

Karyn Megy

List of Publications by Year in descending order

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Version: 2024-02-01

28
papers

5,326
citations

279798

23
h-index

477307

29
g-index

35
all docs

35
docs citations

35
times ranked

13105
citing authors

#	ARTICLE	IF	CITATIONS
1	Exploring the relevance of NUP93 variants in steroid-resistant nephrotic syndrome using next generation sequencing and a fly kidney model. <i>Pediatric Nephrology</i> , 2022, 37, 2643-2656.	1.7	5
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	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, .	3.6	29
4	Advances in understanding the pathogenesis of hereditary macrothrombocytopenia. <i>British Journal of Haematology</i> , 2021, 195, 25-45.	2.5	9
5	Next-generation sequencing for the diagnosis of <i>MYH9</i> : Predicting pathogenic variants. <i>Human Mutation</i> , 2020, 41, 277-290.	2.5	30
6	The Polygenic and Monogenic Basis of Blood Traits and Diseases. <i>Cell</i> , 2020, 182, 1214-1231.e11.	28.9	388
7	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
8	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. <i>Blood</i> , 2020, 136, 1956-1967.	1.4	34
9	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , 2020, 583, 90-95.	27.8	148
10	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020, 583, 96-102.	27.8	338
11	Mutational and phenotypic characterization of hereditary hemorrhagic telangiectasia. <i>Blood</i> , 2020, 136, 1907-1918.	1.4	40
12	Germline mutations in the transcription factor <i>IKZF5</i> cause thrombocytopenia. <i>Blood</i> , 2019, 134, 2070-2081.	1.4	29
13	Curated disease-causing genes for bleeding, thrombotic, and platelet disorders: Communication from the SSC of the ISTH. <i>Journal of Thrombosis and Haemostasis</i> , 2019, 17, 1253-1260.	3.8	56
14	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2019, 134, 2082-2091.	1.4	131
15	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
16	How common are single gene mutations as a cause for lacunar stroke?. <i>Neurology</i> , 2019, 93, e2007-e2020.	1.1	26
17	Loss-of-function nuclear factor κ B subunit 1 (<i>NFKB1</i>) 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
18	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. <i>Genome Medicine</i> , 2018, 10, 95.	8.2	111

#	ARTICLE	IF	CITATIONS
19	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
20	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. American Journal of Human Genetics, 2018, 103, 3-18.	6.2	46
21	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	6.2	26
22	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. American Journal of Human Genetics, 2017, 100, 75-90.	6.2	343
23	Phenotypic Characterization of <i>EIF2AK4</i> Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
24	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood, 2016, 127, 2791-2803.	1.4	157
25	The Allelic Landscape of Human Blood Cell Trait Variation and Links to Common Complex Disease. Cell, 2016, 167, 1415-1429.e19.	28.9	1,052
26	ArrayExpress update—simplifying data submissions. Nucleic Acids Research, 2015, 43, D1113-D1116.	14.5	688
27	Comparative genomics allows the discovery of <i>cis</i> -regulatory elements in mosquitoes. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 3053-3058.	7.1	45
28	Genome Sequence of <i>Aedes aegypti</i> , a Major Arbovirus Vector. Science, 2007, 316, 1718-1723.	12.6	1,025