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 g-index
44 all docs	44 docs citations	44 times ranked	7144 citing authors

#	Article	IF	Citations
1	Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema. Science Translational Medicine, 2022, 14, eabm4869.	12.4	14
2	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
3	Mimicking BehÃ§etâ€™s disease: GMâ€CSF gain of function mutation in a family suffering from a BehÃ§etâ€™s diseaseâ€like disorder marked by extreme pathergy. Clinical and Experimental Immunology, 2021, 204, 189-198.	2.6	2
4	Compound heterozygous variants in LAMC3 in association with posterior periventricular nodular heterotopia. BMC Medical Genomics, 2021, 14, 64.	1.5	5
5	Impact of rare and common genetic variation in the interleukin-1 pathway on human cytokine responses. Genome Medicine, 2021, 13, 94.	8.2	5
6	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. Haematologica, 2021, 106, 3004-3007.	3.5	29
7	GATA2 deficiency syndrome: A decade of discovery. Human Mutation, 2021, 42, 1399-1421.	2.5	30
8	OUP accepted manuscript. Human Molecular Genetics, 2021, 30, 2068-2081.	2.9	7
9	Pseudodiastrophic dysplasia expands the known phenotypic spectrum of defects in proteoglycan biosynthesis. Journal of Medical Genetics, 2020, 57, 454-460.	3.2	8
10	RUNX1-mutated families show phenotype heterogeneity and a somatic mutation profile unique to germline predisposed AML. Blood Advances, 2020, 4, 1131-1144.	5.2	102
11	Paternal mosaicism for a novel <scp>PBX1</scp> mutation associated with recurrent perinatal death: Phenotypic expansion of the <scp>PBX1</scp>â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1273-1277.	1.2	12
12	Rare genetic variants in interleukin-37 link this anti-inflammatory cytokine to the pathogenesis and treatment of gout. Annals of the Rheumatic Diseases, 2020, 79, 536-544.	0.9	44
13	A synonymous GATA2 variant underlying familial myeloid malignancy with striking intrafamilial phenotypic variability. British Journal of Haematology, 2020, 190, e297-e301.	2.5	14
14	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	8.2	49
15	A systems genomics approach identifies SIGLEC15 as a susceptibility factor in recurrent vulvovaginal candidiasis. Science Translational Medicine, 2019, 11, .	12.4	38
16	The mutational burden of therapy-related myeloid neoplasms is similar to primary myelodysplastic syndrome but has a distinctive distribution. Leukemia, 2019, 33, 2842-2853.	7.2	43
17	Identification and targeted management of a neurodegenerative disorder caused by biallelic mutations in SLC5A6. Npj Genomic Medicine, 2019, 4, 28.	3.8	16
18	Australian Familial Haematological Cancer Study - Findings from 15 Years of Aggregated Clinical, Genomic and Transcriptomic Data. Blood, 2019, 134, 1439-1439.	1.4	2

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19	Familial Clustering of Hematological Malignancies: Harbingers of Wider Germline Cancer Susceptibility. Blood, 2019, 134, 3794-3794.	1.4	0
20	OP0285â€…Identification of rare coding variants in il-1-related pathways in patients with adult-onset stillâ€™s disease. , 2018, , .		0
21	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
22	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
23	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
24	Rare NOX3 Variants Confer Susceptibility to Agranulocytosis During Thyrostatic Treatment of Graves' Disease. Clinical Pharmacology and Therapeutics, 2017, 102, 1017-1024.	4.7	12
25	MST1R mutation as a genetic cause of Lady Windermere syndrome. European Respiratory Journal, 2017, 49, 1601478.	6.7	18
26	Quantification of differential gene expression by multiplexed targeted resequencing of cDNA. Nature Communications, 2017, 8, 15190.	12.8	19
27	Immunologic defects in severe mucocutaneous HSV-2 infections: Response to IFN-Î³ therapy. Journal of Allergy and Clinical Immunology, 2016, 138, 895-898.	2.9	6
28	Progressive multifocal leukoencephalopathy in an immunocompetent patient. Annals of Clinical and Translational Neurology, 2016, 3, 226-232.	3.7	19
29	A missense mutation underlies defective <scp>SOCS</scp>4 function in a family with autoimmunity. Journal of Internal Medicine, 2015, 278, 203-210.	6.0	6
30	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. Human Molecular Genetics, 2015, 24, 2000-2010.	2.9	25
31	Human TLR10 is an anti-inflammatory pattern-recognition receptor. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, E4478-84.	7.1	211
32	<scp>MLL2</scp> mutation detection in 86 patients with Kabuki syndrome: a genotypeâ€“phenotype study. Clinical Genetics, 2013, 84, 539-545.	2.0	85
33	Functional genomics identifies type I interferon pathway as central for host defense against Candida albicans. Nature Communications, 2013, 4, 1342.	12.8	157
34	STAT1Mutations in Autosomal Dominant Chronic Mucocutaneous Candidiasis. New England Journal of Medicine, 2011, 365, 54-61.	27.0	614
35	Next-Generation Sequencing of a 40 Mb Linkage Interval Reveals TSPAN12 Mutations in Patients with Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 240-247.	6.2	202
36	Exome Sequencing Identifies WDR35 Variants Involved in Sensenbrenner Syndrome. American Journal of Human Genetics, 2010, 87, 418-423.	6.2	260

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37	Targeted Next-Generation Sequencing of a 12.5 Mb Homozygous Region Reveals ANO10 Mutations in Patients with Autosomal-Recessive Cerebellar Ataxia. American Journal of Human Genetics, 2010, 87, 813-819.	6.2	125
38	Massively parallel sequencing of ataxia genes after array-based enrichment. Human Mutation, 2010, 31, 494-499.	2.5	86
39	De novo mutations of SETBP1 cause Schinzel-Giedion syndrome. Nature Genetics, 2010, 42, 483-485.	21.4	417
40	A de novo paradigm for mental retardation. Nature Genetics, 2010, 42, 1109-1112.	21.4	751