Daniel Greene

List of Publications by Year
in descending order

31	3,131	257450 24	434195 31
papers	citations	h-index	g-index

37	37	37	6839
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
2	Identification of a homozygous recessive variant in <i>PTGS1</i> resulting in a congenital aspirin-like defect in platelet function. Haematologica, 2021, 106, 1423-1432.	3.5	7
3	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. Nucleic Acids Research, 2021, 49, 9686-9695.	14.5	14
4	Nextâ€generation sequencing for the diagnosis ofMYH9â€RD: Predicting pathogenic variants. Human Mutation, 2020, 41, 277-290.	2.5	30
5	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. Blood, 2020, 136, 1956-1967.	1.4	34
6	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95.	27.8	148
7	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
8	MonoallelicÂloss-of-function THPOÂvariants cause heritable thrombocytopenia. Blood Advances, 2020, 4, 920-924.	5.2	10
9	Characterization of the clinical and immunologic phenotype and management of 157 individuals with 56 distinct heterozygous NFKB1 mutations. Journal of Allergy and Clinical Immunology, 2020, 146, 901-911.	2.9	78
10	Germline mutations in the transcription factor IKZF5 cause thrombocytopenia. Blood, 2019, 134, 2070-2081.	1.4	29
11	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. Blood, 2019, 134, 2082-2091.	1.4	131
12	A novel missense variant in SLC18A2 causes recessive brain monoamine vesicular transport disease and absent serotonin in platelets. JIMD Reports, 2019, 47, 9-16.	1.5	18
13	Abnormal differentiation of B cells and megakaryocytes in patients with Roifman syndrome. Journal of Allergy and Clinical Immunology, 2018, 142, 630-646.	2.9	36
14	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
15	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	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. American Journal of Human Genetics, 2018, 103, 3-18.	6.2	46
17	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
18	ontologyX: a suite of R packages for working with ontological data. Bioinformatics, 2017, 33, 1104-1106.	4.1	86

#	Article	IF	Citations
19	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
20	Rare variants in GP1BB are responsible for autosomal dominant macrothrombocytopenia. Blood, 2017, 129, 520-524.	1.4	42
21	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	4.1	40
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 EIF2AK4 Mutation Carriers in a Large Cohort of Patients Diagnosed Clinically With Pulmonary Arterial Hypertension. Circulation, 2017, 136, 2022-2033.	1.6	111
24	Expanded repertoire of RASGRP2 variants responsible for platelet dysfunction and severe bleeding. Blood, 2017, 130, 1026-1030.	1.4	38
25	A Fast Association Test for Identifying Pathogenic Variants Involved in Rare Diseases. American Journal of Human Genetics, 2017, 101, 104-114.	6.2	31
26	A dominant gain-of-function mutation in universal tyrosine kinase SRC causes thrombocytopenia, myelofibrosis, bleeding, and bone pathologies. Science Translational Medicine, 2016, 8, 328ra30.	12.4	87
27	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood, 2016, 127, 2791-2803.	1.4	157
28	A gain-of-function variant in DIAPH1 causes dominant macrothrombocytopenia and hearing loss. Blood, 2016, 127, 2903-2914.	1.4	121
29	Phenotype Similarity Regression for Identifying the Genetic Determinants of Rare Diseases. American Journal of Human Genetics, 2016, 98, 490-499.	6.2	49
30	Human phenotype ontology annotation and cluster analysis to unravel genetic defects in 707 cases with unexplained bleeding and platelet disorders. Genome Medicine, 2015, 7, 36.	8.2	119
31	Transureteral Natural Orifice Translumenal Endoscopic Surgery Nephrectomy: A Feasibility Study in the Porcine Model. Videourology (New Rochelle, N Y), 2011, 25, .	0.1	0