Christopher A Walsh

List of Publications by Citations

259 papers	34,473 citations	100 h-index	184 g-index
313 ext. papers	40,322 ext. citations	15.4 avg, IF	7.01 L-index

#	Paper	IF	Citations
259	Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature*, **2014**, 515, 209-15	50.4	1581
258	Association between microdeletion and microduplication at 16p11.2 and autism. *New England Journal of Medicine*, **2008**, 358, 667-75	59.2	1249
257	Regulation of cerebral cortical size by control of cell cycle exit in neural precursors. *Science*, **2002**, 297, 365-9	33.3	1157
256	Doublecortin is a microtubule-associated protein and is expressed widely by migrating neurons. *Neuron*, **1999**, 23, 257-71	13.9	1061
255	Multiple recurrent de novo CNVs, including duplications of the 7q11.23 Williams syndrome region, are strongly associated with autism. *Neuron*, **2011**, 70, 863-85	13.9	932
254	Directed migration of neural stem cells to sites of CNS injury by the stromal cell-derived factor 1alpha/CXC chemokine receptor 4 pathway. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 18117-22	11.5	926
253	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	56.2	904
252	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. *Neuron*, **2015**, 87, 1215-1233	13.9	806
251	Mutations in filamin 1 prevent migration of cerebral cortical neurons in human periventricular heterotopia. *Neuron*, **1998**, 21, 1315-25	13.9	729
250	Autosomal recessive lissencephaly with cerebellar hypoplasia is associated with human RELN mutations. *Nature Genetics*, **2000**, 26, 93-6	36.3	700
249	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. *Cell*, **2020**, 180, 568-584.e23	56.2	578
248	Identifying autism loci and genes by tracing recent shared ancestry. *Science*, **2008**, 321, 218-23	33.3	578
247	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, 1033-43	11	573
246	Reelin binds alpha3beta1 integrin and inhibits neuronal migration. *Neuron*, **2000**, 27, 33-44	13.9	489
245	ASPM is a major determinant of cerebral cortical size. *Nature Genetics*, **2002**, 32, 316-20	36.3	472
244	A centrosomal mechanism involving CDK5RAP2 and CENPJ controls brain size. *Nature Genetics*, **2005**, 37, 353-5	36.3	447
243	The cerebrospinal fluid provides a proliferative niche for neural progenitor cells. *Neuron*, **2011**, 69, 893-905	13.9	430

242	Single-neuron sequencing analysis of L1 retrotransposition and somatic mutation in the human brain. *Cell*, **2012**, 151, 483-96	56.2	404
241	G protein-coupled receptor-dependent development of human frontal cortex. *Science*, **2004**, 303, 2033-6	33.3	404
240	The many faces of filamin: a versatile molecular scaffold for cell motility and signalling. *Nature Cell Biology*, **2004**, 6, 1034-8	23.4	398
239	Somatic mutation, genomic variation, and neurological disease. *Science*, **2013**, 341, 1237758	33.3	390
238	PAK3 mutation in nonsyndromic X-linked mental retardation. *Nature Genetics*, **1998**, 20, 25-30	36.3	387
237	Expression of Cux-1 and Cux-2 in the subventricular zone and upper layers II-IV of the cerebral cortex. *Journal of Comparative Neurology*, **2004**, 479, 168-80	3.4	383
236	Somatic mutation in single human neurons tracks developmental and transcriptional history. *Science*, **2015**, 350, 94-98	33.3	364
235	Characterization of Foxp2 and Foxp1 mRNA and protein in the developing and mature brain. *Journal of Comparative Neurology*, **2003**, 460, 266-79	3.4	362
234	Somatic activation of AKT3 causes hemispheric developmental brain malformations. *Neuron*, **2012**, 74, 41-8	13.9	341
233	Abnormal cerebellar development and axonal decussation due to mutations in AHI1 in Joubert syndrome. *Nature Genetics*, **2004**, 36, 1008-13	36.3	324
232	Aging and neurodegeneration are associated with increased mutations in single human neurons. *Science*, **2018**, 359, 555-559	33.3	315
231	Early asymmetry of gene transcription in embryonic human left and right cerebral cortex. *Science*, **2005**, 308, 1794-8	33.3	303
230	Using whole-exome sequencing to identify inherited causes of autism. *Neuron*, **2013**, 77, 259-73	13.9	297
229	Mitotic spindle regulation by Nde1 controls cerebral cortical size. *Neuron*, **2004**, 44, 279-93	13.9	293
228	Mutations in ARFGEF2 implicate vesicle trafficking in neural progenitor proliferation and migration in the human cerebral cortex. *Nature Genetics*, **2004**, 36, 69-76	36.3	292
227	Neuronal migration disorders: from genetic diseases to developmental mechanisms. *Trends in Neurosciences*, **2000**, 23, 352-9	13.3	291
226	Clinical genetic testing for patients with autism spectrum disorders. *Pediatrics*, **2010**, 125, e727-35	7.4	281
225	Doublecortin is required in mice for lamination of the hippocampus but not the neocortex. *Journal of Neuroscience*, **2002**, 22, 7548-57	6.6	269

224	Somatic mutations in cerebral cortical malformations. *New England Journal of Medicine*, **2014**, 371, 733-43	59.2	265
223	LIS1 regulates CNS lamination by interacting with mNudE, a central component of the centrosome. *Neuron*, **2000**, 28, 665-79	13.9	252
222	Molecular approaches to brain asymmetry and handedness. *Nature Reviews Neuroscience*, **2006**, 7, 655-62	23.5	250
221	Aberrant splicing of a mouse disabled homolog, mdab1, in the scrambler mouse. *Neuron*, **1997**, 19, 239-49	13.9	237
220	Patterning of the dorsal telencephalon and cerebral cortex by a roof plate-Lhx2 pathway. *Neuron*, **2001**, 32, 591-604	13.9	237
219	Mutations in WDR62, encoding a centrosome-associated protein, cause microcephaly with simplified gyri and abnormal cortical architecture. *Nature Genetics*, **2010**, 42, 1015-20	36.3	236
218	Filamin A (FLNA) is required for cell-cell contact in vascular development and cardiac morphogenesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2006**, 103, 19836-41	11.5	222
217	Genetic interactions between doublecortin and doublecortin-like kinase in neuronal migration and axon outgrowth. *Neuron*, **2006**, 49, 41-53	13.9	218
216	Mutations in PNKP cause microcephaly, seizures and defects in DNA repair. *Nature Genetics*, **2010**, 42, 245-9	36.3	210
215	Increased neuronal production, enlarged forebrains and cytoarchitectural distortions in beta-catenin overexpressing transgenic mice. *Cerebral Cortex*, **2003**, 13, 599-606	5.1	201
214	Human brain malformations and their lessons for neuronal migration. *Annual Review of Neuroscience*, **2001**, 24, 1041-70	17	200
213	Single-cell, genome-wide sequencing identifies clonal somatic copy-number variation in the human brain. *Cell Reports*, **2014**, 8, 1280-9	10.6	198
212	Cdk5rap2 regulates centrosome function and chromosome segregation in neuronal progenitors. *Development (Cambridge)*, **2010**, 137, 1907-17	6.6	197
211	Single-cell analysis reveals transcriptional heterogeneity of neural progenitors in human cortex. *Nature Neuroscience*, **2015**, 18, 637-46	25.5	195
210	Cux1 and Cux2 regulate dendritic branching, spine morphology, and synapses of the upper layer neurons of the cortex. *Neuron*, **2010**, 66, 523-35	13.9	190
209	Mammalian target of rapamycin pathway mutations cause hemimegalencephaly and focal cortical dysplasia. *Annals of Neurology*, **2015**, 77, 720-5	9.4	183
208	Cell lineage analysis in human brain using endogenous retroelements. *Neuron*, **2015**, 85, 49-59	13.9	183
207	GPR56 regulates pial basement membrane integrity and cortical lamination. *Journal of Neuroscience*, **2008**, 28, 5817-26	6.6	175

206	Protein-protein interactions, cytoskeletal regulation and neuronal migration. *Nature Reviews Neuroscience*, **2001**, 2, 408-16	13.5	169
205	Molecular insights into human brain evolution. *Nature*, **2005**, 437, 64-7	50.4	167
204	Human mutations in NDE1 cause extreme microcephaly with lissencephaly [corrected]. *American Journal of Human Genetics*, **2011**, 88, 536-47	11	165
203	A mapping label required for normal scale of body representation in the cortex. *Nature Neuroscience*, **2000**, 3, 358-65	25.5	163
202	Evolutionarily dynamic alternative splicing of GPR56 regulates regional cerebral cortical patterning. *Science*, **2014**, 343, 764-8	33.3	161
201	DCAMKL1 encodes a protein kinase with homology to doublecortin that regulates microtubule polymerization. *Journal of Neuroscience*, **2000**, 20, 9152-61	6.6	160
200	Cell-Type-Specific Alternative Splicing Governs Cell Fate in the Developing Cerebral Cortex. *Cell*, **2016**, 166, 1147-1162.e15	56.2	159
199	Genetic causes of microcephaly and lessons for neuronal development. *Wiley Interdisciplinary Reviews: Developmental Biology*, **2013**, 2, 461-78	5.9	157
198	Characterization of mutations in the gene doublecortin in patients with double cortex syndrome. *Annals of Neurology*, **1999**, 45, 146-53	9.4	157
197	Deletions of NRXN1 (neurexin-1) predispose to a wide spectrum of developmental disorders. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 937-47	3.5	156
196	Building a lineage from single cells: genetic techniques for cell lineage tracking. *Nature Reviews Genetics*, **2017**, 18, 230-244	30.1	155
195	Mutations in Human Accelerated Regions Disrupt Cognition and Social Behavior. *Cell*, **2016**, 167, 341-354.e12	30.12	154
194	Intersection of diverse neuronal genomes and neuropsychiatric disease: The Brain Somatic Mosaicism Network. *Science*, **2017**, 356,	33.3	152
193	Accelerated evolution of the ASPM gene controlling brain size begins prior to human brain expansion. *PLoS Biology*, **2004**, 2, E126	9.7	146
192	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. *Nature Neuroscience*, **2017**, 20, 1217-1224	25.5	144
191	Exome sequencing and functional validation in zebrafish identify GTDC2 mutations as a cause of Walker-Warburg syndrome. *American Journal of Human Genetics*, **2012**, 91, 541-7	11	144
190	Somatic Mutations Activating the mTOR Pathway in Dorsal Telencephalic Progenitors Cause a Continuum of Cortical Dysplasias. *Cell Reports*, **2017**, 21, 3754-3766	10.6	143
189	Disruption of neural progenitors along the ventricular and subventricular zones in periventricular heterotopia. *Human Molecular Genetics*, **2009**, 18, 497-516	5.6	143

188	Protein-truncating mutations in ASPM cause variable reduction in brain size. *American Journal of Human Genetics*, **2003**, 73, 1170-7	11	143
187	The microcephaly ASPM gene is expressed in proliferating tissues and encodes for a mitotic spindle protein. *Human Molecular Genetics*, **2005**, 14, 2155-65	5.6	143
186	Neurogenesis at the brain-cerebrospinal fluid interface. *Annual Review of Cell and Developmental Biology*, **2011**, 27, 653-79	12.6	140
185	Autism and brain development. *Cell*, **2008**, 135, 396-400	56.2	140
184	Mutations in B3GALNT2 cause congenital muscular dystrophy and hypoglycosylation of -dystroglycan. *American Journal of Human Genetics*, **2013**, 92, 354-65	11	139
183	The hyh mutation uncovers roles for alpha Snap in apical protein localization and control of neural cell fate. *Nature Genetics*, **2004**, 36, 264-70	36.3	139
182	Whole-exome sequencing and homozygosity analysis implicate depolarization-regulated neuronal genes in autism. *PLoS Genetics*, **2012**, 8, e1002635	6	134
181	Molecular genetics of human microcephaly. *Current Opinion in Neurology*, **2001**, 14, 151-6	7.1	134
180	Genetic malformations of the human cerebral cortex. *Neuron*, **1999**, 23, 19-29	13.9	134
179	What disorders of cortical development tell us about the cortex: one plus one does not always make two. *Current Opinion in Genetics and Development*, **2011**, 21, 333-9	4.9	130
178	The Genetics of Primary Microcephaly. *Annual Review of Genomics and Human Genetics*, **2018**, 19, 177-200	9.7	130
177	Microcephaly gene links trithorax and REST/NRSF to control neural stem cell proliferation and differentiation. *Cell*, **2012**, 151, 1097-112	56.2	122
176	Somatic and germline mosaic mutations in the doublecortin gene are associated with variable phenotypes. *American Journal of Human Genetics*, **2000**, 67, 574-81	11	122
175	Somatic mosaicism and neurodevelopmental disease. *Nature Neuroscience*, **2018**, 21, 1504-1514	25.5	122
174	Both doublecortin and doublecortin-like kinase play a role in cortical interneuron migration. *Journal of Neuroscience*, **2007**, 27, 3875-83	6.6	121
173	The exon junction complex component Magoh controls brain size by regulating neural stem cell division. *Nature Neuroscience*, **2010**, 13, 551-8	25.5	120
172	Patient mutations in doublecortin define a repeated tubulin-binding domain. *Journal of Biological Chemistry*, **2000**, 275, 34442-50	5.4	120
171	The diverse genetic landscape of neurodevelopmental disorders. *Annual Review of Genomics and Human Genetics*, **2014**, 15, 195-213	9.7	116

170	A genome-wide association study of autism using the Simons Simplex Collection: Does reducing phenotypic heterogeneity in autism increase genetic homogeneity?. *Biological Psychiatry*, **2015**, 77, 775-84	7.9	113
169	Smooth, rough and upside-down neocortical development. *Current Opinion in Genetics and Development*, **2002**, 12, 320-7	4.9	113
168	Missense mutation in PAK3, R67C, causes X-linked nonspecific mental retardation. *American Journal of Medical Genetics Part A*, **2000**, 93, 294-8		112
167	Bilateral frontoparietal polymicrogyria: clinical and radiological features in 10 families with linkage to chromosome 16. *Annals of Neurology*, **2003**, 53, 596-606	9.4	110
166	Mechanisms of cerebral cortical patterning in mice and humans. *Nature Neuroscience*, **2001**, 4 Suppl, 1199-206	25.5	109
165	Resolving rates of mutation in the brain using single-neuron genomics. *ELife*, **2016**, 5,	8.9	109
164	A comparative proteomic analysis of human and rat embryonic cerebrospinal fluid. *Journal of Proteome Research*, **2007**, 6, 3537-48	5.6	107
163	Impaired neuronal positioning and dendritogenesis in the neocortex after cell-autonomous Dab1 suppression. *Journal of Neuroscience*, **2006**, 26, 1767-75	6.6	107
162	Filamin A and Filamin B are co-expressed within neurons during periods of neuronal migration and can physically interact. *Human Molecular Genetics*, **2002**, 11, 2845-54	5.6	107
161	The DCX-domain tandems of doublecortin and doublecortin-like kinase. *Nature Structural and Molecular Biology*, **2003**, 10, 324-33	17.6	103
160	Genotype-phenotype analysis of human frontoparietal polymicrogyria syndromes. *Annals of Neurology*, **2005**, 58, 680-7	9.4	103
159	Targeted DNA Sequencing from Autism Spectrum Disorder Brains Implicates Multiple Genetic Mechanisms. *Neuron*, **2015**, 88, 910-917	13.9	100
158	An autosomal recessive form of bilateral frontoparietal polymicrogyria maps to chromosome 16q12.2-21. *American Journal of Human Genetics*, **2002**, 70, 1028-33	11	97
157	Birthdate and cell marker analysis of scrambler: a novel mutation affecting cortical development with a reeler-like phenotype. *Journal of Neuroscience*, **1997**, 17, 9204-11	6.6	94
156	Homozygous PLCB1 deletion associated with malignant migrating partial seizures in infancy. *Epilepsia*, **2012**, 53, e146-50	6.4	93
155	Molecular basis for specific regulation of neuronal kinesin-3 motors by doublecortin family proteins. *Molecular Cell*, **2012**, 47, 707-21	17.6	93
154	Disease-associated mutations affect GPR56 protein trafficking and cell surface expression. *Human Molecular Genetics*, **2007**, 16, 1972-85	5.6	92
153	SLC25A22 is a novel gene for migrating partial seizures in infancy. *Annals of Neurology*, **2013**, 74, 873-82	9.4	89

152	Genetic and neuroradiological heterogeneity of double cortex syndrome. *Annals of Neurology*, **2000**, 47, 265-269	9.4	89
151	Genetic changes shaping the human brain. *Developmental Cell*, **2015**, 32, 423-34	10.2	87
150	Mutations in QARS, encoding glutaminyl-tRNA synthetase, cause progressive microcephaly, cerebral-cerebellar atrophy, and intractable seizures. *American Journal of Human Genetics*, **2014**, 94, 547-58	11	87
149	Cux-2 controls the proliferation of neuronal intermediate precursors of the cortical subventricular zone. *Cerebral Cortex*, **2008**, 18, 1758-70	5.1	87
148	Centriolar satellites assemble centrosomal microcephaly proteins to recruit CDK2 and promote centriole duplication. *ELife*, **2015**, 4,	8.9	85
147	Microcephaly Proteins Wdr62 and Aspm Define a Mother Centriole Complex Regulating Centriole Biogenesis, Apical Complex, and Cell Fate. *Neuron*, **2016**, 92, 813-828	13.9	82
146	The apical complex couples cell fate and cell survival to cerebral cortical development. *Neuron*, **2010**, 66, 69-84	13.9	81
145	Aspm knockout ferret reveals an evolutionary mechanism governing cerebral cortical size. *Nature*, **2018**, 556, 370-375	50.4	77
144	Identification of neural outgrowth genes using genome-wide RNAi. *PLoS Genetics*, **2008**, 4, e1000111	6	77
143	Cortical malformations and epilepsy. *Mental Retardation and Developmental Disabilities Research Reviews*, **2000**, 6, 268-80		77
142	Consistent chromosome abnormalities identify novel polymicrogyria loci in 1p36.3, 2p16.1-p23.1, 4q21.21-q22.1, 6q26-q27, and 21q2. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1637-54	2.5	76
141	Insights into the gyrification of developing ferret brain by magnetic resonance imaging. *Journal of Anatomy*, **2007**, 210, 66-77	2.9	75
140	Innovations present in the primate interneuron repertoire. *Nature*, **2020**, 586, 262-269	50.4	74
139	Bilateral periventricular nodular heterotopia due to filamin 1 gene mutation: widespread glomeruloid microvascular anomaly and dysplastic cytoarchitecture in the cerebral cortex. *Acta Neuropathologica*, **2002**, 104, 649-57	14.3	73
138	A homozygous mutation in the tight-junction protein JAM3 causes hemorrhagic destruction of the brain, subependymal calcification, and congenital cataracts. *American Journal of Human Genetics*, **2010**, 87, 882-9	11	71
137	Evolution of Osteocrin as an activity-regulated factor in the primate brain. *Nature*, **2016**, 539, 242-247	50.4	69
136	CHMP1A encodes an essential regulator of BMI1-INK4A in cerebellar development. *Nature Genetics*, **2012**, 44, 1260-4	36.3	68
135	Sodium Channel SCN3A (Na1.3) Regulation of Human Cerebral Cortical Folding and Oral Motor Development. *Neuron*, **2018**, 99, 905-913.e7	13.9	64

134	Lis1-Nde1-dependent neuronal fate control determines cerebral cortical size and lamination. *Human Molecular Genetics*, **2008**, 17, 2441-55	5.6	64
133	Katanin p80 regulates human cortical development by limiting centriole and cilia number. *Neuron*, **2014**, 84, 1240-57	13.9	63
132	Targeted disruption of Tgif, the mouse ortholog of a human holoprosencephaly gene, does not result in holoprosencephaly in mice. *Molecular and Cellular Biology*, **2005**, 25, 3639-47	4.8	63
131	Recessive gene disruptions in autism spectrum disorder. *Nature Genetics*, **2019**, 51, 1092-1098	36.3	56
130	Genetic basis of developmental malformations of the cerebral cortex. *Archives of Neurology*, **2004**, 61, 637-40		56
129	Ethnically diverse causes of Walker-Warburg syndrome (WWS): FCMD mutations are a more common cause of WWS outside of the Middle East. *Human Mutation*, **2008**, 29, E231-41	4.7	54
128	A 2-Mb critical region implicated in the microcephaly associated with terminal 1q deletion syndrome. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1692-8	2.5	53
127	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. *Human Molecular Genetics*, **2014**, 23, 5781-92	5.6	51
126	The ESCRT-III Protein CHMP1A Mediates Secretion of Sonic Hedgehog on a Distinctive Subtype of Extracellular Vesicles. *Cell Reports*, **2018**, 24, 973-986.e8	10.6	49
125	Modest impact on risk for autism spectrum disorder of rare copy number variants at 15q11.2, specifically breakpoints 1 to 2. *Autism Research*, **2014**, 7, 355-62	5.1	49
124	Impact of PNKP mutations associated with microcephaly, seizures and developmental delay on enzyme activity and DNA strand break repair. *Nucleic Acids Research*, **2012**, 40, 6608-19	20.1	49
123	Etiological heterogeneity of familial periventricular heterotopia and hydrocephalus. *Brain and Development*, **2004**, 26, 326-34	2.2	49
122	The role of RELN in lissencephaly and neuropsychiatric disease. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2007**, 144B, 58-63	3.5	48
121	Periventricular heterotopia and the genetics of neuronal migration in the cerebral cortex. *American Journal of Human Genetics*, **1999**, 65, 19-24	11	48
120	Coexistence of widespread clones and large radial clones in early embryonic ferret cortex. *Cerebral Cortex*, **1999**, 9, 636-45	5.1	48
119	CC2D1A regulates human intellectual and social function as well as NF-Β signaling homeostasis. *Cell Reports*, **2014**, 8, 647-55	10.6	45
118	Genomic and evolutionary analyses of asymmetrically expressed genes in human fetal left and right cerebral cortex. *Cerebral Cortex*, **2006**, 16 Suppl 1, i18-25	5.1	44
117	Allelic diversity in human developmental neurogenetics: insights into biology and disease. *Neuron*, **2010**, 68, 245-53	13.9	43

116	Disorders of Microtubule Function in Neurons: Imaging Correlates. *American Journal of Neuroradiology*, **2016**, 37, 528-35	4.4	43
115	Linked-read analysis identifies mutations in single-cell DNA-sequencing data. *Nature Genetics*, **2019**, 51, 749-754	36.3	42
114	Bilateral frontoparietal polymicrogyria, Lennox-Gastaut syndrome, and GPR56 gene mutations. *Epilepsia*, **2009**, 50, 1344-53	6.4	39
113	Periventricular nodular heterotopia and Williams syndrome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1305-11	2.5	39
112	Cerebral cortical neuron diversity and development at single-cell resolution. *Current Opinion in Neurobiology*, **2017**, 42, 9-16	7.6	38
111	Genes that regulate neuronal migration in the cerebral cortex. *Epilepsy Research*, **1999**, 36, 143-54	3	38
110	Biallelic mutations in human DCC cause developmental split-brain syndrome. *Nature Genetics*, **2017**, 49, 606-612	36.3	37
109	Ion Channel Functions in Early Brain Development. *Trends in Neurosciences*, **2020**, 43, 103-114	13.3	37
108	Impaired proliferation and migration in human Miller-Dieker neural precursors. *Annals of Neurology*, **2006**, 60, 137-44	9.4	34
107	Loss of PCLO function underlies pontocerebellar hypoplasia type III. *Neurology*, **2015**, 84, 1745-50	6.5	33
106	Periventricular heterotopia: new insights into Ehlers-Danlos syndrome. *Clinical Medicine and Research*, **2005**, 3, 229-33	1.4	33
105	G-protein coupled receptor 56 promotes myoblast fusion through serum response factor- and nuclear factor of activated T-cell-mediated signalling but is not essential for muscle development in vivo. *FEBS Journal*, **2013**, 280, 6097-113	5.7	31
104	Transcription factor Lmo4 defines the shape of functional areas in developing cortices and regulates sensorimotor control. *Developmental Biology*, **2009**, 327, 132-42	3.1	29
103	Developmental and degenerative features in a complicated spastic paraplegia. *Annals of Neurology*, **2010**, 67, 516-25	9.4	28
102	Cytoplasmic LEK1 is a regulator of microtubule function through its interaction with the LIS1 pathway. *Proceedings of the National Academy of Sciences of the United States of America*, **2005**, 102, 8549-54	11.5	28
101	Integrated genome and transcriptome sequencing identifies a noncoding mutation in the genome replication factor as the cause of microcephaly-micromelia syndrome. *Genome Research*, **2017**, 27, 1323-1335	9.7	27
100	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. *PLoS Genetics*, **2018**, 14, e1007281	6	27
99	METTL23, a transcriptional partner of GABPA, is essential for human cognition. *Human Molecular Genetics*, **2014**, 23, 3456-66	5.6	27

98	Thoracic aortic aneurysm in patients with loss of function Filamin A mutations: Clinical characterization, genetics, and recommendations. *American Journal of Medical Genetics, Part A*, **2018**, 176, 337-350	2.5	26
97	Novel loss-of-function variants in DIAPH1 associated with syndromic microcephaly, blindness, and early onset seizures. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 435-440	2.5	26
96	DCC mutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. *Human Mutation*, **2018**, 39, 23-39	4.7	26
95	Genomic variants and variations in malformations of cortical development. *Pediatric Clinics of North America*, **2015**, 62, 571-85	3.6	25
94	Detecting natural selection by empirical comparison to random regions of the genome. *Human Molecular Genetics*, **2009**, 18, 4853-67	5.6	25
93	An autosomal recessive form of spastic cerebral palsy (CP) with microcephaly and mental retardation. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1504-10	2.5	24
92	Control of a neuronal morphology program by an RNA-binding zinc finger protein, Unkempt. *Genes and Development*, **2015**, 29, 501-12	12.6	23
91	Developmental genetic malformations of the cerebral cortex. *Current Neurology and Neuroscience Reports*, **2003**, 3, 433-41	6.6	23
90	Genetics of neuronal migration in the cerebral cortex. *Mental Retardation and Developmental Disabilities Research Reviews*, **2000**, 6, 34-40		23
89	The Epigenetic State of PRDM16-Regulated Enhancers in Radial Glia Controls Cortical Neuron Position. *Neuron*, **2018**, 98, 945-962.e8	13.9	23
88	Cc2d1a Loss of Function Disrupts Functional and Morphological Development in Forebrain Neurons Leading to Cognitive and Social Deficits. *Cerebral Cortex*, **2017**, 27, 1670-1685	5.1	22
87	Genome aging: somatic mutation in the brain links age-related decline with disease and nominates pathogenic mechanisms. *Human Molecular Genetics*, **2019**, 28, R197-R206	5.6	22
86	Genetics of disorders of cortical development. *Neuroimaging Clinics of North America*, **2004**, 14, 219-29, viii	3	22
85	Cryptic t(1;12)(q44;p13.3) translocation in a previously described syndrome with polymicrogyria, segregating as an apparently X-linked trait. *American Journal of Medical Genetics Part A*, **2003**, 117A, 65-71		21
84	New innovations: therapeutic opportunities for intellectual disabilities. *Annals of Neurology*, **2013**, 74, 382-90	9.4	20
83	A microRNA negative feedback loop downregulates vesicle transport and inhibits fear memory. *ELife*, **2016**, 5,	8.9	20
82	Accurate detection of mosaic variants in sequencing data without matched controls. *Nature Biotechnology*, **2020**, 38, 314-319	44.5	20
81	Delineation of the clinical, molecular and cellular aspects of novel JAM3 mutations underlying the autosomal recessive hemorrhagic destruction of the brain, subependymal calcification, and congenital cataracts. *Human Mutation*, **2013**, 34, 498-505	4.7	19

80	Brain evolution and uniqueness in the human genome. *Cell*, **2006**, 126, 1033-5	56.2	19
79	The landscape of somatic mutation in cerebral cortex of autistic and neurotypical individuals revealed by ultra-deep whole-genome sequencing. *Nature Neuroscience*, **2021**, 24, 176-185	25.5	19
78	Doublecortin is expressed in articular chondrocytes. *Biochemical and Biophysical Research Communications*, **2007**, 363, 694-700	3.4	18
77	Clonal mixing, clonal restriction, and specification of cell types in the developing rat olfactory bulb **1999**, 403, 106-118		18
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