Karyn Megy

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/11070806/publications.pdf

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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	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
2	Genome Sequence of Aedes aegypti, a Major Arbovirus Vector. Science, 2007, 316, 1718-1723.	12.6	1,025
3	ArrayExpress updateâ€"simplifying data submissions. Nucleic Acids Research, 2015, 43, D1113-D1116.	14.5	688
4	The Polygenic and Monogenic Basis of Blood Traits and Diseases. Cell, 2020, 182, 1214-1231.e11.	28.9	388
5	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
6	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
7	Loss-of-function nuclear factor ÎºB subunit 1 (NFKB1) variants are the most common monogenic cause of common variable immunodeficiency in Europeans. Journal of Allergy and Clinical Immunology, 2018, 142, 1285-1296.	2.9	185
8	Whole genome sequencing reveals that genetic conditions are frequent in intensively ill children. Intensive Care Medicine, 2019, 45, 627-636.	8.2	183
9	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood, 2016, 127, 2791-2803.	1.4	157
10	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95.	27.8	148
11	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. Blood, 2019, 134, 2082-2091.	1.4	131
12	Phenotypic Characterization of EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
13	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. Genome Medicine, 2018, 10, 95.	8.2	111
14	Curated diseaseâ€causing genes for bleeding, thrombotic, and platelet disorders: Communication from the SSC of the ISTH. Journal of Thrombosis and Haemostasis, 2019, 17, 1253-1260.	3.8	56
15	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
16	Comparative genomics allows the discovery of cis -regulatory elements in mosquitoes. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 3053-3058.	7.1	45
17	Mutational and phenotypic characterization of hereditary hemorrhagic telangiectasia. Blood, 2020, 136, 1907-1918.	1.4	40
18	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36

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19	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. Blood, 2020, 136, 1956-1967.	1.4	34
20	Development and validation of a universal blood donor genotyping platform: a multinational prospective study. Blood Advances, 2020, 4, 3495-3506.	5.2	31
21	Nextâ€generation sequencing for the diagnosis ofMYH9â€RD: Predicting pathogenic variants. Human Mutation, 2020, 41, 277-290.	2.5	30
22	Germline mutations in the transcription factor IKZF5 cause thrombocytopenia. Blood, 2019, 134, 2070-2081.	1.4	29
23	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
24	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
25	How common are single gene mutations as a cause for lacunar stroke?. Neurology, 2019, 93, e2007-e2020.	1.1	26
26	Advances in understanding the pathogenesis of hereditary macrothrombocytopenia. British Journal of Haematology, 2021, 195, 25-45.	2.5	9
27	Exploring the relevance of NUP93 variants in steroid-resistant nephrotic syndrome using next generation sequencing and a fly kidney model. Pediatric Nephrology, 2022, 37, 2643-2656.	1.7	5
28	Refinements and considerations for trio whole-genome sequence analysis when investigating Mendelian diseases presenting in early childhood. Human Genetics and Genomics Advances, 2022, 3, 100113.	1.7	4