William B Dobyns

List of Publications by Year
in descending order

401 papers	41,006 citations	1799 103 h-index	3407 183 g-index
418 all docs	418 docs citations	418 times ranked	32598 citing authors

#	Article	IF	Citations
1	Autosomal dominant cerebellar ataxia (SCA6) associated with small polyglutamine expansions in the Î±1A-voltage-dependent calcium channel. Nature Genetics, 1997, 15, 62-69.	21.4	1,606
2	Isolation of a Millerâ€“Dicker lissencephaly gene containing G protein Î²-subunit-like repeats. Nature, 1993, 364, 717-721.	27.8	1,036
3	doublecortin, a Brain-Specific Gene Mutated in Human X-Linked Lissencephaly and Double Cortex Syndrome, Encodes a Putative Signaling Protein. Cell, 1998, 92, 63-72.	28.9	1,007
4	A developmental and genetic classification for malformations of cortical development: update 2012. Brain, 2012, 135, 1348-1369.	7.6	849
5	Mutations in filamin 1 Prevent Migration of Cerebral Cortical Neurons in Human Periventricular Heterotopia. Neuron, 1998, 21, 1315-1325.	8.1	811
6	Characterizing the Pattern of Anomalies in Congenital Zika Syndrome for Pediatric Clinicians. JAMA Pediatrics, 2017, 171, 288.	6.2	746
7	A developmental and genetic classification for malformations of cortical development. Neurology, 2005, 65, 1873-1887.	1.1	711
8	Mutation of ARX causes abnormal development of forebrain and testes in mice and X-linked lissencephaly with abnormal genitalia in humans. Nature Genetics, 2002, 32, 359-369.	21.4	647
9	Mutations in the O-Mannosyltransferase Gene POMT1 Give Rise to the Severe Neuronal Migration Disorder Walker-Warburg Syndrome. American Journal of Human Genetics, 2002, 71, 1033-1043.	6.2	636
10	Mutation of the PAX2 gene in a family with optic nerve colobomas, renal anomalies and vesicoureteral reflux. Nature Genetics, 1995, 9, 358-364.	21.4	623
11	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	21.4	621
12	Recurrent 16p11.2 microdeletions in autism. Human Molecular Genetics, 2007, 17, 628-638.	2.9	614
13	G Protein-Coupled Receptor-Dependent Development of Human Frontal Cortex. Science, 2004, 303, 2033-2036.	12.6	498
14	Mutations in the Na+/K+-ATPase Î±3 Gene ATP1A3 Are Associated with Rapid-Onset Dystonia Parkinsonism. Neuron, 2004, 43, 169-175.	8.1	466
15	Diagnostic criteria for Walkerâ€Warburg syndrome. American Journal of Medical Genetics Part A, 1989, 32, 195-210.	2.4	439
16	Lymphatic and Other Vascular Malformative/Overgrowth Disorders AreÂCaused by Somatic Mutations in PIK3CA. Journal of Pediatrics, 2015, 166, 1048-1054.e5.	1.8	429
17	PIK3CAâ€related overgrowth spectrum (PROS): Diagnostic and testing eligibility criteria, differential diagnosis, and evaluation. American Journal of Medical Genetics, Part A, 2015, 167, 287-295.	1.2	399
18	Malformations of cortical development: clinical features and genetic causes. Lancet Neurology, The, 2014, 13, 710-726.	10.2	382

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19	14-3-3Îµ is important for neuronal migration by binding to NUDEL: a molecular explanation for Millerâ€“Dieker syndrome. Nature Genetics, 2003, 34, 274-285.	21.4	374
20	Description of 13 Infants Born During October 2015â€“January 2016 With Congenital Zika Virus Infection Without Microcephaly at Birth â€” Brazil. Morbidity and Mortality Weekly Report, 2016, 65, 1343-1348.	15.1	368
21	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	6.2	352
22	Consensus Paper: Cerebellar Development. Cerebellum, 2016, 15, 789-828.	2.5	337
23	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	28.9	322
24	Periventricular heterotopia: phenotypic heterogeneity and correlation with Filamin A mutations. Brain, 2006, 129, 1892-1906.	7.6	315
25	Point Mutations and an Intragenic Deletion in LIS1, the Lissencephaly Causative Gene in Isolated Lissencephaly Sequence and Miller-Dieker Syndrome. Human Molecular Genetics, 1997, 6, 157-164.	2.9	297
26	Mutations ofARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. Human Mutation, 2004, 23, 147-159.	2.5	293
27	Lissencephaly. JAMA - Journal of the American Medical Association, 1993, 270, 2838.	7.4	287
28	PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. Brain, 2015, 138, 1613-1628.	7.6	286
29	Lissencephaly and the molecular basis of neuronal migration. Human Molecular Genetics, 2003, 12, 89R-96.	2.9	274
30	Infantile hydrocephalus: A review of epidemiology, classification and causes. European Journal of Medical Genetics, 2014, 57, 359-368.	1.3	273
31	Novel Submicroscopic Chromosomal Abnormalities Detected in Autism Spectrum Disorder. Biological Psychiatry, 2008, 63, 1111-1117.	1.3	268
32	Spinocerebellar ataxia type 6: Gazeâ€evoked and vertical nystagmus, Purkinje cell degeneration, and variable age of onset. Annals of Neurology, 1997, 42, 933-950.	5.3	267
33	A developmental and genetic classification for midbrain-hindbrain malformations. Brain, 2009, 132, 3199-3230.	7.6	262
34	Mutations in WDR62, encoding a centrosome-associated protein, cause microcephaly with simplified gyri and abnormal cortical architecture. Nature Genetics, 2010, 42, 1015-1020.	21.4	259
35	WDR62 is associated with the spindle pole and is mutated in human microcephaly. Nature Genetics, 2010, 42, 1010-1014.	21.4	255
36	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	28.9	253

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37	Mutations of CASK cause an X-linked brain malformation phenotype with microcephaly and hypoplasia of the brainstem and cerebellum. Nature Genetics, 2008, 40, 1065-1067.	21.4	252
38	The phenotypic spectrum of rapid-onset dystonia-parkinsonism (RDP) and mutations in the ATP1A3 gene. Brain, 2007, 130, 828-835.	7.6	251
39	Practice Parameter: Evaluation of the child with microcephaly (an evidence-based review) [RETIRED]. Neurology, 2009, 73, 887-897.	1.1	244
40	Fetal brain lesions after subcutaneous inoculation of Zika virus in a pregnant nonhuman primate. Nature Medicine, 2016, 22, 1256-1259.	30.7	241
41	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. Nature Genetics, 2012, 44, 440-444.	21.4	237
42	FOXC1 is required for normal cerebellar development and is a major contributor to chromosome 6p25.3 Dandy-Walker malformation. Nature Genetics, 2009, 41, 1037-1042.	21.4	234
43	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	9.0	234
44	The NPHP1 Gene Deletion Associated with Juvenile Nephronophthisis Is Present in a Subset of Individuals with Joubert Syndrome. American Journal of Human Genetics, 2004, 75, 82-91.	6.2	228
45	Abnormal development of the human cerebral cortex: genetics, functional consequences and treatment options. Trends in Neurosciences, 2008, 31, 154-162.	8.6	227
46	The core FOXG1 syndrome phenotype consists of postnatal microcephaly, severe mental retardation, absent language, dyskinesia, and corpus callosum hypogenesis. Journal of Medical Genetics, 2011, 48, 396-406.	3.2	220
47	Syndromes with lissencephaly. I: Millerdieker and Norman-Roberts syndromes and isolated lissencephaly. American Journal of Medical Genetics Part A, 1984, 18, 509-526.	2.4	218
48	Neonatal adrenoleukodystrophy: New cases, biochemical studies, and differentiation from Zellweger and related peroxisomal polydystrophy syndromes. American Journal of Medical Genetics Part A, 1986, 23, 869-901.	2.4	216
49	Refinement of a 400-kb Critical Region Allows Genotypic Differentiation between Isolated Lissencephaly, Miller-Dieker Syndrome, and Other Phenotypes Secondary to Deletions of 17p13.3. American Journal of Human Genetics, 2003, 72, 918-930.	6.2	215
50	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. Brain, 2010, 133, 1415-1427.	7.6	215
51	Molar tooth sign of the midbrain-hindbrain junction: Occurrence in multiple distinct syndromes. American Journal of Medical Genetics Part A, 2004, 125A, 125-134.	2.4	213
52	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. Nature Genetics, 2012, 44, 575-580.	21.4	212
53	Heterozygous deletion of the linked genes ZIC1 and ZIC4 is involved in Dandy-Walker malformation. Nature Genetics, 2004, 36, 1053-1055.	21.4	206
54	Human malformations of the midbrain and hindbrain: review and proposed classification scheme. Molecular Genetics and Metabolism, 2003, 80, 36-53.	1.1	205

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55	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. Nature Genetics, 2005, 37, 221-224.	21.4	201
56	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. Brain, 2011, 134, 143-156.	7.6	200
57	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	3.2	190
58	Megalencephalyâ€capillary malformation (MCAP) and megalencephalyâ€polydactylyâ€polymicrogyriaâ€hydrocephalus (MPPH) syndromes: Two closely related disorders of brain overgrowth and abnormal brain and body morphogenesis. American Journal of Medical Genetics, Part A, 2012, 158A, 269-291.	1.2	188
59	Targeted loss of Arx results in a developmental epilepsy mouse model and recapitulates the human phenotype in heterozygous females. Brain, 2009, 132, 1563-1576.	7.6	178
60	A Revision of the Lissencephaly and Miller-Dieker Syndrome Critical Regions in Chromosome 17p13.3. Human Molecular Genetics, 1997, 6, 147-155.	2.9	176
61	TUBA1A mutations cause wide spectrum lissencephaly (smooth brain) and suggest that multiple neuronal migration pathways converge on alpha tubulins. Human Molecular Genetics, 2010, 19, 2817-2827.	2.9	176
62	Malformations of cortical development and epilepsy. Dialogues in Clinical Neuroscience, 2008, 10, 47-62.	3.7	176
63	Characterization of mutations in the genedoublecortin in patients with double cortex syndrome. Annals of Neurology, 1999, 45, 146-153.	5.3	175
64	Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 92, 354-365.	6.2	172
65	Genetic links between brain development and brain evolution. Nature Reviews Genetics, 2005, 6, 581-590.	16.3	169
66	Miller-Dieker syndrome: Lissencephaly andmonosomy 17p. Journal of Pediatrics, 1983, 102, 552-558.	1.8	166
67	Loss-of-Function Mutations in RAB18 Cause Warburg Micro Syndrome. American Journal of Human Genetics, 2011, 88, 499-507.	6.2	158
68	Genetic Basis of Brain Malformations. Molecular Syndromology, 2016, 7, 220-233.	0.8	156
69	Syndromes with lissencephaly. II: Walker-Warburg and Cerebro-Oculo-Muscular syndromes and a new syndrome with type II lissencephaly. American Journal of Medical Genetics Part A, 1985, 22, 157-195.	2.4	151
70	Mutation analysis of the DCX gene and genotype/phenotype correlation in subcortical band heterotopia. European Journal of Human Genetics, 2001, 9, 5-12.	2.8	144
71	Mapping of Deletion and Translocation Breakpoints in 1q44 Implicates the Serine/Threonine Kinase AKT3 in Postnatal Microcephaly and Agenesis of the Corpus Callosum. American Journal of Human Genetics, 2007, 81, 292-303.	6.2	144
72	Genetic and Biologic Classification of Infantile Spasms. Pediatric Neurology, 2011, 45, 355-367.	2.1	144

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73	X-Linked Lissencephaly With Abnormal Genitalia as a Tangential Migration Disorder Causing Intractable Epilepsy: Proposal for a New Term, â€œInterneuronopathyâ€. Journal of Child Neurology, 2005, 20, 392-397.	1.4	143
74	Expansion of the first PolyA tract of ARX causes infantile spasms and status dystonicus. Neurology, 2007, 69, 427-433.	1.1	143
75	Subcortical Band Heterotopia in Rare Affected Males Can be Caused by Missense Mutations in DCX (XLIS) or LIS1. Human Molecular Genetics, 1999, 8, 1757-1760.	2.9	142
76	Cerebello-oculo-renal syndromes including Arima, Senior-Lï¿½ken and COACH syndromes: More than just variants of Joubert syndrome. American Journal of Medical Genetics Part A, 1999, 86, 459-469.	2.4	142
77	Macrocephaly-Cutis marmorata telangiectatica congenita: A distinct disorder with developmental delay and connective tissue abnormalities. American Journal of Medical Genetics Part A, 1997, 70, 67-73.	2.4	141
78	Inheritance of most X-linked traits is not dominant or recessive, just X-linked. American Journal of Medical Genetics Part A, 2004, 129A, 136-143.	2.4	140
79	Megalencephaly Syndromes and Activating Mutations in the PI3Kâ€AKT Pathway: MPPH and MCAP. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 122-130.	1.6	139
80	Somatic and Germline Mosaic Mutations in the doublecortin Gene Are Associated with Variable Phenotypes. American Journal of Human Genetics, 2000, 67, 574-581.	6.2	135
81	PIK3CA-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. JCI Insight, 2016, 1, .	5.0	134
82	Overlapping cortical malformations and mutations in TUBB2B and TUBA1A. Brain, 2013, 136, 536-548.	7.6	133
83	Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. Brain, 2012, 135, 1370-1386.	7.6	131
84	Mutations in 3 genes (MKS3, CC2D2A and RPGRIP1L) cause COACH syndrome (Joubert syndrome with) Tj ETQq0 0 0 rgBT /Overlock 10	3.2	129
85	Lissencephaly with Cerebellar Hypoplasia (LCH): A Heterogeneous Group of Cortical Malformations. Neuropediatrics, 2001, 32, 256-263.	0.6	128
86	Interstitial deletion of (17)(p11.2p11.2): Report of six additional patients with a new chromosome deletion syndrome. American Journal of Medical Genetics Part A, 1986, 24, 421-432.	2.4	127
87	X-linked lissencephaly with absent corpus callosum and ambiguous genitalia. American Journal of Medical Genetics Part A, 1999, 86, 331-337.	2.4	126
88	Polymicrogyria and deletion 22q11.2 syndrome: Window to the etiology of a common cortical malformation. American Journal of Medical Genetics, Part A, 2006, 140A, 2416-2425.	1.2	125
89	AHI1gene mutations cause specific forms of Joubert syndrome-related disorders. Annals of Neurology, 2006, 59, 527-534.	5.3	125
90	Early-Life Epilepsies and the Emerging Role of Genetic Testing. JAMA Pediatrics, 2017, 171, 863.	6.2	125

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91	Genotype-phenotype analysis of human frontoparietal polymicrogyria syndromes. Annals of Neurology, 2005, 58, 680-687.	5.3	124
92	Recessive Mutations in the Gene Encoding the Tight Junction Protein Occludin Cause Band-like Calcification with Simplified Gyration and Polymicrogyria. American Journal of Human Genetics, 2010, 87, 354-364.	6.2	123
93	COL4A1 Mutations Cause Ocular Dysgenesis, Neuronal Localization Defects, and Myopathy in Mice and Walker-Warburg Syndrome in Humans. PLoS Genetics, 2011, 7, e1002062.	3.5	121
94	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	8.1	121
95	PRKDC mutations in a SCID patient with profound neurological abnormalities. Journal of Clinical Investigation, 2013, 123, 2969-2980.	8.2	121
96	Bilateral frontoparietal polymicrogyria: Clinical and radiological features in 10 families with linkage to chromosome 16. Annals of Neurology, 2003, 53, 596-606.	5.3	120
97	Familial cavernous malformations of the central nervous system and retina. Annals of Neurology, 1987, 21, 578-583.	5.3	119
98	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. Nature Genetics, 2014, 46, 510-515.	21.4	118
99	A novel rasopathy caused by recurrent de novo missense mutations in PPP1CB closely resembles Noonan syndrome with loose anagen hair. American Journal of Medical Genetics, Part A, 2016, 170, 2237-2247.	1.2	117
100	Congenital Zika virus infection as a silent pathology with loss of neurogenic output in the fetal brain. Nature Medicine, 2018, 24, 368-374.	30.7	117
101	Clinical and molecular basis of classical lissencephaly: Mutations in theLIS1 gene (PAFAH1B1). Human Mutation, 2002, 19, 4-15.	2.5	116
102	AHI1 mutations cause both retinal dystrophy and renal cystic disease in Joubert syndrome. Journal of Medical Genetics, 2005, 43, 334-339.	3.2	116
103	Baraitserâ€“Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. European Journal of Human Genetics, 2015, 23, 292-301.	2.8	115
104	Phenotypic spectrum associated with CASK loss-of-function mutations. Journal of Medical Genetics, 2011, 48, 741-751.	3.2	114
105	New chromosomal syndrome: Miller-Dieker syndrome and monosomy 17p13. Human Genetics, 1984, 67, 193-200.	3.8	111
106	Linkage of a human brain malformation, familial holoprosencephaly, to chromosome 7 and evidence for genetic heterogeneity.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 8102-8106.	7.1	111
107	A Locus for Bilateral Perisylvian Polymicrogyria Maps to Xq28. American Journal of Human Genetics, 2002, 70, 1003-1008.	6.2	111
108	Novel mutations in ATP1A3 associated with catastrophic early life epilepsy, episodic prolonged apnea, and postnatal microcephaly. Epilepsia, 2015, 56, 422-430.	5.1	107

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109	Subcortical band heterotopia (SBH) in males: clinical, imaging and genetic findings in comparison with females. Brain, 2002, 125, 2507-2522.	7.6	105
110	Mutation of FOXC1 and PITX2 induces cerebral small-vessel disease. Journal of Clinical Investigation, 2014, 124, 4877-4881.	8.2	105
111	Mechanisms of Interhemispheric Transfer and Patterns of Cognitive Function in Acallosal Patients of Normal Intelligence. Archives of Neurology, 1992, 49, 271-277.	4.5	104
112	Genotypically Defined Lissencephalies Show Distinct Pathologies. Journal of Neuropathology and Experimental Neurology, 2005, 64, 847-857.	1.7	104
113	Malformations of Cortical Development and Epilepsy. Cold Spring Harbor Perspectives in Medicine, 2015, 5, a022392-a022392.	6.2	104
114	Lissencephaly: Expanded imaging and clinical classification. American Journal of Medical Genetics, Part A, 2017, 173, 1473-1488.	1.2	104
115	Neuroimaging findings in macrocephalyâ€“capillary malformation: A longitudinal study of 17 patients. American Journal of Medical Genetics, Part A, 2007, 143A, 2981-3008.	1.2	103
116	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
117	Ultra-High-Field MR Imaging in Polymicrogyria and Epilepsy. American Journal of Neuroradiology, 2015, 36, 309-316.	2.4	100
118	Previously apparently undescribed syndrome: Shallow orbits, ptosis, coloboma, trigonocephaly, gyral malformations, and mental and growth retardation. American Journal of Medical Genetics Part A, 1995, 57, 403-409.	2.4	99
119	De Novo Mutations in the Beta-Tubulin Gene TUBB2A Cause Simplified Gyral Patterning and Infantile-Onset Epilepsy. American Journal of Human Genetics, 2014, 94, 634-641.	6.2	99
120	Albright hereditary osteodystrophy and del(2)(q37.3) in four unrelated individuals. American Journal of Medical Genetics Part A, 1995, 58, 1-7.	2.4	98
121	Spatial and cell type transcriptional landscape of human cerebellar development. Nature Neuroscience, 2021, 24, 1163-1175.	14.8	98
122	Rapid-onset dystonia-parkinsonism: Linkage to chromosome 19q13. Annals of Neurology, 1999, 46, 176-182.	5.3	97
123	Spatiotemporal expansion of primary progenitor zones in the developing human cerebellum. Science, 2019, 366, 454-460.	12.6	97
124	Mutations in LAMB1 Cause Cobblestone Brain Malformation without Muscular or Ocular Abnormalities. American Journal of Human Genetics, 2013, 92, 468-474.	6.2	96
125	Microlissencephaly: A Heterogeneous Malformation of Cortical Development. Neuropediatrics, 1998, 29, 113-119.	0.6	95
126	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239.	8.1	95

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127	Clinical Nosologic and Genetic Aspects of Joubert and Related Syndromes. Journal of Child Neurology, 1999, 14, 660-666.	1.4	94
128	Genetic and neuroradiological heterogeneity of double cortex syndrome. Annals of Neurology, 2000, 47, 265-269.	5.3	94
129	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephalyâ€“capillary malformation syndrome. Nature Genetics, 2013, 45, 556-562.	21.4	94
130	Consistent chromosome abnormalities identify novel polymicrogyria loci in 1p36.3, 2p16.1â€“p23.1, 4q21.21â€“q22.1, 6q26â€“q27, and 21q2. American Journal of Medical Genetics, Part A, 2008, 146A, 1637-1654.	1.2	93
131	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. Genetics in Medicine, 2018, 20, 1354-1364.	2.4	92
132	The molecular landscape of ASPM mutations in primary microcephaly. Journal of Medical Genetics, 2009, 46, 249-253.	3.2	91
133	CHMP1A encodes an essential regulator of BMI1-INK4A in cerebellar development. Nature Genetics, 2012, 44, 1260-1264.	21.4	91
134	High incidence of progressive postnatal cerebellar enlargement in Costello syndrome: Brain overgrowth associated with HRAS mutations as the likely cause of structural brain and spinal cord abnormalities. American Journal of Medical Genetics, Part A, 2010, 152A, 1161-1168.	1.2	89
135	MEF2C Haploinsufficiency features consistent hyperkinesis, variable epilepsy, and has a role in dorsal and ventral neuronal developmental pathways. Neurogenetics, 2013, 14, 99-111.	1.4	89
136	Clinical spectrum of SIX3-associated mutations in holoprosencephaly: correlation between genotype, phenotype and function. Journal of Medical Genetics, 2009, 46, 389-398.	3.2	88
137	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	6.2	88
138	Semiquantitative analysis of hypothalamic damage on <scp>MRI</scp> predicts risk for hypothalamic obesity. Obesity, 2015, 23, 1226-1233.	3.0	87
139	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. Brain, 2013, 136, 3378-3394.	7.6	85
140	Flores hominid: New species or microcephalic dwarf?. The Anatomical Record Part A: Discoveries in Molecular, Cellular, and Evolutionary Biology, 2006, 288A, 1123-1145.	2.0	83
141	Mutations in CENPE define a novel kinetochore-centromeric mechanism for microcephalic primordial dwarfism. Human Genetics, 2014, 133, 1023-1039.	3.8	82
142	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. Brain, 2017, 140, 2322-2336.	7.6	82
143	Linkage and physical mapping of X-linked lissencephaly/SBH (XLIS): a gene causing neuronal migration defects in human brain. Human Molecular Genetics, 1997, 6, 555-562.	2.9	81
144	Association and Mutation Analyses of 16p11.2 Autism Candidate Genes. PLoS ONE, 2009, 4, e4582.	2.5	80

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145	Mouse models of human PIK3CA-related brain overgrowth have acutely treatable epilepsy. ELife, 2015, 4, .	6.0	79
146	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. Human Molecular Genetics, 2015, 24, 5313-5325.	2.9	77
147	A developmental classification of malformations of the brainstem. Annals of Neurology, 2007, 62, 625-639.	5.3	75
148	Identification of a Duplication of Xq28 Associated with Bilateral Periventricular Nodular Heterotopia. American Journal of Human Genetics, 1997, 61, 379-387.	6.2	74
149	Chiari I malformation, delayed gross motor skills, severe speech delay, and epileptiform discharges in a child with FOXP1 haploinsufficiency. European Journal of Human Genetics, 2010, 18, 1216-1220.	2.8	74
150	Copy number variants and infantile spasms: evidence for abnormalities in ventral forebrain development and pathways of synaptic function. European Journal of Human Genetics, 2011, 19, 1238-1245.	2.8	74
151	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. Lancet Neurology, The, 2015, 14, 1182-1195.	10.2	74
152	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. American Journal of Human Genetics, 2017, 101, 23-36.	6.2	74
153	Periventricular nodular heterotopia with overlying polymicrogyria. Brain, 2005, 128, 2811-2821.	7.6	73
154	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. Brain, 2018, 141, 698-712.	7.6	72
155	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
156	Bilateral periventricular nodular heterotopia with mental retardation and frontonasal malformation. Neurology, 1998, 51, 499-503.	1.1	70
157	A Recurrent Mosaic Mutation in SMO , Encoding the Hedgehog Signal Transducer Smoothened, Is the Major Cause of Curry-Jones Syndrome. American Journal of Human Genetics, 2016, 98, 1256-1265.	6.2	70
158	Biallelic loss of human CTNNA2, encoding Î±N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	21.4	70
159	Costello syndrome: Clinical phenotype, genotype, and management guidelines. American Journal of Medical Genetics, Part A, 2019, 179, 1725-1744.	1.2	70
160	Both Rare and De Novo Copy Number Variants Are Prevalent in Agenesis of the Corpus Callosum but Not in Cerebellar Hypoplasia or Polymicrogyria. PLoS Genetics, 2013, 9, e1003823.	3.5	69
161	Mutations inEXOSC2are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. Journal of Medical Genetics, 2016, 53, 419-425.	3.2	69
162	Megalencephaly syndromes associated with mutations of core components of the PI3Kâ€AKTâ€“MTOR pathway: PIK3CA, PIK3R2, AKT3, and MTOR. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 582-590.	1.6	69

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