Hakon H Hakonarson

List of Publications by Year
in descending order

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657	103,705	354	339
papers	citations	139 h-index	293 g-index

700	700	700	124064
all docs	docs citations	times ranked	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	Common Genetic Variation and Age of Onset of Anorexia Nervosa. Biological Psychiatry Global Open Science, 2022, 2, 368-378.	1.0	10
4	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
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	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
10	Identification of Mitochondrial DNA Variants Associated With Risk of Neuroblastoma. Journal of the National Cancer Institute, 2022, 114, 910-913.	3.0	4
11	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
12	Circulating LIGHT (TNFSF14) and Interleukin-18 Levels in Sepsis-Induced Multi-Organ Injuries. Biomedicines, 2022, 10, 264.	1.4	7
13	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
14	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
15	The reckoning: The return of genomic results to 1444 participants across the eMERGE3 Network. Genetics in Medicine, 2022, 24, 1130-1138.	1.1	12
16	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
17	Genetic analysis for type 1 diabetes genes in juvenile dermatomyositis unveils genetic disease overlap. Rheumatology, 2022, , .	0.9	2
18	Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study. Circulation, 2022, 145, 877-891.	1.6	18

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19	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
20	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
21	Identification of Novel Loci Shared by Juvenile Idiopathic Arthritis Subtypes Through Integrative Genetic Analysis. Arthritis and Rheumatology, 2022, 74, 1420-1429.	2.9	4
22	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
23	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
24	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
25	Maternal effect genes as risk factors for congenital heart defects. Human Genetics and Genomics Advances, 2022, 3, 100098.	1.0	2
26	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
27	Manifestations of Alzheimerâ€™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.	1.6	12
28	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncology, 2022, 8, 835.	3.4	25
29	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
30	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
31	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
32	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
33	Multiancestral polygenic risk score for pediatric asthma. Journal of Allergy and Clinical Immunology, 2022, 150, 1086-1096.	1.5	14
34	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
35	Genetics of early-life head circumference and genetic correlations with neurological, psychiatric and cognitive outcomes. BMC Medical Genomics, 2022, 15, .	0.7	2
36	An electronic health record (EHR) phenotype algorithm to identify patients with attention deficit hyperactivity disorders (ADHD) and psychiatric comorbidities. Journal of Neurodevelopmental Disorders, 2022, 14, .	1.5	9

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37	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
38	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
39	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
40	Risk of preâ€•eclampsia in patients with a maternal genetic predisposition to common medical conditions: a caseâ€“control study. BJOG: an International Journal of Obstetrics and Gynaecology, 2021, 128, 55-65.	1.1	19
41	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
42	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
43	Novel Alzheimer Disease Risk Loci and Pathways in African American Individuals Using the African Genome Resources Panel. JAMA Neurology, 2021, 78, 102.	4.5	144
44	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
45	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
46	Lossless integration of multiple electronic health records for identifying pleiotropy using summary statistics. Nature Communications, 2021, 12, 168.	5.8	2
47	Association of novel rare coding variants with juvenile idiopathic arthritis. Annals of the Rheumatic Diseases, 2021, 80, 626-631.	0.5	6
48	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
49	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
50	Integrative analysis of genome-wide association studies identifies novel loci associated with neuropsychiatric disorders. Translational Psychiatry, 2021, 11, 69.	2.4	39
51	Epigenetics in child psychiatry. , 2021, , 553-573.		0
52	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
53	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
54	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

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55	MONTAGE: a new tool for high-throughput detection of mosaic copy number variation. BMC Genomics, 2021, 22, 133.	1.2	4
56	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, 32, 805-820.	3.0	17
57	Performance of model-based multifactor dimensionality reduction methods for epistasis detection by controlling population structure. BioData Mining, 2021, 14, 16.	2.2	2
58	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
59	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
60	Response to Li and Hopper. American Journal of Human Genetics, 2021, 108, 527-529.	2.6	5
61	Macrophages in SHH subgroup medulloblastoma display dynamic heterogeneity that varies with treatment modality. Cell Reports, 2021, 34, 108917.	2.9	27
62	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
63	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
64	Model-based deep embedding for constrained clustering analysis of single cell RNA-seq data. Nature Communications, 2021, 12, 1873.	5.8	46
65	Common Variation in Cytoskeletal Genes Is Associated with Conotruncal Heart Defects. Genes, 2021, 12, 655.	1.0	2
66	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
67	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. Human Genetics, 2021, 140, 1061-1076.	1.8	4
68	Inducible knockout of Clec16a in mice results in sensory neurodegeneration. Scientific Reports, 2021, 11, 9319.	1.6	7
69	JAK/STAT inhibitor therapy partially rescues the lipodystrophic autoimmune phenotype in Clec16a KO mice. Scientific Reports, 2021, 11, 7372.	1.6	6
70	New insights into hallux valgus by whole exome sequencing study. Experimental Biology and Medicine, 2021, 246, 1607-1616.	1.1	2
71	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
72	RUNX-1 haploinsufficiency causes a marked deficiency of megakaryocyte-biased hematopoietic progenitor cells. Blood, 2021, 137, 2662-2675.	0.6	16

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73	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
74	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	1.0	21
75	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	4.7	17
76	Large trans-ethnic meta-analysis identifies AKR1C4 as a novel gene associated with age at menarche. Human Reproduction, 2021, 36, 1999-2010.	0.4	10
77	Constrained chromatin accessibility in PU.1-mutated agammaglobulinemia patients. Journal of Experimental Medicine, 2021, 218, .	4.2	31
78	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.	2.6	35
79	Novel EDGE encoding method enhances ability to identify genetic interactions. PLoS Genetics, 2021, 17, e1009534.	1.5	5
80	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
81	Genomic considerations for FHIRÂ®; eMERGE implementation lessons. Journal of Biomedical Informatics, 2021, 118, 103795.	2.5	15
82	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
83	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
84	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
85	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
86	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
87	Genome-Wide Association Studies of Conotruncal Heart Defects with Normally Related Great Vessels in the United States. Genes, 2021, 12, 1030.	1.0	1
88	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	1.1	3
89	Genetic architecture of type 1 diabetes with low genetic risk score informed by 41 unreported loci. Communications Biology, 2021, 4, 908.	2.0	9
90	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

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91	HIF-1Î± Pulmonary Phenotype Wide Association Study Unveils a Link to Inflammatory Airway Conditions. Frontiers in Genetics, 2021, 12, 756645.	1.1	6
92	DeepCNV: a deep learning approach for authenticating copy number variations. Briefings in Bioinformatics, 2021, 22, .	3.2	15
93	Ciliopathies: Coloring outside of the lines. American Journal of Medical Genetics, Part A, 2021, 185, 687-694.	0.7	7
94	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	1.5	9
95	Rare variant analysis in eczema identifies exonic variants in DUSP1, NOTCH4 and SLC9A4. Nature Communications, 2021, 12, 6618.	5.8	17
96	The power of genetic diversity in genome-wide association studies of lipids. Nature, 2021, 600, 675-679.	13.7	353
97	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
98	Association of Genetic Risk of Obesity with Postoperative Complications Using Mendelian Randomization. World Journal of Surgery, 2020, 44, 84-94.	0.8	4
99	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
100	Genetic Underpinnings of Asthma and Related Traits. , 2020, , 341-360.		0
101	Evaluating sequence data quality from the Swift Accel-Amplicon CFTR Panel. Scientific Data, 2020, 7, 8.	2.4	2
102	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
103	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
104	Xâ€¢chromosome association studies of congenital heart defects. American Journal of Medical Genetics, Part A, 2020, 182, 250-254.	0.7	1
105	Genetic Discrimination Between LADA and Childhood-Onset Type 1 Diabetes Within the MHC. Diabetes Care, 2020, 43, 418-425.	4.3	23
106	Genetic Architecture of Abdominal Aortic Aneurysm in the Million Veteran Program. Circulation, 2020, 142, 1633-1646.	1.6	78
107	The Multi-Omics Architecture of Juvenile Idiopathic Arthritis. Cells, 2020, 9, 2301.	1.8	18
108	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

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109	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. PLoS Genetics, 2020, 16, e1008718.	1.5	95
110	European genetic ancestry associated with risk of childhood ependymoma. Neuro-Oncology, 2020, 22, 1637-1646.	0.6	16
111	Severe Lymphatic Disorder Resolved With MEK Inhibition in a Patient With Noonan Syndrome and SOS1 Mutation. Pediatrics, 2020, 146, .	1.0	56
112	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
113	Biliary-Atresia-Associated Mannosidase-1-Alpha-2 Gene Regulates Biliary and Ciliary Morphogenesis and Laterality. Frontiers in Physiology, 2020, 11, 538701.	1.3	13
114	Genome-wide Modeling of Polygenic Risk Score in Colorectal Cancer Risk. American Journal of Human Genetics, 2020, 107, 432-444.	2.6	124
115	Participant choices for return of genomic results in the eMERGE Network. Genetics in Medicine, 2020, 22, 1821-1829.	1.1	25
116	Genetic predisposition to longer telomere length and risk of childhood, adolescent and adult-onset ependymoma. Acta Neuropathologica Communications, 2020, 8, 173.	2.4	15
117	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
118	Genomic risk scores for juvenile idiopathic arthritis and its subtypes. Annals of the Rheumatic Diseases, 2020, 79, 1572-1579.	0.5	12
119	Genetic Studies of Leptin Concentrations Implicate Leptin in the Regulation of Early Adiposity. Diabetes, 2020, 69, 2806-2818.	0.3	26
120	Mitochondrial DNA haplogroups and risk of attention deficit and hyperactivity disorder in European Americans. Translational Psychiatry, 2020, 10, 370.	2.4	11
121	COVID-19: Look to the Future, Learn from the Past. Viruses, 2020, 12, 1226.	1.5	8
122	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
123	Variants in the Kisspeptin-GnRH Pathway Modulate the Hormonal Profile and Reproductive Outcomes. DNA and Cell Biology, 2020, 39, 1012-1022.	0.9	3
124	Distinct features of SARS-CoV-2-specific IgA response in COVID-19 patients. European Respiratory Journal, 2020, 56, 2001526.	3.1	292
125	The polygenic architecture of left ventricular mass mirrors the clinical epidemiology. Scientific Reports, 2020, 10, 7561.	1.6	13
126	Frequency of genomic secondaryÂfindings among 21,915 eMERGE network participants. Genetics in Medicine, 2020, 22, 1470-1477.	1.1	61

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127	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	3.7	47
128	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
129	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
130	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
131	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
132	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
133	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
134	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
135	Mitochondrial DNA Haplogroups and Susceptibility to Neuroblastoma. Journal of the National Cancer Institute, 2020, 112, 1259-1266.	3.0	10
136	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. Molecular Genetics and Metabolism, 2020, 130, 49-57.	0.5	15
137	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. Nature Communications, 2020, 11, 255.	5.8	48
138	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
139	Role of the ADCY9 gene in cardiac abnormalities of the Rubinstein-Taybi syndrome. Orphanet Journal of Rare Diseases, 2020, 15, 101.	1.2	2
140	TNFAIP8 controls murine intestinal stem cell homeostasis and regeneration by regulating microbiome-induced Akt signaling. Nature Communications, 2020, 11, 2591.	5.8	19
141	Type I IFN response associated with mTOR activation in the TAFRO subtype of idiopathic multicentric Castleman disease. JCI Insight, 2020, 5, .	2.3	35
142	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
143	Kaposiform lymphangiomatosis effectively treated with <scp>MEK</scp> inhibition. EMBO Molecular Medicine, 2020, 12, e12324.	3.3	51
144	Rare Genetic Variants of Large Effect Influence Risk of Type 1 Diabetes. Diabetes, 2020, 69, 784-795.	0.3	69

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145	Genetic architecture study of rheumatoid arthritis and juvenile idiopathic arthritis. PeerJ, 2020, 8, e8234.	0.9	3
146	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
147	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
148	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.	0.7	2
149	Exome sequencing identifies mutations in three cases diagnosed with Retinitis Pigmentosa and hearing impairment. Molecular Vision, 2020, 26, 216-225.	1.1	2
150	Mutations in topoisomerase IIÎ² result in a B cell immunodeficiency. Nature Communications, 2019, 10, 3644.	5.8	37
151	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.	0.5	2
152	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.	2.6	237
153	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
154	Gene-based genome-wide association studies and meta-analyses of conotruncal heart defects. PLoS ONE, 2019, 14, e0219926.	1.1	15
155	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
156	Genetic risk for Alzheimer's disease and functional brain connectivity in children and adolescents. Neurobiology of Aging, 2019, 82, 10-17.	1.5	23
157	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
158	Genetic comparison of sickle cell anaemia cohorts from Brazil andÂthe United States reveals high levels of divergence. Scientific Reports, 2019, 9, 10896.	1.6	9
159	ARAF recurrent mutation causes central conducting lymphatic anomaly treatable with a MEK inhibitor. Nature Medicine, 2019, 25, 1116-1122.	15.2	136
160	Epistasis Detection in Genome-Wide Screening for Complex Human Diseases in Structured Populations. Systems Medicine (New Rochelle, N Y), 2019, 2, 19-27.	1.4	9
161	Phenome-wide association analysis of LDL-cholesterol lowering genetic variants in PCSK9. BMC Cardiovascular Disorders, 2019, 19, 240.	0.7	22
162	The Genetic Contribution to Type 1 Diabetes. Current Diabetes Reports, 2019, 19, 116.	1.7	48

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163	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	5.8	84
164	Detecting multiple differentially methylated CpG sites and regions related to dimensional psychopathology in youths. Clinical Epigenetics, 2019, 11, 146.	1.8	13
165	Target Genes of Autism Risk Loci in Brain Frontal Cortex. Frontiers in Genetics, 2019, 10, 707.	1.1	16
166	Application of ACMG criteria to classify variants in the human gene mutation database. Journal of Human Genetics, 2019, 64, 1091-1095.	1.1	10
167	GWAS on longitudinal growth traits reveals different genetic factors influencing infant, child, and adult BMI. Science Advances, 2019, 5, eaaw3095.	4.7	86
168	Germline 16p11.2 Microdeletion Predisposes to Neuroblastoma. American Journal of Human Genetics, 2019, 105, 658-668.	2.6	31
169	Variants in the fetal genome near pro-inflammatory cytokine genes on 2q13 associate with gestational duration. Nature Communications, 2019, 10, 3927.	5.8	49
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