

Joseph G Gleeson

List of Publications by Year in descending order

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Version: 2024-02-01

146
papers

11,988
citations

38660

50
h-index

30848

102
g-index

164
all docs

164
docs citations

164
times ranked

20421
citing authors

#	ARTICLE	IF	CITATIONS
1	doublecortin, a Brain-Specific Gene Mutated in Human X-Linked Lissencephaly and Double Cortex Syndrome, Encodes a Putative Signaling Protein. <i>Cell</i> , 1998, 92, 63-72.	13.5	1,007
2	De novo somatic mutations in components of the PI3K-AKT3-mTOR pathway cause hemimegalencephaly. <i>Nature Genetics</i> , 2012, 44, 941-945.	9.4	628
3	Aberrant methylation of tRNA links cellular stress to neurodevelopmental disorders. <i>EMBO Journal</i> , 2014, 33, 2020-2039.	3.5	490
4	Functional genomic screen for modulators of ciliogenesis and cilium length. <i>Nature</i> , 2010, 464, 1048-1051.	13.7	473
5	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. <i>Science</i> , 2014, 343, 506-511.	6.0	466
6	Lis1 and doublecortin function with dynein to mediate coupling of the nucleus to the centrosome in neuronal migration. <i>Journal of Cell Biology</i> , 2004, 165, 709-721.	2.3	390
7	Ndel1 Operates in a Common Pathway with LIS1 and Cytoplasmic Dynein to Regulate Cortical Neuronal Positioning. <i>Neuron</i> , 2004, 44, 263-277.	3.8	334
8	Characterization of Greater Middle Eastern genetic variation for enhanced disease gene discovery. <i>Nature Genetics</i> , 2016, 48, 1071-1076.	9.4	314
9	Mutations in <i>BCKD-kinase</i> Lead to a Potentially Treatable Form of Autism with Epilepsy. <i>Science</i> , 2012, 338, 394-397.	6.0	272
10	Impaired Amino Acid Transport at the Blood Brain Barrier Is a Cause of Autism Spectrum Disorder. <i>Cell</i> , 2016, 167, 1481-1494.e18.	13.5	265
11	The mutation significance cutoff: gene-level thresholds for variant predictions. <i>Nature Methods</i> , 2016, 13, 109-110.	9.0	249
12	Primary Cilia in the Developing and Mature Brain. <i>Neuron</i> , 2014, 82, 511-521.	3.8	243
13	Zika Virus Infects Neural Progenitors in the Adult Mouse Brain and Alters Proliferation. <i>Cell Stem Cell</i> , 2016, 19, 593-598.	5.2	242
14	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. <i>Cell</i> , 2014, 157, 651-663.	13.5	228
15	Exome Sequencing Can Improve Diagnosis and Alter Patient Management. <i>Science Translational Medicine</i> , 2012, 4, 138ra78.	5.8	226
16	Primary cilia in neurodevelopmental disorders. <i>Nature Reviews Neurology</i> , 2014, 10, 27-36.	4.9	215
17	An siRNA-based functional genomics screen for the identification of regulators of ciliogenesis and ciliopathy genes. <i>Nature Cell Biology</i> , 2015, 17, 1074-1087.	4.6	215
18	The human gene damage index as a gene-level approach to prioritizing exome variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 13615-13620.	3.3	213

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19	Diagnosis of genetic diseases in seriously ill children by rapid whole-genome sequencing and automated phenotyping and interpretation. <i>Science Translational Medicine</i> , 2019, 11, .	5.8	203
20	Inactivating mutations in MFSD2A, required for omega-3 fatty acid transport in brain, cause a lethal microcephaly syndrome. <i>Nature Genetics</i> , 2015, 47, 809-813.	9.4	180
21	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. <i>American Journal of Human Genetics</i> , 2012, 91, 1051-1064.	2.6	179
22	Paternally inherited cis-regulatory structural variants are associated with autism. <i>Science</i> , 2018, 360, 327-331.	6.0	174
23	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. <i>Nature Genetics</i> , 2012, 44, 193-199.	9.4	157
24	Patient Mutations in Doublecortin Define a Repeated Tubulin-binding Domain. <i>Journal of Biological Chemistry</i> , 2000, 275, 34442-34450.	1.6	138
25	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. <i>Nature Genetics</i> , 2015, 47, 528-534.	9.4	111
26	The Neurobiology of Zika Virus. <i>Neuron</i> , 2016, 92, 949-958.	3.8	101
27	Defining the phenotypic spectrum of <i>SLC6A1</i> mutations. <i>Epilepsia</i> , 2018, 59, 389-402.	2.6	99
28	Mutations in LAMB1 Cause Cobblestone Brain Malformation without Muscular or Ocular Abnormalities. <i>American Journal of Human Genetics</i> , 2013, 92, 468-474.	2.6	96
29	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. <i>Neuron</i> , 2014, 84, 1226-1239.	3.8	95
30	AMPD2 Regulates GTP Synthesis and Is Mutated in a Potentially Treatable Neurodegenerative Brainstem Disorder. <i>Cell</i> , 2013, 154, 505-517.	13.5	94
31	A human three-dimensional neural-perivascular "assembloid"™ promotes astrocytic development and enables modeling of SARS-CoV-2 neuropathology. <i>Nature Medicine</i> , 2021, 27, 1600-1606.	15.2	94
32	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. <i>Genetics in Medicine</i> , 2018, 20, 1354-1364.	1.1	92
33	Cytosine-5 RNA methylation links protein synthesis to cell metabolism. <i>PLoS Biology</i> , 2019, 17, e3000297.	2.6	87
34	Loss of tubulin deglutamylase <i>CCP1</i> causes infantile-onset neurodegeneration. <i>EMBO Journal</i> , 2018, 37, .	3.5	86
35	NSUN2 introduces 5-methylcytosines in mammalian mitochondrial tRNAs. <i>Nucleic Acids Research</i> , 2019, 47, 8720-8733.	6.5	84
36	Off-Target Effect of doublecortin Family shRNA on Neuronal Migration Associated with Endogenous MicroRNA Dysregulation. <i>Neuron</i> , 2014, 82, 1255-1262.	3.8	79

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37	Pathogenetic mechanisms of focal cortical dysplasia. <i>Epilepsia</i> , 2014, 55, 970-978.	2.6	76
38	Autism risk in offspring can be assessed through quantification of male sperm mosaicism. <i>Nature Medicine</i> , 2020, 26, 143-150.	15.2	76
39	Mutations in CSPP1 Lead to Classical Joubert Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 80-86.	2.6	75
40	The landscape of somatic mutation in cerebral cortex of autistic and neurotypical individuals revealed by ultra-deep whole-genome sequencing. <i>Nature Neuroscience</i> , 2021, 24, 176-185.	7.1	73
41	Biallelic Mutations in Citron Kinase Link Mitotic Cytokinesis to Human Primary Microcephaly. <i>American Journal of Human Genetics</i> , 2016, 99, 501-510.	2.6	70
42	Biallelic loss of human CTNNA2, encoding β -catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. <i>Nature Genetics</i> , 2018, 50, 1093-1101.	9.4	70
43	Mutations in MBOAT7, Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features. <i>American Journal of Human Genetics</i> , 2016, 99, 912-916.	2.6	69
44	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. <i>Journal of Clinical Investigation</i> , 2019, 129, 1240-1256.	3.9	68
45	Biallelic mutations in the 3' exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. <i>Nature Genetics</i> , 2017, 49, 457-464.	9.4	66
46	A phenotypic spectrum of autism is attributable to the combined effects of rare variants, polygenic risk and sex. <i>Nature Genetics</i> , 2022, 54, 1284-1292.	9.4	66
47	Identification of a novel recessive RELN mutation using a homozygous balanced reciprocal translocation. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 939-944.	0.7	65
48	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. <i>ELife</i> , 2015, 4, e06602.	2.8	64
49	Biallelic Truncating Mutations in FMN2, Encoding the Actin-Regulatory Protein Formin 2, Cause Nonsyndromic Autosomal-Recessive Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 95, 721-728.	2.6	62
50	Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. <i>European Journal of Human Genetics</i> , 2015, 23, 1482-1487.	1.4	62
51	Homozygous mutation in NUP107 leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome. <i>Journal of Medical Genetics</i> , 2017, 54, 399-403.	1.5	62
52	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. <i>American Journal of Human Genetics</i> , 2018, 103, 431-439.	2.6	62
53	Redefining the Etiologic Landscape of Cerebellar Malformations. <i>American Journal of Human Genetics</i> , 2019, 105, 606-615.	2.6	61
54	Mutations in CEP120 cause Joubert syndrome as well as complex ciliopathy phenotypes. <i>Journal of Medical Genetics</i> , 2016, 53, 608-615.	1.5	55

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55	Zika Virus Protease Cleavage of Host Protein Septin-2 Mediates Mitotic Defects in Neural Progenitors. <i>Neuron</i> , 2019, 101, 1089-1098.e4.	3.8	55
56	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2017, 100, 676-688.	2.6	54
57	Biallelic variants in KIF14 cause intellectual disability with microcephaly. <i>European Journal of Human Genetics</i> , 2018, 26, 330-339.	1.4	52
58	Blacklisting variants common in private cohorts but not in public databases optimizes human exome analysis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 950-959.	3.3	52
59	Mutations in GPAA1 , Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. <i>American Journal of Human Genetics</i> , 2017, 101, 856-865.	2.6	49
60	Identification of a novel ARL13B variant in a Joubert syndrome-affected patient with retinal impairment and obesity. <i>European Journal of Human Genetics</i> , 2015, 23, 621-627.	1.4	48
61	DCLK1 phosphorylates the microtubule-associated protein MAP7D1 to promote axon elongation in cortical neurons. <i>Developmental Neurobiology</i> , 2017, 77, 493-510.	1.5	48
62	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyposis. <i>American Journal of Human Genetics</i> , 2019, 105, 689-705.	2.6	48
63	Closing in on Mechanisms of Open Neural Tube Defects. <i>Trends in Neurosciences</i> , 2020, 43, 519-532.	4.2	47
64	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. <i>American Journal of Human Genetics</i> , 2019, 105, 534-548.	2.6	46
65	Novel congenital disorder of <i>O</i> -linked glycosylation caused by GALNT2 loss of function. <i>Brain</i> , 2020, 143, 1114-1126.	3.7	46
66	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. <i>American Journal of Human Genetics</i> , 2017, 101, 552-563.	2.6	45
67	Autosomal-Recessive Mutations in the tRNA Splicing Endonuclease Subunit TSEN15 Cause Pontocerebellar Hypoplasia and Progressive Microcephaly. <i>American Journal of Human Genetics</i> , 2016, 99, 228-235.	2.6	44
68	Loss of NARS1 impairs progenitor proliferation in cortical brain organoids and leads to microcephaly. <i>Nature Communications</i> , 2020, 11, 4038.	5.8	44
69	Homozygous Mutations in TBC1D23 Lead to a Non-degenerative Form of Pontocerebellar Hypoplasia. <i>American Journal of Human Genetics</i> , 2017, 101, 441-450.	2.6	43
70	Somatic double-hit in MTOR and RPS6 in hemimegalencephaly with intractable epilepsy. <i>Human Molecular Genetics</i> , 2019, 28, 3755-3765.	1.4	42
71	Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). <i>Human Genetics</i> , 2018, 137, 753-768.	1.8	38
72	Mutations in UNC80, Encoding Part of the UNC79-UNC80-NALCN Channel Complex, Cause Autosomal-Recessive Severe Infantile Encephalopathy. <i>American Journal of Human Genetics</i> , 2016, 98, 210-215.	2.6	37

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73	A homozygous founder mutation in <i>TRAPPC6B</i> associates with a neurodevelopmental disorder characterised by microcephaly, epilepsy and autistic features. <i>Journal of Medical Genetics</i> , 2018, 55, 48-54.	1.5	37
74	<i>PYCR2</i> Mutations cause a lethal syndrome of microcephaly and failure to thrive. <i>Annals of Neurology</i> , 2016, 80, 59-70.	2.8	35
75	Diencephalic-mesencephalic junction dysplasia: a novel recessive brain malformation. <i>Brain</i> , 2012, 135, 2416-2427.	3.7	34
76	Loss of Oxidation Resistance 1, <i>OXR1</i> , Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. <i>American Journal of Human Genetics</i> , 2019, 105, 1237-1253.	2.6	34
77	Loss of the neural-specific BAF subunit <i>ACTL6B</i> relieves repression of early response genes and causes recessive autism. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 10055-10066.	3.3	34
78	Molybdenum cofactor and isolated sulphite oxidase deficiencies: Clinical and molecular spectrum among Egyptian patients. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 714-722.	0.7	33
79	Negative selection on human genes underlying inborn errors depends on disease outcome and both the mode and mechanism of inheritance. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	33
80	Non-manifesting <i>AHI1</i> truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. <i>Human Molecular Genetics</i> , 2015, 24, 2594-2603.	1.4	32
81	De Novo and Bi-allelic Pathogenic Variants in <i>NARS1</i> Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. <i>American Journal of Human Genetics</i> , 2020, 107, 311-324.	2.6	32
82	Mutation spectrum of Joubert syndrome and related disorders among Arabs. <i>Human Genome Variation</i> , 2014, 1, 14020.	0.4	31
83	Mutations in Spliceosomal Genes <i>PPIL1</i> and <i>PRP17</i> Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. <i>Neuron</i> , 2021, 109, 241-256.e9.	3.8	31
84	Biallelic Mutations in <i>TMTC3</i> , Encoding a Transmembrane and TPR-Containing Protein, Lead to Cobblestone Lissencephaly. <i>American Journal of Human Genetics</i> , 2016, 99, 1181-1189.	2.6	30
85	Homozygous Missense Variants in <i>NTNG2</i> , Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 105, 1048-1056.	2.6	30
86	Clinical and Genetic Aspects of the Segmental Overgrowth Spectrum Due to Somatic Mutations in <i>PIK3CA</i> . <i>Journal of Pediatrics</i> , 2015, 167, 957-962.	0.9	29
87	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. <i>Brain</i> , 2020, 143, 2929-2944.	3.7	29
88	Biallelic mutations in valyl-tRNA synthetase gene <i>VARS</i> are associated with a progressive neurodevelopmental epileptic encephalopathy. <i>Nature Communications</i> , 2019, 10, 707.	5.8	28
89	<i>MINPP1</i> prevents intracellular accumulation of the chelator inositol hexakisphosphate and is mutated in Pontocerebellar Hypoplasia. <i>Nature Communications</i> , 2020, 11, 6087.	5.8	28
90	Loss of function mutations in <i>GEMIN5</i> cause a neurodevelopmental disorder. <i>Nature Communications</i> , 2021, 12, 2558.	5.8	28

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91	Developmental and temporal characteristics of clonal sperm mosaicism. <i>Cell</i> , 2021, 184, 4772-4783.e15.	13.5	27
92	Extending the mutation spectrum for Gallowayâ€“Mowat syndrome to include homozygous missense mutations in the WDR73 gene. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 992-998.	0.7	26
93	Genome-wide screen identifies novel machineries required for both ciliogenesis and cell cycle arrest upon serum starvation. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2016, 1863, 1307-1318.	1.9	26
94	Comprehensive identification of somatic nucleotide variants in human brain tissue. <i>Genome Biology</i> , 2021, 22, 92.	3.8	26
95	Somatic mosaicism reveals clonal distributions of neocortical development. <i>Nature</i> , 2022, 604, 689-696.	13.7	26
96	Homozygous mutation of STXBP5L explains an autosomal recessive infantile-onset neurodegenerative disorder. <i>Human Molecular Genetics</i> , 2015, 24, 2000-2010.	1.4	25
97	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctive cerebellar, ocular, craniofacial and genital features (COFG). <i>Tj ETQq1 1 0.784314 rgBT / Overlock</i>	1.4	25
98	Pathogenic ARH3 mutations result in ADP-ribose chromatin scars during DNA strand break repair. <i>Nature Communications</i> , 2020, 11, 3391.	5.8	25
99	Mutations in LNPK, Encoding the Endoplasmic Reticulum Junction Stabilizer Lunapark, Cause a Recessive Neurodevelopmental Syndrome. <i>American Journal of Human Genetics</i> , 2018, 103, 296-304.	2.6	24
100	Bi-allelic Mutations in FAM149B1 Cause Abnormal Primary Cilium and a Range of Ciliopathy Phenotypes in Humans. <i>American Journal of Human Genetics</i> , 2019, 104, 731-737.	2.6	23
101	A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. <i>New England Journal of Medicine</i> , 2021, 385, 1292-1301.	13.9	23
102	Pyruvate dehydrogenase complex-E2 deficiency causes paroxysmal exercise-induced dyskinesia. <i>Neurology</i> , 2017, 89, 2297-2298.	1.5	22
103	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. <i>American Journal of Human Genetics</i> , 2020, 106, 484-495.	2.6	22
104	Biallelic loss of function variants in <i>SYT2</i> cause a treatable congenital onset presynaptic myasthenic syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2272-2283.	0.7	20
105	A Zika virus mutation enhances transmission potential and confers escape from protective dengue virus immunity. <i>Cell Reports</i> , 2022, 39, 110655.	2.9	20
106	Loss of <i>Protocadherin 12</i> leads to <i>Diencephalic Mesencephalic Junction Dysplasia Syndrome</i> . <i>Annals of Neurology</i> , 2018, 84, 638-647.	2.8	19
107	Identification of a homozygous nonsense mutation in KIAA0556 in a consanguineous family displaying Joubert syndrome. <i>Human Genetics</i> , 2016, 135, 919-921.	1.8	18
108	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 105, 844-853.	2.6	17

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109	Biallelic variants in HPDL, encoding 4-hydroxyphenylpyruvate dioxygenase-like protein, lead to an infantile neurodegenerative condition. <i>Genetics in Medicine</i> , 2021, 23, 524-533.	1.1	17
110	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	0.7	17
111	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	3.7	17
112	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. <i>American Journal of Human Genetics</i> , 2021, 108, 134-147.	2.6	15
113	Polo-like kinase 2 regulates angiogenic sprouting and blood vessel development. <i>Developmental Biology</i> , 2015, 404, 49-60.	0.9	14
114	When size matters: CHD8 in autism. <i>Nature Neuroscience</i> , 2016, 19, 1430-1432.	7.1	14
115	Regulation of human cerebral cortical development by EXOC7 and EXOC8, components of the exocyst complex, and roles in neural progenitor cell proliferation and survival. <i>Genetics in Medicine</i> , 2020, 22, 1040-1050.	1.1	13
116	Implication of folate deficiency in CYP2U1 loss of function. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	13
117	Sperm mosaicism: implications for genomic diversity and disease. <i>Trends in Genetics</i> , 2021, 37, 890-902.	2.9	13
118	Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene <i>GSX2</i> . <i>Brain</i> , 2019, 142, 2965-2978.	3.7	12
119	Uner Tan syndrome caused by a homozygous <i>TUBB2B</i> mutation affecting microtubule stability. <i>Human Molecular Genetics</i> , 2016, 26, ddw383.	1.4	11
120	Early life experience shapes neural genome. <i>Science</i> , 2018, 359, 1330-1331.	6.0	11
121	Expanding the phenotype of <i>PIGS</i> associated early onset epileptic developmental encephalopathy. <i>Epilepsia</i> , 2021, 62, e35-e41.	2.6	11
122	Biallelic variants in <i>KARS1</i> are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. <i>Genetics in Medicine</i> , 2021, 23, 1933-1943.	1.1	11
123	Loss of <i>C2orf69</i> defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 1301-1317.	2.6	11
124	Clinical Pertinence Metric Enables Hypothesis-Independent Genome-Phenome Analysis for Neurologic Diagnosis. <i>Journal of Child Neurology</i> , 2015, 30, 881-888.	0.7	10
125	Insight into developmental mechanisms of global and focal migration disorders of cortical development. <i>Current Opinion in Neurobiology</i> , 2021, 66, 77-84.	2.0	9
126	<i>ABHD16A</i> deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. <i>American Journal of Human Genetics</i> , 2021, 108, 2017-2023.	2.6	9

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127	Biallelic <i>FRA10AC1</i> variants cause a neurodevelopmental disorder with growth retardation. <i>Brain</i> , 2022, 145, 1551-1563.	3.7	9
128	Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. <i>Human Mutation</i> , 2022, 43, 403-419.	1.1	9
129	Oligonucleotide correction of an intronic <i>TIMMDC1</i> variant in cells of patients with severe neurodegenerative disorder. <i>Npj Genomic Medicine</i> , 2022, 7, 9.	1.7	8
130	El-Hattab-Alkuraya syndrome caused by biallelic <i>WDR45B</i> pathogenic variants: Further delineation of the phenotype and genotype. <i>Clinical Genetics</i> , 2022, 101, 530-540.	1.0	7
131	Dandy-Walker malformation, genitourinary abnormalities, and intellectual disability in two families. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2503-2507.	0.7	6
132	Recurrent homozygous damaging mutation in <i>TMX2</i> , encoding a protein disulfide isomerase, in four families with microlissencephaly. <i>Journal of Medical Genetics</i> , 2020, 57, 274-282.	1.5	6
133	<i>RSRC1</i> loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. <i>Brain</i> , 2020, 143, e31-e31.	3.7	6
134	Monoallelic and biallelic mutations in <i>RELN</i> underlie a graded series of neurodevelopmental disorders. <i>Brain</i> , 2022, 145, 3274-3287.	3.7	6
135	Novel mutation in the fukutin gene in an Egyptian family with Fukuyama congenital muscular dystrophy and microcephaly. <i>Gene</i> , 2014, 539, 279-282.	1.0	5
136	Unbiased mosaic variant assessment in sperm: a cohort study to test predictability of transmission. <i>ELife</i> , 0, 11, .	2.8	5
137	The ciliary proteins Meckelin and Jouberin are required for retinoic acid-dependent neural differentiation of mouse embryonic stem cells. <i>Differentiation</i> , 2014, 87, 134-146.	1.0	4
138	Clinical, biomarker and genetic spectrum of Niemann-Pick type C in Egypt: The detection of nine novel <i>NPC1</i> mutations. <i>Clinical Genetics</i> , 2019, 95, 537-539.	1.0	4
139	Molecular diagnosis in recessive pediatric neurogenetic disease can help reduce disease recurrence in families. <i>BMC Medical Genomics</i> , 2020, 13, 68.	0.7	4
140	The Neurobiology of Modern Viral Scourges: ZIKV and COVID-19. <i>Neuroscientist</i> , 2022, 28, 438-452.	2.6	4
141	Bi-allelic <i>TTC5</i> variants cause delayed developmental milestones and intellectual disability. <i>Journal of Medical Genetics</i> , 2021, 58, 237-246.	1.5	4
142	Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDC) syndrome. <i>Journal of Medical Genetics</i> , 2021, 58, 815-831.	1.5	3
143	Biallelic <i>BICD2</i> variant is a novel candidate for Cohen-like syndrome. <i>Journal of Human Genetics</i> , 2022, 67, 553-556.	1.1	3
144	A founder mutation in <i>PEX12</i> among Egyptian patients in peroxisomal biogenesis disorder. <i>Neurological Sciences</i> , 2020, 42, 2737-2745.	0.9	1

#	ARTICLE	IF	CITATIONS
145	Primary Cilia and Brain Wiring, Connecting the Dots. <i>Developmental Cell</i> , 2019, 51, 661-663.	3.1	0
146	Editorial overview: Neurodevelopment Diseases and Neurogenetics pivot towards mechanisms and therapies. <i>Current Opinion in Genetics and Development</i> , 2020, 65, iii-vii.	1.5	0