

Karine Megy

List of Publications by Year in descending order

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Version: 2024-02-01

30
papers

6,463
citations

257450

24
h-index

434195

31
g-index

37
all docs

37
docs citations

37
times ranked

13544
citing authors

#	ARTICLE	IF	CITATIONS
1	Multiparameter platelet function analysis of bleeding patients with a prolonged platelet function analyser closure time. <i>British Journal of Haematology</i> , 2022, 196, 1388-1400.	2.5	2
2	Refinements and considerations for trio whole-genome sequence analysis when investigating Mendelian diseases presenting in early childhood. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100113.	1.7	4
3	Whole Genome Interpretation for a Family of Five. <i>Frontiers in Genetics</i> , 2021, 12, 535123.	2.3	3
4	Advances in understanding the pathogenesis of hereditary macrothrombocytopenia. <i>British Journal of Haematology</i> , 2021, 195, 25-45.	2.5	9
5	The Polygenic and Monogenic Basis of Blood Traits and Diseases. <i>Cell</i> , 2020, 182, 1214-1231.e11.	28.9	388
6	Development and validation of a universal blood donor genotyping platform: a multinational prospective study. <i>Blood Advances</i> , 2020, 4, 3495-3506.	5.2	31
7	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. <i>Blood</i> , 2020, 136, 1956-1967.	1.4	34
8	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , 2020, 583, 90-95.	27.8	148
9	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020, 583, 96-102.	27.8	338
10	Transcriptomic analysis of insecticide resistance in the lymphatic filariasis vector <i>Culex quinquefasciatus</i> . <i>Scientific Reports</i> , 2019, 9, 11406.	3.3	11
11	Germline mutations in the transcription factor IKZF5 cause thrombocytopenia. <i>Blood</i> , 2019, 134, 2070-2081.	1.4	29
12	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2019, 134, 2082-2091.	1.4	131
13	Whole genome sequencing reveals that genetic conditions are frequent in intensively ill children. <i>Intensive Care Medicine</i> , 2019, 45, 627-636.	8.2	183
14	How common are single gene mutations as a cause for lacunar stroke?. <i>Neurology</i> , 2019, 93, e2007-e2020.	1.1	26
15	Loss-of-function nuclear factor κ B subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 1285-1296.	2.9	185
16	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 75-90.	6.2	343
17	Phenotypic Characterization of <i>EIF2AK4</i> Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. <i>Circulation</i> , 2017, 136, 2022-2033.	1.6	111
18	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2016, 127, 2791-2803.	1.4	157

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19	The Allelic Landscape of Human Blood Cell Trait Variation and Links to Common Complex Disease. <i>Cell</i> , 2016, 167, 1415-1429.e19.	28.9	1,052
20	Genomic insights into the Ixodes scapularis tick vector of Lyme disease. <i>Nature Communications</i> , 2016, 7, 10507.	12.8	450
21	Genome Sequence of the Tsetse Fly (<i>Glossina morsitans</i>): Vector of African Trypanosomiasis. <i>Science</i> , 2014, 344, 380-386.	12.6	254
22	VectorBase: improvements to a bioinformatics resource for invertebrate vector genomics. <i>Nucleic Acids Research</i> , 2012, 40, D729-D734.	14.5	143
23	Ensembl Genomes: an integrative resource for genome-scale data from non-vertebrate species. <i>Nucleic Acids Research</i> , 2012, 40, D91-D97.	14.5	179
24	Sequencing of <i>Culex quinquefasciatus</i> Establishes a Platform for Mosquito Comparative Genomics. <i>Science</i> , 2010, 330, 86-88.	12.6	424
25	Ensembl's 10th year. <i>Nucleic Acids Research</i> , 2010, 38, D557-D562.	14.5	251
26	Pathogenomics of <i>Culex quinquefasciatus</i> and Meta-Analysis of Infection Responses to Diverse Pathogens. <i>Science</i> , 2010, 330, 88-90.	12.6	150
27	VectorBase: a data resource for invertebrate vector genomics. <i>Nucleic Acids Research</i> , 2009, 37, D583-D587.	14.5	234
28	Comparative genomics allows the discovery of cis-regulatory elements in mosquitoes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 3053-3058.	7.1	45
29	VectorBase: a home for invertebrate vectors of human pathogens. <i>Nucleic Acids Research</i> , 2007, 35, D503-D505.	14.5	107
30	Genome Sequence of <i>Aedes aegypti</i> , a Major Arbovirus Vector. <i>Science</i> , 2007, 316, 1718-1723.	12.6	1,025