

Peer Arts

List of Publications by Year in descending order

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40
papers

3,440
citations

430874

18
h-index

361022

35
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44
all docs

44
docs citations

44
times ranked

7144
citing authors

#	ARTICLE	IF	CITATIONS
1	A de novo paradigm for mental retardation. <i>Nature Genetics</i> , 2010, 42, 1109-1112.	21.4	751
2	<i>STAT1</i> Mutations in Autosomal Dominant Chronic Mucocutaneous Candidiasis. <i>New England Journal of Medicine</i> , 2011, 365, 54-61.	27.0	614
3	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. <i>Nature Genetics</i> , 2010, 42, 483-485.	21.4	417
4	Exome Sequencing Identifies WDR35 Variants Involved in Sensenbrenner Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 418-423.	6.2	260
5	Human TLR10 is an anti-inflammatory pattern-recognition receptor. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, E4478-84.	7.1	211
6	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. <i>American Journal of Human Genetics</i> , 2010, 86, 240-247.	6.2	202
7	Functional genomics identifies type I interferon pathway as central for host defense against <i>Candida albicans</i> . <i>Nature Communications</i> , 2013, 4, 1342.	12.8	157
8	Targeted Next-Generation Sequencing of a 12.5 Mb Homozygous Region Reveals ANO10 Mutations in Patients with Autosomal-Recessive Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2010, 87, 813-819.	6.2	125
9	RUNX1-mutated families show phenotype heterogeneity and a somatic mutation profile unique to germline predisposed AML. <i>Blood Advances</i> , 2020, 4, 1131-1144.	5.2	102
10	Massively parallel sequencing of ataxia genes after array-based enrichment. <i>Human Mutation</i> , 2010, 31, 494-499.	2.5	86
11	<i>MLL2</i> mutation detection in 86 patients with Kabuki syndrome: a genotype-phenotype study. <i>Clinical Genetics</i> , 2013, 84, 539-545.	2.0	85
12	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. <i>Genome Medicine</i> , 2019, 11, 38.	8.2	49
13	Rare genetic variants in interleukin-37 link this anti-inflammatory cytokine to the pathogenesis and treatment of gout. <i>Annals of the Rheumatic Diseases</i> , 2020, 79, 536-544.	0.9	44
14	The mutational burden of therapy-related myeloid neoplasms is similar to primary myelodysplastic syndrome but has a distinctive distribution. <i>Leukemia</i> , 2019, 33, 2842-2853.	7.2	43
15	A systems genomics approach identifies <i>SIGLEC15</i> as a susceptibility factor in recurrent vulvovaginal candidiasis. <i>Science Translational Medicine</i> , 2019, 11, .	12.4	38
16	GATA2 deficiency syndrome: A decade of discovery. <i>Human Mutation</i> , 2021, 42, 1399-1421.	2.5	30
17	The RUNX1 database (RUNX1db): establishment of an expert curated RUNX1 registry and genomics database as a public resource for familial platelet disorder with myeloid malignancy. <i>Haematologica</i> , 2021, 106, 3004-3007.	3.5	29
18	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. <i>Human Molecular Genetics</i> , 2015, 24, 2000-2010.	2.9	25

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19	Progressive multifocal leukoencephalopathy in an immunocompetent patient. <i>Annals of Clinical and Translational Neurology</i> , 2016, 3, 226-232.	3.7	19
20	Quantification of differential gene expression by multiplexed targeted resequencing of cDNA. <i>Nature Communications</i> , 2017, 8, 15190.	12.8	19
21	<i>MST1R</i> mutation as a genetic cause of Lady Windermere syndrome. <i>European Respiratory Journal</i> , 2017, 49, 1601478.	6.7	18
22	Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in <i>SLC5A6</i> . <i>Npj Genomic Medicine</i> , 2019, 4, 28.	3.8	16
23	A synonymous <i>GATA2</i> variant underlying familial myeloid malignancy with striking intrafamilial phenotypic variability. <i>British Journal of Haematology</i> , 2020, 190, e297-e301.	2.5	14
24	Pathogenic variants in <i>MDFIC</i> cause recessive central conducting lymphatic anomaly with lymphedema. <i>Science Translational Medicine</i> , 2022, 14, eabm4869.	12.4	14
25	Rare <i>NOX3</i> Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of Graves' Disease. <i>Clinical Pharmacology and Therapeutics</i> , 2017, 102, 1017-1024.	4.7	12
26	Paternal mosaicism for a novel <i>PBX1</i> mutation associated with recurrent perinatal death: Phenotypic expansion of the <i>PBX1</i> -related syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1273-1277.	1.2	12
27	Pseudodiastrophic dysplasia expands the known phenotypic spectrum of defects in proteoglycan biosynthesis. <i>Journal of Medical Genetics</i> , 2020, 57, 454-460.	3.2	8
28	OUP accepted manuscript. <i>Human Molecular Genetics</i> , 2021, 30, 2068-2081.	2.9	7
29	A missense mutation underlies defective <i>SOCS4</i> function in a family with autoimmunity. <i>Journal of Internal Medicine</i> , 2015, 278, 203-210.	6.0	6
30	Immunologic defects in severe mucocutaneous HSV-2 infections: Response to IFN- γ therapy. <i>Journal of Allergy and Clinical Immunology</i> , 2016, 138, 895-898.	2.9	6
31	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. <i>Genetics in Medicine</i> , 2022, 24, 1753-1760.	2.4	6
32	Compound heterozygous variants in <i>LAMC3</i> in association with posterior periventricular nodular heterotopia. <i>BMC Medical Genomics</i> , 2021, 14, 64.	1.5	5
33	Impact of rare and common genetic variation in the interleukin-1 pathway on human cytokine responses. <i>Genome Medicine</i> , 2021, 13, 94.	8.2	5
34	Mimicking Behçet's disease: GM-CSF gain of function mutation in a family suffering from a Behçet's disease-like disorder marked by extreme pathergy. <i>Clinical and Experimental Immunology</i> , 2021, 204, 189-198.	2.6	2
35	Australian Familial Haematological Cancer Study - Findings from 15 Years of Aggregated Clinical, Genomic and Transcriptomic Data. <i>Blood</i> , 2019, 134, 1439-1439.	1.4	2
36	OPO285...Identification of rare coding variants in il-1-related pathways in patients with adult-onset still's disease. , 2018, , .		0

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37	Genetic Predisposition to Therapy-Related Myeloid Neoplasm By Rare, Deleterious Germline Variants in DNA Repair Pathway and Myeloid Driver Genes. Blood, 2018, 132, 1802-1802.	1.4	0
38	Therapy-Related Myeloid Neoplasms (T-MN) and Primary MDS (PMDS) Patients with Very Low (VL) or Low (L) IPSS-R Score Share Clinical and Biological Characteristics and Have Similar Outcome. Blood, 2018, 132, 3078-3078.	1.4	0
39	Development of a Data Portal for Aggregation and Analysis of Genomics Data in Familial Platelet Disorder with Predisposition to Myeloid Malignancy - the RUNX1.DB. Blood, 2018, 132, 5241-5241.	1.4	0
40	Familial Clustering of Hematological Malignancies: Harbingers of Wider Germline Cancer Susceptibility. Blood, 2019, 134, 3794-3794.	1.4	0