James O'sullivan

List of Publications by Year
in descending order

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34 papers	3,122 citations	236925 25 h-index	377865 34 g-index
35 all docs	35 docs citations	35 times ranked	6198 citing authors

#	Article	IF	Citations
1	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. Nature Genetics, 2014, 46, 503-509.	21.4	490
2	Germline Mutations in SUFU Cause Gorlin Syndromeâ€“Associated Childhood Medulloblastoma and Redefine the Risk Associated With PTCH1 Mutations. Journal of Clinical Oncology, 2014, 32, 4155-4161.	1.6	236
3	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	21.4	234
4	Identification of mutations in CUL7 in 3-M syndrome. Nature Genetics, 2005, 37, 1119-1124.	21.4	158
5	Missense Mutations in a Retinal Pigment Epithelium Protein, Bestrophin-1, Cause Retinitis Pigmentosa. American Journal of Human Genetics, 2009, 85, 581-592.	6.2	156
6	Protein Kinase CÎ´ Deficiency Causes Mendelian Systemic Lupus Erythematosus With B Cellâ€Ðefective Apoptosis and Hyperproliferation. Arthritis and Rheumatism, 2013, 65, 2161-2171.	6.7	155
7	Personalized Diagnosis and Management of Congenital Cataract by Next-Generation Sequencing. Ophthalmology, 2014, 121, 2124-2137.e2.	5.2	153
8	Whole-Exome Sequencing Identifies FAM20A Mutations as a Cause of Amelogenesis Imperfecta and Gingival Hyperplasia Syndrome. American Journal of Human Genetics, 2011, 88, 616-620.	6.2	147
9	A paradigm shift in the delivery of services for diagnosis of inherited retinal disease. Journal of Medical Genetics, 2012, 49, 322-326.	3.2	143
10	Molecular findings from 537 individuals with inherited retinal disease. Journal of Medical Genetics, 2016, 53, 761-767.	3.2	135
11	Whole Genome Sequencing Increases Molecular Diagnostic Yield Compared with Current Diagnostic Testing for Inherited Retinal Disease. Ophthalmology, 2016, 123, 1143-1150.	5.2	122
12	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
13	Exome Sequencing Identifies CCDC8 Mutations in 3-M Syndrome, Suggesting that CCDC8 Contributes in a Pathway with CUL7 and OBSL1 to Control Human Growth. American Journal of Human Genetics, 2011, 89, 148-153.	6.2	98
14	Mutations in LZTR1 add to the complex heterogeneity of schwannomatosis. Neurology, 2015, 84, 141-147.	1.1	90
15	MiR-204 is responsible for inherited retinal dystrophy associated with ocular coloboma. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E3236-45.	7.1	90
16	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
17	Exome Sequence Identifies RIPK4 as the Bartsocas- Papas Syndrome Locus. American Journal of Human Genetics, 2012, 90, 69-75.	6.2	82
18	Compound Heterozygosity of Low-Frequency Promoter Deletions and Rare Loss-of-Function Mutations in TXNL4A Causes Burn-McKeown Syndrome. American Journal of Human Genetics, 2014, 95, 698-707.	6.2	55

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19	Autozygosity Mapping with Exome Sequence Data. Human Mutation, 2013, 34, 50-56.	2.5	49
20	Childhood-Onset Autosomal Recessive Bestrophinopathy. JAMA Ophthalmology, 2011, 129, 1088.	2.4	46
21	Expanding the genetic and phenotypic spectrum of popliteal pterygium disorders. American Journal of Medical Genetics, Part A, 2015, 167, 545-552.	1.2	38
22	Contribution of rare and predicted pathogenic gene variants to childhood-onset lupus: a large, genetic panel analysis of British and French cohorts. Lancet Rheumatology, The, 2020, 2, e99-e109.	3.9	38
23	Intracranial Angiomatoid Fibrous Histiocytoma with EWSR1-CREB Family Fusions: A Report of 2 Pediatric Cases. World Neurosurgery, 2019, 126, 113-119.	1.3	35
24	Non lethal Raine syndrome and differential diagnosis. European Journal of Medical Genetics, 2016, 59, 577-583.	1.3	33
25	Analysis of head and neck carcinoma progression reveals novel and relevant stage-specific changes associated with immortalisation and malignancy. Scientific Reports, 2019, 9, 11992.	3.3	32
26	Pinpointing clinical diagnosis through whole exome sequencing to direct patient care: a case of Senior-Loken syndrome. Lancet, The, 2015, 385, 1916.	13.7	29
27	Bi-allelic variants in the mitochondrial RNase P subunit PRORP cause mitochondrial tRNA processing defects and pleiotropic multisystem presentations. American Journal of Human Genetics, 2021, 108, 2195-2204.	6.2	26
28	Exome Sequencing Identifies a Dominant <i>TNNT3</i> Mutation in a Large Family with Distal Arthrogryposis. Molecular Syndromology, 2014, 5, 218-228.	0.8	11
29	Novel C8orf37 mutations cause retinitis pigmentosa in consanguineous families of Pakistani origin. Molecular Vision, 2015, 21, 236-43.	1.1	10
30	A homozygous missense variant in CHRM3 associated with familial urinary bladder disease. Clinical Genetics, 2019, 96, 515-520.	2.0	9
31	Marfanoid habitus is a nonspecific feature of Perrault syndrome. Clinical Dysmorphology, 2017, 26, 200-204.	0.3	7
32	Ligase IV syndrome can present with microcephaly and radial ray anomalies similar to Fanconi anaemia plus fatal kidney malformations. European Journal of Medical Genetics, 2020, 63, 103974.	1.3	5
33	Biallelic loss of function variants in STAG3 result in primary ovarian insufficiency. Reproductive BioMedicine Online, 2021, 43, 899-902.	2.4	5
34	Genome sequencing identifies a large deletion at 13q32.1 as the cause of microcoria and childhoodâ€onset glaucoma. Acta Ophthalmologica, 2017, 95, e249-e250.	1.1	4