

Daniel Greene

List of Publications by Year in descending order

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Version: 2024-02-01

31
papers

3,131
citations

257101

24
h-index

433756

31
g-index

37
all docs

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docs citations

37
times ranked

6839
citing authors

#	ARTICLE	IF	CITATIONS
1	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, .	1.6	29
2	Identification of a homozygous recessive variant in <i>PTGS1</i> resulting in a congenital aspirin-like defect in platelet function. <i>Haematologica</i> , 2021, 106, 1423-1432.	1.7	7
3	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. <i>Nucleic Acids Research</i> , 2021, 49, 9686-9695.	6.5	14
4	Next-generation sequencing for the diagnosis of <i>MYH9</i> : Predicting pathogenic variants. <i>Human Mutation</i> , 2020, 41, 277-290.	1.1	30
5	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. <i>Blood</i> , 2020, 136, 1956-1967.	0.6	34
6	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , 2020, 583, 90-95.	13.7	148
7	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020, 583, 96-102.	13.7	338
8	Monoallelic loss-of-function <i>THPO</i> variants cause heritable thrombocytopenia. <i>Blood Advances</i> , 2020, 4, 920-924.	2.5	10
9	Characterization of the clinical and immunologic phenotype and management of 157 individuals with 56 distinct heterozygous <i>NFKB1</i> mutations. <i>Journal of Allergy and Clinical Immunology</i> , 2020, 146, 901-911.	1.5	78
10	Germline mutations in the transcription factor <i>IKZF5</i> cause thrombocytopenia. <i>Blood</i> , 2019, 134, 2070-2081.	0.6	29
11	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2019, 134, 2082-2091.	0.6	131
12	A novel missense variant in <i>SLC18A2</i> causes recessive brain monoamine vesicular transport disease and absent serotonin in platelets. <i>JIMD Reports</i> , 2019, 47, 9-16.	0.7	18
13	Abnormal differentiation of B cells and megakaryocytes in patients with Roifman syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 142, 630-646.	1.5	36
14	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.	1.5	185
15	De Novo Truncating Mutations in <i>WASF1</i> Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018, 103, 144-153.	2.6	36
16	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 3-18.	2.6	46
17	The Human Phenotype Ontology in 2017. <i>Nucleic Acids Research</i> , 2017, 45, D865-D876.	6.5	699
18	ontologyX: a suite of R packages for working with ontological data. <i>Bioinformatics</i> , 2017, 33, 1104-1106.	1.8	86

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19	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. <i>American Journal of Human Genetics</i> , 2017, 100, 334-342.	2.6	26
20	Rare variants in GP1BB are responsible for autosomal dominant macrothrombocytopenia. <i>Blood</i> , 2017, 129, 520-524.	0.6	42
21	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. <i>Bioinformatics</i> , 2017, 33, 2421-2423.	1.8	40
22	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.	2.6	343
23	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
24	Expanded repertoire of RASGRP2 variants responsible for platelet dysfunction and severe bleeding. <i>Blood</i> , 2017, 130, 1026-1030.	0.6	38
25	A Fast Association Test for Identifying Pathogenic Variants Involved in Rare Diseases. <i>American Journal of Human Genetics</i> , 2017, 101, 104-114.	2.6	31
26	A dominant gain-of-function mutation in universal tyrosine kinase <i>SRC</i> causes thrombocytopenia, myelofibrosis, bleeding, and bone pathologies. <i>Science Translational Medicine</i> , 2016, 8, 328ra30.	5.8	87
27	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. <i>Blood</i> , 2016, 127, 2791-2803.	0.6	157
28	A gain-of-function variant in DIAPH1 causes dominant macrothrombocytopenia and hearing loss. <i>Blood</i> , 2016, 127, 2903-2914.	0.6	121
29	Phenotype Similarity Regression for Identifying the Genetic Determinants of Rare Diseases. <i>American Journal of Human Genetics</i> , 2016, 98, 490-499.	2.6	49
30	Human phenotype ontology annotation and cluster analysis to unravel genetic defects in 707 cases with unexplained bleeding and platelet disorders. <i>Genome Medicine</i> , 2015, 7, 36.	3.6	119
31	Transureteral Natural Orifice Translumenal Endoscopic Surgery Nephrectomy: A Feasibility Study in the Porcine Model. <i>Videourology (New Rochelle, N Y)</i> , 2011, 25, .	0.1	0