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List of Publications by Year
in descending order

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83 papers	14,292 citations	43973 48 h-index	56606 83 g-index
95 all docs	95 docs citations	95 times ranked	25158 citing authors

#	Article	IF	Citations
1	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. Nature, 2012, 482, 226-231.	13.7	2,129
2	Hotspot Mutations in H3F3A and IDH1 Define Distinct Epigenetic and Biological Subgroups of Glioblastoma. Cancer Cell, 2012, 22, 425-437.	7.7	1,551
3	K27M mutation in histone H3.3 defines clinically and biologically distinct subgroups of pediatric diffuse intrinsic pontine gliomas. Acta Neuropathologica, 2012, 124, 439-447.	3.9	799
4	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. Nature Genetics, 2013, 45, 927-932.	9.4	674
5	Mapping the human genetic architecture of COVID-19. Nature, 2021, 600, 472-477.	13.7	640
6	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	9.4	621
7	Recurrent somatic mutations in ACVR1 in pediatric midline high-grade astrocytoma. Nature Genetics, 2014, 46, 462-466.	9.4	381
8	Frequent ATRX mutations and loss of expression in adult diffuse astrocytic tumors carrying IDH1/IDH2 and TP53 mutations. Acta Neuropathologica, 2012, 124, 615-625.	3.9	376
9	Clonal selection drives genetic divergence of metastatic medulloblastoma. Nature, 2012, 482, 529-533.	13.7	376
10	What can exome sequencing do for you?. Journal of Medical Genetics, 2011, 48, 580-589.	1.5	321
11	Open Targets Genetics: systematic identification of trait-associated genes using large-scale genetics and functional genomics. Nucleic Acids Research, 2021, 49, D1311-D1320.	6.5	295
12	Open Targets Platform: supporting systematic drugâ€“target identification and prioritisation. Nucleic Acids Research, 2021, 49, D1302-D1310.	6.5	265
13	Genome-wide meta-analysis, fine-mapping and integrative prioritization implicate new Alzheimerâ€™s disease risk genes. Nature Genetics, 2021, 53, 392-402.	9.4	258
14	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
15	Mutations in SETD2 and genes affecting histone H3K36 methylation target hemispheric high-grade gliomas. Acta Neuropathologica, 2013, 125, 659-669.	3.9	250
16	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817.	2.6	219
17	An open approach to systematically prioritize causal variants and genes at all published human GWAS trait-associated loci. Nature Genetics, 2021, 53, 1527-1533.	9.4	208
18	Mutations inSYNGAP1Cause Intellectual Disability, Autism, and a Specific Form of Epilepsy by Inducing Haploinsufficiency. Human Mutation, 2013, 34, 385-394.	1.1	196

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19	Molecular and functional variation in iPSC-derived sensory neurons. Nature Genetics, 2018, 50, 54-61.	9.4	191
20	Haploinsufficiency of SF3B4, a Component of the Pre-mRNA Spliceosomal Complex, Causes Nager Syndrome. American Journal of Human Genetics, 2012, 90, 925-933.	2.6	188
21	Haploinsufficiency of a Spliceosomal GTPase Encoded by EFTUD2 Causes Mandibulofacial Dysostosis with Microcephaly. American Journal of Human Genetics, 2012, 90, 369-377.	2.6	180
22	Mutations in NMNAT1 cause Leber congenital amaurosis and identify a new disease pathway for retinal degeneration. Nature Genetics, 2012, 44, 1035-1039.	9.4	177
23	CTP synthase 1 deficiency in humans reveals its central role in lymphocyte proliferation. Nature, 2014, 510, 288-292.	13.7	174
24	Fusion of TTYH1 with the C19MC microRNA cluster drives expression of a brain-specific DNMT3B isoform in the embryonal brain tumor ETMR. Nature Genetics, 2014, 46, 39-44.	9.4	167
25	Mutations in DDHD2, Encoding an Intracellular Phospholipase A1, Cause a Recessive Form of Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1073-1081.	2.6	159
26	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	2.6	157
27	Mutations in PIK3R1 Cause SHORT Syndrome. American Journal of Human Genetics, 2013, 93, 158-166.	2.6	156
28	Mutations inFLNCare Associated with Familial Restrictive Cardiomyopathy. Human Mutation, 2016, 37, 269-279.	1.1	138
29	A Recurrent PDGFRB Mutation Causes Familial Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 996-1000.	2.6	135
30	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. American Journal of Human Genetics, 2017, 100, 865-884.	2.6	131
31	Mutations in C5ORF42 Cause Joubert Syndrome in the French Canadian Population. American Journal of Human Genetics, 2012, 90, 693-700.	2.6	118
32	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. Nature Genetics, 2014, 46, 510-515.	9.4	118
33	A map of transcriptional heterogeneity and regulatory variation in human microglia. Nature Genetics, 2021, 53, 861-868.	9.4	115
34	Mutations in KAT6B, Encoding a Histone Acetyltransferase, Cause Genitopatellar Syndrome. American Journal of Human Genetics, 2012, 90, 282-289.	2.6	112
35	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephalyâ€“capillary malformation syndrome. Nature Genetics, 2013, 45, 556-562.	9.4	94
36	Mutations in NOTCH2 in families with Hajdu-Cheney syndrome. Human Mutation, 2011, 32, 1114-1117.	1.1	93

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37	Exome Sequencing as a Diagnostic Tool for Pediatricâ€Onset Ataxia. Human Mutation, 2014, 35, 45-49.	1.1	91
38	Mutations in NFKB2and potential genetic heterogeneity in patients with DAVID syndrome, having variable endocrine and immune deficiencies. BMC Medical Genetics, 2014, 15, 139.	2.1	84
39	Whole genome sequencing and imputation in isolated populations identify genetic associations with medically-relevant complex traits. Nature Communications, 2017, 8, 15606.	5.8	79
40	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79.	2.6	77
41	Mutations inTMEM231cause Joubert syndrome in French Canadians. Journal of Medical Genetics, 2012, 49, 636-641.	1.5	72
42	Novel inborn error of folate metabolism: identification by exome capture and sequencing of mutations in the MTHFD1 gene in a single proband. Journal of Medical Genetics, 2011, 48, 590-592.	1.5	66
43	Mutation in The Nuclear-Encoded Mitochondrial Isoleucyl-tRNA SynthetaseIARS2in Patients with Cataracts, Growth Hormone Deficiency with Short Stature, Partial Sensorineural Deafness, and Peripheral Neuropathy or with Leigh Syndrome. Human Mutation, 2014, 35, n/a-n/a.	1.1	66
44	Differential stability of 2â€²F-ANAâ€¢RNA and ANAâ€¢RNA hybrid duplexes: roles of structure, pseudohydrogen bonding, hydration, ion uptake and flexibility. Nucleic Acids Research, 2010, 38, 2498-2511.	6.5	65
45	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. Journal of Medical Genetics, 2014, 51, 470-474.	1.5	64
46	Specific combination of compound heterozygous mutations in 17Î²-hydroxysteroid dehydrogenase type 4 (HSD17B4) defines a new subtype of D-bifunctional protein deficiency. Orphanet Journal of Rare Diseases, 2012, 7, 90.	1.2	60
47	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebroâ€“costoâ€“mandibular syndrome. Nature Communications, 2014, 5, 4483.	5.8	57
48	GeneMatcher Aids in the Identification of a New Malformation Syndrome with Intellectual Disability, Unique Facial Dysmorphisms, and Skeletal and Connective Tissue Abnormalities Caused by De Novo Variants inHNRNPK. Human Mutation, 2015, 36, 1009-1014.	1.1	56
49	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	2.6	56
50	Bioinactive ACTH Causing Glucocorticoid Deficiency. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 736-742.	1.8	51
51	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. Neurobiology of Aging, 2015, 36, 1222.e1-1222.e5.	1.5	50
52	Intellectual disability associated with a homozygous missense mutation in THOC6. Orphanet Journal of Rare Diseases, 2013, 8, 62.	1.2	48
53	Congenital Visual Impairment and Progressive Microcephaly Due to Lysylâ€“Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations. Journal of Child Neurology, 2015, 30, 1037-1043.	0.7	47
54	CCDC88B is a novel regulator of maturation and effector functions of T cells during pathological inflammation. Journal of Experimental Medicine, 2014, 211, 2519-2535.	4.2	44

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55	Compound heterozygous mutations in glycyl-tRNA synthetase are a proposed cause of systemic mitochondrial disease. BMC Medical Genetics, 2014, 15, 36.	2.1	41
56	Mutations in ALDH6A1 encoding methylmalonate semialdehyde dehydrogenase are associated with dysmyelination and transient methylmalonic aciduria. Orphanet Journal of Rare Diseases, 2013, 8, 98.	1.2	37
57	An inherited immunoglobulin class-switch recombination deficiency associated with a defect in the INO80 chromatin remodeling complex. Journal of Allergy and Clinical Immunology, 2015, 135, 998-1007.e6.	1.5	37
58	CYRI/FAM49B negatively regulates RAC1-driven cytoskeletal remodelling and protects against bacterial infection. Nature Microbiology, 2019, 4, 1516-1531.	5.9	37
59	Wholeâ€exome sequencing in an individual with severe global developmental delay and intractable epilepsy identifies a novel, de novo GRIN2A mutation. Epilepsia, 2014, 55, e75-9.	2.6	36
60	A novel multisystem disease associated with recessive mutations in the tyrosylâ€tRNA synthetase (YARS) gene. American Journal of Medical Genetics, Part A, 2017, 173, 126-134.	0.7	36
61	A family segregating lethal neonatal coenzyme Q₁₀ deficiency caused by mutations in COQ9. Journal of Inherited Metabolic Disease, 2018, 41, 719-729.	1.7	30
62	Metaphyseal Dysplasia with Maxillary Hypoplasia and Brachydactyly Is Caused by a Duplication in RUNX2. American Journal of Human Genetics, 2013, 92, 252-258.	2.6	29
63	Histone H3 Mutations in Pediatric Brain Tumors. Cold Spring Harbor Perspectives in Biology, 2014, 6, a018689-a018689.	2.3	29
64	Whole exome sequencing identifies the TNNI3K gene as a cause of familial conduction system disease and congenital junctional ectopic tachycardia. International Journal of Cardiology, 2015, 185, 114-116.	0.8	29
65	Neuropathologic Features of Pontocerebellar Hypoplasia Type 6. Journal of Neuropathology and Experimental Neurology, 2014, 73, 1009-1025.	0.9	28
66	THEMIS Is Required for Pathogenesis of Cerebral Malaria and Protection against Pulmonary Tuberculosis. Infection and Immunity, 2015, 83, 759-768.	1.0	26
67	A proteome-wide genetic investigation identifies several SARS-CoV-2-exploited host targets of clinical relevance. ELife, 2021, 10, .	2.8	23
68	An N-terminal formyl methionine on COX 1 is required for the assembly of cytochrome c oxidase. Human Molecular Genetics, 2015, 24, 4103-4113.	1.4	22
69	Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. JIMD Reports, 2016, 30, 73-79.	0.7	21
70	A mutation in the TMEM65 gene results in mitochondrial myopathy with severe neurological manifestations. European Journal of Human Genetics, 2017, 25, 744-751.	1.4	21
71	Genome-Wide Mouse Mutagenesis Reveals CD45-Mediated T Cell Function as Critical in Protective Immunity to HSV-1. PLoS Pathogens, 2013, 9, e1003637.	2.1	20
72	Mutations in riboflavin transporter present with severe sensory loss and deafness in childhood. Muscle and Nerve, 2014, 50, 775-779.	1.0	20

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73	Iron Refractory Iron Deficiency Anemia: Presentation With Hyperferritinemia and Response to Oral Iron Therapy. Pediatrics, 2013, 131, e620-e625.	1.0	19
74	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a coâ€occurrence of multiple events. American Journal of Medical Genetics, Part A, 2016, 170, 1820-1825.	0.7	19
75	Molecular Genetics of Achromatopsia in Newfoundland Reveal Genetic Heterogeneity, Founder Effects and the First Cases of Jalili Syndrome in North America. Ophthalmic Genetics, 2013, 34, 119-129.	0.5	18
76	The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome. BMC Neurology, 2014, 14, 22.	0.8	18
77	Altered IFN-Î³â€“Mediated Immunity and Transcriptional Expression Patterns in N-Ethyl-N-Nitrosoureaâ€“Induced STAT4 Mutants Confer Susceptibility to Acute Typhoid-like Disease. Journal of Immunology, 2014, 192, 259-270.	0.4	17
78	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression. , 2017, 58, 1736.		17
79	A novel CCBE1 mutation leading to a mild form of hennekam syndrome: case report and review of the literature. BMC Medical Genetics, 2015, 16, 28.	2.1	14
80	Bridging the Gap between Single Molecule and Ensemble Methods for Measuring Lateral Dynamics in the Plasma Membrane. PLoS ONE, 2013, 8, e78096.	1.1	11
81	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. Journal of Child Neurology, 2019, 34, 74-80.	0.7	9
82	Screening for functional transcriptional and splicing regulatory variants with GenIE. Nucleic Acids Research, 2020, 48, e131-e131.	6.5	8
83	Expansion of the clinical phenotype of the distal 10q26.3 deletion syndrome to include ataxia and hyperemia of the hands and feet. American Journal of Medical Genetics, Part A, 2017, 173, 1611-1619.	0.7	4