2020, 11, 538701.	1.3	13
122	Elucidation of DNA methylation on N6-adenine with deep learning. Nature Machine Intelligence, 2020, 2, 466-475.	8.3	7
123	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. American Journal of Human Genetics, 2020, 107, 432-444.	2.6	124
124	Participant choices for return of genomic results in the eMERGE Network. Genetics in Medicine, 2020, 22, 1821-1829.	1.1	25
125	Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in Medicine, 2020, 22, 2114-2119.	1.1	15
126	Genetic predisposition to longer telomere length and risk of childhood, adolescent and adult-onset ependymoma. Acta Neuropathologica Communications, 2020, 8, 173.	2.4	15

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127	Expression Pattern of the SARS-CoV-2 Entry Genes ACE2 and TMPRSS2 in the Respiratory Tract. Viruses, 2020, 12, 1174.	1.5	27
128	High prevalence of elevated serum liver enzymes in Chinese children suggests metabolic syndrome as a common risk factor. Journal of Paediatrics and Child Health, 2020, 56, 1590-1596.	0.4	1
129	Non-coding structural variation differentially impacts attention-deficit hyperactivity disorder (ADHD) gene networks in African American vs Caucasian children. Scientific Reports, 2020, 10, 15252.	1.6	5
130	Autophagy mitigates ethanol-induced mitochondrial dysfunction and oxidative stress in esophageal keratinocytes. PLoS ONE, 2020, 15, e0239625.	1.1	18
131	Genomic risk scores for juvenile idiopathic arthritis and its subtypes. Annals of the Rheumatic Diseases, 2020, 79, 1572-1579.	0.5	12
132	Genetic Studies of Leptin Concentrations Implicate Leptin in the Regulation of Early Adiposity. Diabetes, 2020, 69, 2806-2818.	0.3	26
133	Mitochondrial DNA haplogroups and risk of attention deficit and hyperactivity disorder in European Americans. Translational Psychiatry, 2020, 10, 370.	2.4	11
134	COVID-19: Look to the Future, Learn from the Past. Viruses, 2020, 12, 1226.	1.5	8
135	Lung Function in African American Children with Asthma Is Associated with Novel Regulatory Variants of the KIT LigandKITLG/SCFand Gene-By-Air-Pollution Interaction. Genetics, 2020, 215, 869-886.	1.2	11
136	Variants in the Kisspeptin-GnRH Pathway Modulate the Hormonal Profile and Reproductive Outcomes. DNA and Cell Biology, 2020, 39, 1012-1022.	0.9	3
137	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
138	Distinct features of SARS-CoV-2-specific IgA response in COVID-19 patients. European Respiratory Journal, 2020, 56, 2001526.	3.1	292
139	The polygenic architecture of left ventricular mass mirrors the clinical epidemiology. Scientific Reports, 2020, 10, 7561.	1.6	13
140	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
141	Activating variants in <scp>PDGFRB</scp> result in a spectrum of disorders responsive to imatinib monotherapy. American Journal of Medical Genetics, Part A, 2020, 182, 1576-1591.	0.7	21
142	Application of exome sequencing to diagnose a novel presentation of the Cornelia de Lange syndrome in an Afroâ€Caribbean family. Molecular Genetics & Genomic Medicine, 2020, 8, e1318.	0.6	2
143	Gene-based analyses of the maternal genome implicate maternal effect genes as risk factors for conotruncal heart defects. PLoS ONE, 2020, 15, e0234357.	1.1	8
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	A homozygous truncating NALCN variant in two Afroâ€Caribbean siblings with hypotonia and dolichocephaly. American Journal of Medical Genetics, Part A, 2020, 182, 1877-1880.	0.7	3
146	Detection of maternal X chromosome abnormalities using single nucleotide polymorphismâ€“based noninvasive prenatal testing. American Journal of Obstetrics & Gynecology MFM, 2020, 2, 100152.	1.3	13
147	Mitochondrial DNA Haplogroups and Susceptibility to Neuroblastoma. Journal of the National Cancer Institute, 2020, 112, 1259-1266.	3.0	10
148	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. Molecular Genetics and Metabolism, 2020, 130, 49-57.	0.5	15
149	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. Nature Communications, 2020, 11, 255.	5.8	48
150	Regulation of Janus Kinase 2 by an Inflammatory Bowel Disease Causal Non-coding Single Nucleotide Polymorphism. Journal of Crohn's and Colitis, 2020, 14, 646-653.	0.6	5
151	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
152	Role of the ADCY9 gene in cardiac abnormalities of the Rubinstein-Taybi syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 101.	1.2	2
153	Association of Neighborhood Deprivation With Epigenetic Aging Using 4 Clock Metrics. JAMA Network Open, 2020, 3, e2024329.	2.8	50
154	TNFAIP8 controls murine intestinal stem cell homeostasis and regeneration by regulating microbiome-induced Akt signaling. Nature Communications, 2020, 11, 2591.	5.8	19
155	Type I IFN response associated with mTOR activation in the TAFRO subtype of idiopathic multicentric Castleman disease. JCI Insight, 2020, 5, .	2.3	35
156	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.	1.1	16
157	Kaposiform lymphangiomatosis effectively treated with <scp>MEK</scp> inhibition. EMBO Molecular Medicine, 2020, 12, e12324.	3.3	51
158	Rare Genetic Variants of Large Effect Influence Risk of Type 1 Diabetes. Diabetes, 2020, 69, 784-795.	0.3	69
159	Genetic architecture study of rheumatoid arthritis and juvenile idiopathic arthritis. PeerJ, 2020, 8, e8234.	0.9	3
160	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, e3197-e3206.	1.8	6
161	Development and validation of targeted treatments for the rare tumor syndrome infantile myofibromatosis.. Journal of Clinical Oncology, 2020, 38, e22519-e22519.	0.8	0
162	The Infection Rate of COVID-19 in Wuhan, China: Combined Analysis of Population Samples. Journal of Medical Internet Research, 2020, 22, e20914.	2.1	2

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163	TNFAIP8 is a central regulator of intestinal homeostasis and regeneration. FASEB Journal, 2020, 34, 1-1.	0.2	0
164	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. Molecular Vision, 2020, 26, 216-225.	1.1	2
165	Title is missing!. , 2020, 15, e0234357.		0
166	Title is missing!. , 2020, 15, e0234357.		0
167	Title is missing!. , 2020, 15, e0234357.		0
168	Title is missing!. , 2020, 15, e0234357.		0
169	Title is missing!. , 2020, 15, e0239625.		0
170	Title is missing!. , 2020, 15, e0239625.		0
171	Title is missing!. , 2020, 15, e0239625.		0
172	Title is missing!. , 2020, 15, e0239625.		0
173	Implications of an admixed Brazilian population in schizophrenia polygenic risk score. Schizophrenia Research, 2019, 204, 404-406.	1.1	6
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