Eamonn Sheridan

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5859344/publications.pdf

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25 papers	1,229 citations	471509 17 h-index	580821 25 g-index
27 all docs	27 docs citations	27 times ranked	2964 citing authors

#	Article	IF	Citations
1	Risk factors for congenital anomaly in a multiethnic birth cohort: an analysis of the Born in Bradford study. Lancet, The, 2013, 382, 1350-1359.	13.7	190
2	CCDC151 Mutations Cause Primary Ciliary Dyskinesia by Disruption of the Outer Dynein Arm Docking Complex Formation. American Journal of Human Genetics, 2014, 95, 257-274.	6.2	149
3	Loss-of-Function Mutations in TBC1D20 Cause Cataracts and Male Infertility in blind sterile Mice and Warburg Micro Syndrome in Humans. American Journal of Human Genetics, 2013, 93, 1001-1014.	6.2	119
4	Diagnosis of Constitutional Mismatch Repair-Deficiency Syndrome Based on Microsatellite Instability and Lymphocyte Tolerance to Methylating Agents. Gastroenterology, 2015, 149, 1017-1029.e3.	1.3	76
5	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	6.2	71
6	HEATR2 Plays a Conserved Role in Assembly of the Ciliary Motile Apparatus. PLoS Genetics, 2014, 10, e1004577.	3.5	67
7	Biallelic Mutations in PDE10A Lead to Loss of Striatal PDE10A and a Hyperkinetic Movement Disorder with Onset in Infancy. American Journal of Human Genetics, 2016, 98, 735-743.	6.2	65
8	High prevalence of CCDC103 p.His154Pro mutation causing primary ciliary dyskinesia disrupts protein oligomerisation and is associated with normal diagnostic investigations. Thorax, 2018, 73, 157-166.	5.6	63
9	Genome-wide association study identifies susceptibility loci for B-cell childhood acute lymphoblastic leukemia. Nature Communications, 2018, 9, 1340.	12.8	58
10	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
11	DNAAF1 links heart laterality with the AAA+ ATPase RUVBL1 and ciliary intraflagellar transport. Human Molecular Genetics, 2018, 27, 529-545.	2.9	45
12	Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. American Journal of Human Genetics, 2015, 96, 948-954.	6.2	42
13	Use of zebrafish models to investigate rare human disease. Journal of Medical Genetics, 2018, 55, 641-649.	3.2	42
14	Cas9-based enrichment and single-molecule sequencing for precise characterization of genomic duplications. Laboratory Investigation, 2020, 100, 135-146.	3.7	33
15	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	8.1	31
16	The 9p21.3 risk of childhood acute lymphoblastic leukaemia is explained by a rare high-impact variant in CDKN2A. Scientific Reports, 2015, 5, 15065.	3.3	24
17	A tubulin alpha 8 mouse knockout model indicates a likely role in spermatogenesis but not in brain development. PLoS ONE, 2017, 12, e0174264.	2.5	23
18	Fine-scale population structure and demographic history of British Pakistanis. Nature Communications, 2021, 12, 7189.	12.8	21

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19	Deficiency of the myogenic factor MyoD causes a perinatally lethal fetal akinesia. Journal of Medical Genetics, 2016, 53, 264-269.	3.2	15
20	Differentiation of MISSLA and Fanconi anaemia by computer-aided image analysis and presentation of two novel MISSLA siblings. European Journal of Human Genetics, 2019, 27, 1827-1835.	2.8	9
21	Novel loss-of-function mutation in HERC2 is associated with severe developmental delay and paediatric lethality. Journal of Medical Genetics, 2021, 58, 334-341.	3.2	9
22	Whole Exon Deletion in the GFAP Gene Is a Novel Molecular Mechanism Causing Alexander Disease. Neuropediatrics, 2018, 49, 118-122.	0.6	6
23	A Chromosome 7 Pericentric Inversion Defined at Single-Nucleotide Resolution Using Diagnostic Whole Genome Sequencing in a Patient with Hand-Foot-Genital Syndrome. PLoS ONE, 2016, 11, e0157075.	2.5	5
24	Long-read sequencing to resolve the parent of origin of a de novo pathogenicUBE3Avariant. Journal of Medical Genetics, 2022, 59, 1082-1086.	3.2	4
25	Analysis of the Born in Bradford birth cohort â€“ Authors' reply. Lancet, The, 2014, 383, 123.	13.7	3