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	Biallelic loss of function variants in STAG3 result in primary ovarian insufficiency. Reproductive BioMedicine Online, 2021, 43, 899-902.	2.4	5
2	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
3	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
4	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
5	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
6	A homozygous missense variant in CHRM3 associated with familial urinary bladder disease. Clinical Genetics, 2019, 96, 515-520.	2.0	9
7	Intracranial Angiomatoid Fibrous Histiocytoma with EWSR1-CREB Family Fusions: A Report of 2 Pediatric Cases. World Neurosurgery, 2019, 126, 113-119.	1.3	35
8	Marfanoid habitus is a nonspecific feature of Perrault syndrome. Clinical Dysmorphology, 2017, 26, 200-204.	0.3	7
9	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
10	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
11	Molecular findings from 537 individuals with inherited retinal disease. Journal of Medical Genetics, 2016, 53, 761-767.	3.2	135
12	Non lethal Raine syndrome and differential diagnosis. European Journal of Medical Genetics, 2016, 59, 577-583.	1.3	33
13	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
14	Whole Genome Sequencing Increases Molecular Diagnostic Yield Compared with Current Diagnostic Testing for Inherited Retinal Disease. Ophthalmology, 2016, 123, 1143-1150.	5.2	122
15	Mutations in LZTR1 add to the complex heterogeneity of schwannomatosis. Neurology, 2015, 84, 141-147.	1.1	90
16	Expanding the genetic and phenotypic spectrum of popliteal pterygium disorders. American Journal of Medical Genetics, Part A, 2015, 167, 545-552.	1.2	38
17	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
18	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

#	Article	IF	Citations
19	Novel C8orf37 mutations cause retinitis pigmentosa in consanguineous families of Pakistani origin. Molecular Vision, 2015, 21, 236-43.	1.1	10
20	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
21	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
22	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
23	Personalized Diagnosis and Management of Congenital Cataract by Next-Generation Sequencing. Ophthalmology, 2014, 121, 2124-2137.e2.	5.2	153
24	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
25	Autozygosity Mapping with Exome Sequence Data. Human Mutation, 2013, 34, 50-56.	2.5	49
26	Protein Kinase CÎ´ Deficiency Causes Mendelian Systemic Lupus Erythematosus With B Cellâ€Đefective Apoptosis and Hyperproliferation. Arthritis and Rheumatism, 2013, 65, 2161-2171.	6.7	155
27	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
28	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	21.4	234
29	Exome Sequence Identifies RIPK4 as the Bartsocas- Papas Syndrome Locus. American Journal of Human Genetics, 2012, 90, 69-75.	6.2	82
30	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
31	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
32	Childhood-Onset Autosomal Recessive Bestrophinopathy. JAMA Ophthalmology, 2011, 129, 1088.	2.4	46
33	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
34	Identification of mutations in CUL7 in 3-M syndrome. Nature Genetics, 2005, 37, 1119-1124.	21.4	158