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	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
2	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
3	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
4	Loss-of-function nuclear factor Î¹B 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
5	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood, 2016, 127, 2791-2803.	1.4	157
6	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. Nature, 2020, 583, 90-95.	27.8	148
7	Diagnostic high-throughput sequencing of 2396 patients with bleeding, thrombotic, and platelet disorders. Blood, 2019, 134, 2082-2091.	1.4	131
8	A gain-of-function variant in DIAPH1 causes dominant macrothrombocytopenia and hearing loss. Blood, 2016, 127, 2903-2914.	1.4	121
9	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
10	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
11	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
12	ontologyX: a suite of R packages for working with ontological data. Bioinformatics, 2017, 33, 1104-1106.	4.1	86
13	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
14	Phenotype Similarity Regression for Identifying the Genetic Determinants of Rare Diseases. American Journal of Human Genetics, 2016, 98, 490-499.	6.2	49
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	Rare variants in GP1BB are responsible for autosomal dominant macrothrombocytopenia. Blood, 2017, 129, 520-524.	1.4	42
17	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	4.1	40
18	Expanded repertoire of RASGRP2 variants responsible for platelet dysfunction and severe bleeding. Blood, 2017, 130, 1026-1030.	1.4	38

#	Article	IF	Citations
19	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
20	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	36
21	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. Blood, 2020, 136, 1956-1967.	1.4	34
22	A Fast Association Test for Identifying Pathogenic Variants Involved in Rare Diseases. American Journal of Human Genetics, 2017, 101, 104-114.	6.2	31
23	Nextâ€generation sequencing for the diagnosis ofMYH9â€RD: Predicting pathogenic variants. Human Mutation, 2020, 41, 277-290.	2.5	30
24	Germline mutations in the transcription factor IKZF5 cause thrombocytopenia. Blood, 2019, 134, 2070-2081.	1.4	29
25	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
26	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
27	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
28	MitoPhen database: a human phenotype ontology-based approach to identify mitochondrial DNA diseases. Nucleic Acids Research, 2021, 49, 9686-9695.	14.5	14
29	MonoallelicÂloss-of-function THPOÂvariants cause heritable thrombocytopenia. Blood Advances, 2020, 4, 920-924.	5.2	10
30	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
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