

# Christopher A Walsh

## List of Publications by Year in descending order

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259  
papers

44,083  
citations

1701

104  
h-index

2381

198  
g-index

313  
all docs

313  
docs citations

313  
times ranked

39862  
citing authors

#	ARTICLE	IF	CITATIONS
1	Synaptic, transcriptional and chromatin genes disrupted in autism. <i>Nature</i> , 2014, 515, 209-215.	13.7	2,254
2	Association between Microdeletion and Microduplication at 16p11.2 and Autism. <i>New England Journal of Medicine</i> , 2008, 358, 667-675.	13.9	1,476
3	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. <i>Cell</i> , 2020, 180, 568-584.e23.	13.5	1,422
4	Regulation of Cerebral Cortical Size by Control of Cell Cycle Exit in Neural Precursors. <i>Science</i> , 2002, 297, 365-369.	6.0	1,303
5	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. <i>Neuron</i> , 2015, 87, 1215-1233.	3.8	1,219
6	Doublecortin Is a Microtubule-Associated Protein and Is Expressed Widely by Migrating Neurons. <i>Neuron</i> , 1999, 23, 257-271.	3.8	1,200
7	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. <i>Neuron</i> , 2011, 70, 863-885.	3.8	1,146
8	Directed migration of neural stem cells to sites of CNS injury by the stromal cell-derived factor 1 $\alpha$ /CXCR4 chemokine receptor 4 pathway. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 18117-18122.	3.3	1,023
9	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
10	Mutations in filamin 1 Prevent Migration of Cerebral Cortical Neurons in Human Periventricular Heterotopia. <i>Neuron</i> , 1998, 21, 1315-1325.	3.8	811
11	Autosomal recessive lissencephaly with cerebellar hypoplasia is associated with human RELN mutations. <i>Nature Genetics</i> , 2000, 26, 93-96.	9.4	798
12	Identifying Autism Loci and Genes by Tracing Recent Shared Ancestry. <i>Science</i> , 2008, 321, 218-223.	6.0	688
13	Mutations in the O-Mannosyltransferase Gene POMT1 Give Rise to the Severe Neuronal Migration Disorder Walker-Warburg Syndrome. <i>American Journal of Human Genetics</i> , 2002, 71, 1033-1043.	2.6	636
14	The Cerebrospinal Fluid Provides a Proliferative Niche for Neural Progenitor Cells. <i>Neuron</i> , 2011, 69, 893-905.	3.8	543
15	ASPM is a major determinant of cerebral cortical size. <i>Nature Genetics</i> , 2002, 32, 316-320.	9.4	538
16	Reelin Binds $\alpha$ 3 $\beta$ 1 Integrin and Inhibits Neuronal Migration. <i>Neuron</i> , 2000, 27, 33-44.	3.8	527
17	A centrosomal mechanism involving CDK5RAP2 and CENPJ controls brain size. <i>Nature Genetics</i> , 2005, 37, 353-355.	9.4	520
18	Somatic Mutation, Genomic Variation, and Neurological Disease. <i>Science</i> , 2013, 341, 1237758.	6.0	501

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19	Single-Neuron Sequencing Analysis of L1 Retrotransposition and Somatic Mutation in the Human Brain. <i>Cell</i> , 2012, 151, 483-496.	13.5	500
20	G Protein-Coupled Receptor-Dependent Development of Human Frontal Cortex. <i>Science</i> , 2004, 303, 2033-2036.	6.0	498
21	Aging and neurodegeneration are associated with increased mutations in single human neurons. <i>Science</i> , 2018, 359, 555-559.	6.0	496
22	Somatic mutation in single human neurons tracks developmental and transcriptional history. <i>Science</i> , 2015, 350, 94-98.	6.0	486
23	Expression of Cux-1 and Cux-2 in the subventricular zone and upper layers II-IV of the cerebral cortex. <i>Journal of Comparative Neurology</i> , 2004, 479, 168-180.	0.9	461
24	The many faces of filamin: A versatile molecular scaffold for cell motility and signalling. <i>Nature Cell Biology</i> , 2004, 6, 1034-1038.	4.6	441
25	PAK3 mutation in nonsyndromic X-linked mental retardation. <i>Nature Genetics</i> , 1998, 20, 25-30.	9.4	432
26	Characterization of Foxp2 and Foxp1 mRNA and protein in the developing and mature brain. <i>Journal of Comparative Neurology</i> , 2003, 460, 266-279.	0.9	432
27	Somatic Activation of AKT3 Causes Hemispheric Developmental Brain Malformations. <i>Neuron</i> , 2012, 74, 41-48.	3.8	413
28	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. <i>Neuron</i> , 2013, 77, 259-273.	3.8	383
29	Abnormal cerebellar development and axonal decussation due to mutations in AH11 in Joubert syndrome. <i>Nature Genetics</i> , 2004, 36, 1008-1013.	9.4	374
30	Mutations in ARFGEF2 implicate vesicle trafficking in neural progenitor proliferation and migration in the human cerebral cortex. <i>Nature Genetics</i> , 2004, 36, 69-76.	9.4	340
31	Early Asymmetry of Gene Transcription in Embryonic Human Left and Right Cerebral Cortex. <i>Science</i> , 2005, 308, 1794-1798.	6.0	339
32	Clinical Genetic Testing for Patients With Autism Spectrum Disorders. <i>Pediatrics</i> , 2010, 125, e727-e735.	1.0	339
33	Mitotic Spindle Regulation by Nde1 Controls Cerebral Cortical Size. <i>Neuron</i> , 2004, 44, 279-293.	3.8	327
34	Somatic Mutations in Cerebral Cortical Malformations. <i>New England Journal of Medicine</i> , 2014, 371, 733-743.	13.9	326
35	Neuronal migration disorders: from genetic diseases to developmental mechanisms. <i>Trends in Neurosciences</i> , 2000, 23, 352-359.	4.2	325
36	Filamin A (FLNA) is required for cell-cell contact in vascular development and cardiac morphogenesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 19836-19841.	3.3	306

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37	Doublecortin Is Required in Mice for Lamination of the Hippocampus But Not the Neocortex. <i>Journal of Neuroscience</i> , 2002, 22, 7548-7557.	1.7	294
38	Molecular approaches to brain asymmetry and handedness. <i>Nature Reviews Neuroscience</i> , 2006, 7, 655-662.	4.9	287
39	Mutations in Human Accelerated Regions Disrupt Cognition and Social Behavior. <i>Cell</i> , 2016, 167, 341-354.e12.	13.5	280
40	Cell-Type-Specific Alternative Splicing Governs Cell Fate in the Developing Cerebral Cortex. <i>Cell</i> , 2016, 166, 1147-1162.e15.	13.5	276
41	LIS1 Regulates CNS Lamination by Interacting with mNudE, a Central Component of the Centrosome. <i>Neuron</i> , 2000, 28, 665-679.	3.8	271
42	Patterning of the Dorsal Telencephalon and Cerebral Cortex by a Roof Plate-Lhx2 Pathway. <i>Neuron</i> , 2001, 32, 591-604.	3.8	268
43	Mutations in PNKP cause microcephaly, seizures and defects in DNA repair. <i>Nature Genetics</i> , 2010, 42, 245-249.	9.4	268
44	Genetic Interactions between Doublecortin and Doublecortin-like Kinase in Neuronal Migration and Axon Outgrowth. <i>Neuron</i> , 2006, 49, 41-53.	3.8	263
45	Single-Cell, Genome-wide Sequencing Identifies Clonal Somatic Copy-Number Variation in the Human Brain. <i>Cell Reports</i> , 2014, 8, 1280-1289.	2.9	260
46	Aberrant Splicing of a Mouse disabled Homolog, mdab1, in the scrambler Mouse. <i>Neuron</i> , 1997, 19, 239-249.	3.8	259
47	Mutations in WDR62, encoding a centrosome-associated protein, cause microcephaly with simplified gyri and abnormal cortical architecture. <i>Nature Genetics</i> , 2010, 42, 1015-1020.	9.4	259
48	Cux1 and Cux2 Regulate Dendritic Branching, Spine Morphology, and Synapses of the Upper Layer Neurons of the Cortex. <i>Neuron</i> , 2010, 66, 523-535.	3.8	247
49	Single-cell analysis reveals transcriptional heterogeneity of neural progenitors in human cortex. <i>Nature Neuroscience</i> , 2015, 18, 637-646.	7.1	247
50	Somatic Mutations Activating the mTOR Pathway in Dorsal Telencephalic Progenitors Cause a Continuum of Cortical Dysplasias. <i>Cell Reports</i> , 2017, 21, 3754-3766.	2.9	247
51	Increased Neuronal Production, Enlarged Forebrains and Cytoarchitectural Distortions in beta-Catenin Overexpressing Transgenic Mice. <i>Cerebral Cortex</i> , 2003, 13, 599-606.	1.6	243
52	Evolutionarily Dynamic Alternative Splicing of <i>GPR56</i> Regulates Regional Cerebral Cortical Patterning. <i>Science</i> , 2014, 343, 764-768.	6.0	238
53	Mammalian target of rapamycin pathway mutations cause hemimegalencephaly and focal cortical dysplasia. <i>Annals of Neurology</i> , 2015, 77, 720-725.	2.8	235
54	Cell Lineage Analysis in Human Brain Using Endogenous Retroelements. <i>Neuron</i> , 2015, 85, 49-59.	3.8	234

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55	Cdk5rap2 regulates centrosome function and chromosome segregation in neuronal progenitors. <i>Development (Cambridge)</i> , 2010, 137, 1907-1917.	1.2	233
56	Human Brain Malformations and Their Lessons for Neuronal Migration. <i>Annual Review of Neuroscience</i> , 2001, 24, 1041-1070.	5.0	221
57	The Genetics of Primary Microcephaly. <i>Annual Review of Genomics and Human Genetics</i> , 2018, 19, 177-200.	2.5	220
58	Deletions of <i>NRXN1</i> (neurexin1) predispose to a wide spectrum of developmental disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 937-947.	1.1	217
59	Molecular insights into human brain evolution. <i>Nature</i> , 2005, 437, 64-67.	13.7	214
60	Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. <i>Nature Neuroscience</i> , 2017, 20, 1217-1224.	7.1	212
61	GPR56 Regulates Pial Basement Membrane Integrity and Cortical Lamination. <i>Journal of Neuroscience</i> , 2008, 28, 5817-5826.	1.7	209
62	Intersection of diverse neuronal genomes and neuropsychiatric disease: The Brain Somatic Mosaicism Network. <i>Science</i> , 2017, 356, .	6.0	206
63	Innovations present in the primate interneuron repertoire. <i>Nature</i> , 2020, 586, 262-269.	13.7	206
64	Building a lineage from single cells: genetic techniques for cell lineage tracking. <i>Nature Reviews Genetics</i> , 2017, 18, 230-244.	7.7	204
65	Genetic causes of microcephaly and lessons for neuronal development. <i>Wiley Interdisciplinary Reviews: Developmental Biology</i> , 2013, 2, 461-478.	5.9	199
66	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. <i>American Journal of Human Genetics</i> , 2011, 88, 536-547.	2.6	196
67	DCAMKL1 Encodes a Protein Kinase with Homology to Doublecortin that Regulates Microtubule Polymerization. <i>Journal of Neuroscience</i> , 2000, 20, 9152-9161.	1.7	187
68	Somatic mosaicism and neurodevelopmental disease. <i>Nature Neuroscience</i> , 2018, 21, 1504-1514.	7.1	186
69	Protein-Protein interactions, cytoskeletal regulation and neuronal migration. <i>Nature Reviews Neuroscience</i> , 2001, 2, 408-416.	4.9	184
70	A mapping label required for normal scale of body representation in the cortex. <i>Nature Neuroscience</i> , 2000, 3, 358-365.	7.1	178
71	Accelerated Evolution of the ASPM Gene Controlling Brain Size Begins Prior to Human Brain Expansion. <i>PLoS Biology</i> , 2004, 2, e126.	2.6	176
72	Characterization of mutations in the gene doublecortin in patients with double cortex syndrome. <i>Annals of Neurology</i> , 1999, 45, 146-153.	2.8	175

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73	Autism and Brain Development. <i>Cell</i> , 2008, 135, 396-400.	13.5	175
74	Neurogenesis at the Brain–Cerebrospinal Fluid Interface. <i>Annual Review of Cell and Developmental Biology</i> , 2011, 27, 653-679.	4.0	175
75	The microcephaly ASPM gene is expressed in proliferating tissues and encodes for a mitotic spindle protein. <i>Human Molecular Genetics</i> , 2005, 14, 2155-2165.	1.4	172
76	Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of $\alpha$ -Dystroglycan. <i>American Journal of Human Genetics</i> , 2013, 92, 354-365.	2.6	172
77	Disruption of neural progenitors along the ventricular and subventricular zones in periventricular heterotopia. <i>Human Molecular Genetics</i> , 2009, 18, 497-516.	1.4	169
78	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 541-547.	2.6	167
79	Whole-Exome Sequencing and Homozygosity Analysis Implicate Depolarization-Regulated Neuronal Genes in Autism. <i>PLoS Genetics</i> , 2012, 8, e1002635.	1.5	164
80	Protein-Truncating Mutations in ASPM Cause Variable Reduction in Brain Size. <i>American Journal of Human Genetics</i> , 2003, 73, 1170-1177.	2.6	163
81	Molecular genetics of human microcephaly. <i>Current Opinion in Neurology</i> , 2001, 14, 151-156.	1.8	158
82	The <i>hyh</i> mutation uncovers roles for $\alpha$ -Snap in apical protein localization and control of neural cell fate. <i>Nature Genetics</i> , 2004, 36, 264-270.	9.4	158
83	The exon junction complex component Magoh controls brain size by regulating neural stem cell division. <i>Nature Neuroscience</i> , 2010, 13, 551-558.	7.1	156
84	Microcephaly Gene Links Trithorax and REST/NRSF to Control Neural Stem Cell Proliferation and Differentiation. <i>Cell</i> , 2012, 151, 1097-1112.	13.5	153
85	What disorders of cortical development tell us about the cortex: one plus one does not always make two. <i>Current Opinion in Genetics and Development</i> , 2011, 21, 333-339.	1.5	151
86	Genetic Malformations of the Human Cerebral Cortex. <i>Neuron</i> , 1999, 23, 19-29.	3.8	146
87	The Diverse Genetic Landscape of Neurodevelopmental Disorders. <i>Annual Review of Genomics and Human Genetics</i> , 2014, 15, 195-213.	2.5	146
88	Targeted DNA Sequencing from Autism Spectrum Disorder Brains Implicates Multiple Genetic Mechanisms. <i>Neuron</i> , 2015, 88, 910-917.	3.8	142
89	Resolving rates of mutation in the brain using single-neuron genomics. <i>ELife</i> , 2016, 5, .	2.8	139
90	Patient Mutations in Doublecortin Define a Repeated Tubulin-binding Domain. <i>Journal of Biological Chemistry</i> , 2000, 275, 34442-34450.	1.6	138

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91	Somatic and Germline Mosaic Mutations in the doublecortin Gene Are Associated with Variable Phenotypes. <i>American Journal of Human Genetics</i> , 2000, 67, 574-581.	2.6	135
92	Both Doublecortin and Doublecortin-Like Kinase Play a Role in Cortical Interneuron Migration. <i>Journal of Neuroscience</i> , 2007, 27, 3875-3883.	1.7	133
93	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. <i>Biological Psychiatry</i> , 2015, 77, 775-784.	0.7	133
94	Smooth, rough and upside-down neocortical development. <i>Current Opinion in Genetics and Development</i> , 2002, 12, 320-327.	1.5	132
95	Mechanisms of cerebral cortical patterning in mice and humans. <i>Nature Neuroscience</i> , 2001, 4, 1199-1206.	7.1	130
96	Aspm knockout ferret reveals an evolutionary mechanism governing cerebral cortical size. <i>Nature</i> , 2018, 556, 370-375.	13.7	127
97	Genotype-phenotype analysis of human frontoparietal polymicrogyria syndromes. <i>Annals of Neurology</i> , 2005, 58, 680-687.	2.8	124
98	Filamin A and Filamin B are co-expressed within neurons during periods of neuronal migration and can physically interact. <i>Human Molecular Genetics</i> , 2002, 11, 2845-2854.	1.4	123
99	Missense mutation in PAK3, R67C, causes X-linked nonspecific mental retardation. <i>American Journal of Medical Genetics Part A</i> , 2000, 93, 294-298.	2.4	122
100	The DCX-domain tandems of doublecortin and doublecortin-like kinase. <i>Nature Structural and Molecular Biology</i> , 2003, 10, 324-333.	3.6	122
101	Bilateral frontoparietal polymicrogyria: Clinical and radiological features in 10 families with linkage to chromosome 16. <i>Annals of Neurology</i> , 2003, 53, 596-606.	2.8	120
102	Evolution of Osteocrin as an activity-regulated factor in the primate brain. <i>Nature</i> , 2016, 539, 242-247.	13.7	120
103	Impaired Neuronal Positioning and Dendritogenesis in the Neocortex after Cell-Autonomous Dab1 Suppression. <i>Journal of Neuroscience</i> , 2006, 26, 1767-1775.	1.7	119
104	A Comparative Proteomic Analysis of Human and Rat Embryonic Cerebrospinal Fluid. <i>Journal of Proteome Research</i> , 2007, 6, 3537-3548.	1.8	118
105	Centriolar satellites assemble centrosomal microcephaly proteins to recruit CDK2 and promote centriole duplication. <i>ELife</i> , 2015, 4, .	2.8	118
106	Molecular Basis for Specific Regulation of Neuronal Kinesin-3 Motors by Doublecortin Family Proteins. <i>Molecular Cell</i> , 2012, 47, 707-721.	4.5	116
107	Microcephaly Proteins Wdr62 and Aspm Define a Mother Centriole Complex Regulating Centriole Biogenesis, Apical Complex, and Cell Fate. <i>Neuron</i> , 2016, 92, 813-828.	3.8	116
108	Genetic Changes Shaping the Human Brain. <i>Developmental Cell</i> , 2015, 32, 423-434.	3.1	115

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109	An Autosomal Recessive Form of Bilateral Frontoparietal Polymicrogyria Maps to Chromosome 16q12.2-21. <i>American Journal of Human Genetics</i> , 2002, 70, 1028-1033.	2.6	113
110	Disease-associated mutations affect GPR56 protein trafficking and cell surface expression. <i>Human Molecular Genetics</i> , 2007, 16, 1972-1985.	1.4	109
111	Sodium Channel SCN3A (NaV1.3) Regulation of Human Cerebral Cortical Folding and Oral Motor Development. <i>Neuron</i> , 2018, 99, 905-913.e7.	3.8	109
112	Recessive gene disruptions in autism spectrum disorder. <i>Nature Genetics</i> , 2019, 51, 1092-1098.	9.4	109
113	Mutations in QARS, Encoding Glutamyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. <i>American Journal of Human Genetics</i> , 2014, 94, 547-558.	2.6	106
114	Homozygous <i>PLCB1</i> deletion associated with malignant migrating partial seizures in infancy. <i>Epilepsia</i> , 2012, 53, e146-50.	2.6	104
115	<i>SLC25A22</i> is a novel gene for migrating partial seizures in infancy. <i>Annals of Neurology</i> , 2013, 74, 873-882.	2.8	102
116	Birthdate and Cell Marker Analysis of Scrambler: A Novel Mutation Affecting Cortical Development with a Reeler-Like Phenotype. <i>Journal of Neuroscience</i> , 1997, 17, 9204-9211.	1.7	100
117	The Apical Complex Couples Cell Fate and Cell Survival to Cerebral Cortical Development. <i>Neuron</i> , 2010, 66, 69-84.	3.8	97
118	Cux-2 Controls the Proliferation of Neuronal Intermediate Precursors of the Cortical Subventricular Zone. <i>Cerebral Cortex</i> , 2008, 18, 1758-1770.	1.6	96
119	Genetic and neuroradiological heterogeneity of double cortex syndrome. <i>Annals of Neurology</i> , 2000, 47, 265-269.	2.8	94
120	Consistent chromosome abnormalities identify novel polymicrogyria loci in 1p36.3, 2p16.1-p23.1, 4q21.21-q22.1, 6q26-q27, and 21q2. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1637-1654.	0.7	93
121	Somatic genomic changes in single Alzheimer's disease neurons. <i>Nature</i> , 2022, 604, 714-722.	13.7	92
122	Cortical malformations and epilepsy. <i>Mental Retardation and Developmental Disabilities Research Reviews</i> , 2000, 6, 268-280.	3.5	91
123	CHMP1A encodes an essential regulator of BMI1-INK4A in cerebellar development. <i>Nature Genetics</i> , 2012, 44, 1260-1264.	9.4	91
124	Rewiring of human neurodevelopmental gene regulatory programs by human accelerated regions. <i>Neuron</i> , 2021, 109, 3239-3251.e7.	3.8	91
125	Katanin p80 Regulates Human Cortical Development by Limiting Centriole and Cilia Number. <i>Neuron</i> , 2014, 84, 1240-1257.	3.8	89
126	Insights into the gyrification of developing ferret brain by magnetic resonance imaging. <i>Journal of Anatomy</i> , 2007, 210, 66-77.	0.9	88

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127	The <sc>ILAE</sc> consensus classification of focal cortical dysplasia: An update proposed by an ad hoc task force of the <sc>ILAE</sc> diagnostic methods commission. <i>Epilepsia</i> , 2022, 63, 1899-1919.	2.6	88
128	A Homozygous Mutation in the Tight-Junction Protein JAM3 Causes Hemorrhagic Destruction of the Brain, Subependymal Calcification, and Congenital Cataracts. <i>American Journal of Human Genetics</i> , 2010, 87, 882-889.	2.6	87
129	Identification of Neural Outgrowth Genes using Genome-Wide RNAi. <i>PLoS Genetics</i> , 2008, 4, e1000111.	1.5	85
130	Bilateral periventricular nodular heterotopia due to filamin 1 gene mutation: widespread glomeruloid microvascular anomaly and dysplastic cytoarchitecture in the cerebral cortex. <i>Acta Neuropathologica</i> , 2002, 104, 649-657.	3.9	84
131	The ESCRT-III Protein CHMP1A Mediates Secretion of Sonic Hedgehog on a Distinctive Subtype of Extracellular Vesicles. <i>Cell Reports</i> , 2018, 24, 973-986.e8.	2.9	79
132	Linked-read analysis identifies mutations in single-cell DNA-sequencing data. <i>Nature Genetics</i> , 2019, 51, 749-754.	9.4	76
133	Lis1-dependent neuronal fate control determines cerebral cortical size and lamination. <i>Human Molecular Genetics</i> , 2008, 17, 2441-2455.	1.4	73
134	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
135	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. <i>Human Molecular Genetics</i> , 2014, 23, 5781-5792.	1.4	72
136	Genetic Basis of Developmental Malformations of the Cerebral Cortex. <i>Archives of Neurology</i> , 2004, 61, 637.	4.9	68
137	Targeted Disruption of Tgif , the Mouse Ortholog of a Human Holoprosencephaly Gene, Does Not Result in Holoprosencephaly in Mice. <i>Molecular and Cellular Biology</i> , 2005, 25, 3639-3647.	1.1	68
138	Ethnically diverse causes of Walker-Warburg syndrome (WWS): FCMD mutations are a more common cause of WWS outside of the Middle East. <i>Human Mutation</i> , 2008, 29, E231-E241.	1.1	67
139	Ion Channel Functions in Early Brain Development. <i>Trends in Neurosciences</i> , 2020, 43, 103-114.	4.2	67
140	Landmarks of human embryonic development inscribed in somatic mutations. <i>Science</i> , 2021, 371, 1249-1253.	6.0	65
141	The role of RELN in lissencephaly and neuropsychiatric disease. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2007, 144B, 58-63.	1.1	63
142	A 2-Mb critical region implicated in the microcephaly associated with terminal 1q deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1692-1698.	0.7	62
143	Impact of PNKP mutations associated with microcephaly, seizures and developmental delay on enzyme activity and DNA strand break repair. <i>Nucleic Acids Research</i> , 2012, 40, 6608-6619.	6.5	62
144	Biallelic mutations in human DCC cause developmental split-brain syndrome. <i>Nature Genetics</i> , 2017, 49, 606-612.	9.4	62

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145	CC2D1A Regulates Human Intellectual and Social Function as well as NF- $\kappa$ B Signaling Homeostasis. <i>Cell Reports</i> , 2014, 8, 647-655.	2.9	60
146	Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15q11.2, Specifically Breakpoints 1 to 2. <i>Autism Research</i> , 2014, 7, 355-362.	2.1	59
147	Coexistence of Widespread Clones and Large Radial Clones in Early Embryonic Ferret Cortex. <i>Cerebral Cortex</i> , 1999, 9, 636-645.	1.6	56
148	Etiological heterogeneity of familial periventricular heterotopia and hydrocephalus. <i>Brain and Development</i> , 2004, 26, 326-334.	0.6	56
149	Disorders of Microtubule Function in Neurons: Imaging Correlates. <i>American Journal of Neuroradiology</i> , 2016, 37, 528-535.	1.2	56
150	The Epigenetic State of PRDM16-Regulated Enhancers in Radial Glia Controls Cortical Neuron Position. <i>Neuron</i> , 2018, 98, 945-962.e8.	3.8	54
151	Accurate detection of mosaic variants in sequencing data without matched controls. <i>Nature Biotechnology</i> , 2020, 38, 314-319.	9.4	54
152	Toward a better definition of focal cortical dysplasia: An iterative histopathological and genetic agreement trial. <i>Epilepsia</i> , 2021, 62, 1416-1428.	2.6	54
153	Allelic Diversity in Human Developmental Neurogenetics: Insights into Biology and Disease. <i>Neuron</i> , 2010, 68, 245-253.	3.8	53
154	Genomic and Evolutionary Analyses of Asymmetrically Expressed Genes in Human Fetal Left and Right Cerebral Cortex. <i>Cerebral Cortex</i> , 2006, 16, i18-i25.	1.6	51
155	Cerebral cortical neuron diversity and development at single-cell resolution. <i>Current Opinion in Neurobiology</i> , 2017, 42, 9-16.	2.0	51
156	Periventricular Heterotopia and the Genetics of Neuronal Migration in the Cerebral Cortex. <i>American Journal of Human Genetics</i> , 1999, 65, 19-24.	2.6	49
157	METTL23, a transcriptional partner of GABPA, is essential for human cognition. <i>Human Molecular Genetics</i> , 2014, 23, 3456-3466.	1.4	47
158	Bilateral frontoparietal polymicrogyria, Lennox-Gastaut syndrome, and GPR56 gene mutations. <i>Epilepsia</i> , 2009, 50, 1344-1353.	2.6	46
159	Impaired neurogenesis alters brain biomechanics in a neuroprogenitor-based genetic subtype of congenital hydrocephalus. <i>Nature Neuroscience</i> , 2022, 25, 458-473.	7.1	46
160	Periventricular nodular heterotopia and Williams syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1305-1311.	0.7	45
161	Loss of PCLO function underlies pontocerebellar hypoplasia type III. <i>Neurology</i> , 2015, 84, 1745-1750.	1.5	45
162	Genes that regulate neuronal migration in the cerebral cortex. <i>Epilepsy Research</i> , 1999, 36, 143-154.	0.8	42

#	ARTICLE	IF	CITATIONS
163	<i>DCC</i> mutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. <i>Human Mutation</i> , 2018, 39, 23-39.	1.1	41
164	Impaired proliferation and migration in human Miller-Dieker neural precursors. <i>Annals of Neurology</i> , 2006, 60, 137-144.	2.8	40
165	Integrated genome and transcriptome sequencing identifies a noncoding mutation in the genome replication factor <i>DONSON</i> as the cause of microcephaly-micromelia syndrome. <i>Genome Research</i> , 2017, 27, 1323-1335.	2.4	40
166	Thoracic aortic aneurysm in patients with loss of function <i>Filamin A</i> mutations: Clinical characterization, genetics, and recommendations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 337-350.	0.7	40
167	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. <i>PLoS Genetics</i> , 2018, 14, e1007281.	1.5	40
168	Periventricular Heterotopia: New Insights into Ehlers-Danlos Syndrome. <i>Clinical Medicine and Research</i> , 2005, 3, 229-233.	0.4	39
169	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 <i>in vivo</i>. <i>FEBS Journal</i> , 2013, 280, 6097-6113.	2.2	39
170	Genetic mosaicism in the human brain: from lineage tracing to neuropsychiatric disorders. <i>Nature Reviews Neuroscience</i> , 2022, 23, 275-286.	4.9	39
171	Genome aging: somatic mutation in the brain links age-related decline with disease and nominates pathogenic mechanisms. <i>Human Molecular Genetics</i> , 2019, 28, R197-R206.	1.4	37
172	Novel loss-of-function variants in <i>DIAPH1</i> associated with syndromic microcephaly, blindness, and early onset seizures. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 435-440.	0.7	36
173	Cc2d1a Loss of Function Disrupts Functional and Morphological Development in Forebrain Neurons Leading to Cognitive and Social Deficits. <i>Cerebral Cortex</i> , 2017, 27, 1670-1685.	1.6	36
174	Large mosaic copy number variations confer autism risk. <i>Nature Neuroscience</i> , 2021, 24, 197-203.	7.1	36
175	Control of a neuronal morphology program by an RNA-binding zinc finger protein, Unkempt. <i>Genes and Development</i> , 2015, 29, 501-512.	2.7	35
176	16p11.2 deletion is associated with hyperactivation of human iPSC-derived dopaminergic neuron networks and is rescued by RHOA inhibition in vitro. <i>Nature Communications</i> , 2021, 12, 2897.	5.8	35
177	Transcription factor Lmo4 defines the shape of functional areas in developing cortices and regulates sensorimotor control. <i>Developmental Biology</i> , 2009, 327, 132-142.	0.9	34
178	Parallel RNA and DNA analysis after deep sequencing (PRDD-seq) reveals cell type-specific lineage patterns in human brain. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 13886-13895.	3.3	33
179	New innovations: Therapeutic opportunities for intellectual disabilities. <i>Annals of Neurology</i> , 2013, 74, 382-390.	2.8	32
180	Genomic Variants and Variations in Malformations of Cortical Development. <i>Pediatric Clinics of North America</i> , 2015, 62, 571-585.	0.9	32

#	ARTICLE	IF	CITATIONS
181	Brain Somatic Mutation in Aging and Alzheimer's Disease. Annual Review of Genomics and Human Genetics, 2021, 22, 239-256.	2.5	32
182	Developmental and degenerative features in a complicated spastic paraplegia. Annals of Neurology, 2010, 67, 516-525.	2.8	31
183	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, 8549-8554.	3.3	30
184	Delineation of the Clinical, Molecular and Cellular Aspects of Novel <i>JAM3</i> Mutations Underlying the Autosomal Recessive Hemorrhagic Destruction of the Brain, Subependymal Calcification, and Congenital Cataracts. Human Mutation, 2013, 34, 498-505.	1.1	30
185	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	2.6	30
186	Recent Advances in Understanding the Genetic Architecture of Autism. Annual Review of Genomics and Human Genetics, 2020, 21, 289-304.	2.5	30
187	A microRNA negative feedback loop downregulates vesicle transport and inhibits fear memory. ELife, 2016, 5, .	2.8	29
188	Genetics of disorders of cortical development. Neuroimaging Clinics of North America, 2004, 14, 219-229.	0.5	28
189	Detecting natural selection by empirical comparison to random regions of the genome. Human Molecular Genetics, 2009, 18, 4853-4867.	1.4	27
190	Genetics of neuronal migration in the cerebral cortex. , 2000, 6, 34-40.		26
191	Comprehensive identification of somatic nucleotide variants in human brain tissue. Genome Biology, 2021, 22, 92.	3.8	26
192	Somatic mosaicism reveals clonal distributions of neocortical development. Nature, 2022, 604, 689-696.	13.7	26
193	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.	2.4	25
194	An autosomal recessive form of spastic cerebral palsy (CP) with microcephaly and mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 1504-1510.	0.7	25
195	Developmental genetic malformations of the cerebral cortex. Current Neurology and Neuroscience Reports, 2003, 3, 433-441.	2.0	24
196	Doublecortin is expressed in articular chondrocytes. Biochemical and Biophysical Research Communications, 2007, 363, 694-700.	1.0	23
197	<i>PSMD12</i> haploinsufficiency in a neurodevelopmental disorder with autistic features. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 736-745.	1.1	23
198	Brain ventricles as windows into brain development and disease. Neuron, 2022, 110, 12-15.	3.8	23

#	ARTICLE	IF	CITATIONS
199	Neocortical neuronal arrangement in Miller Dieker syndrome. <i>Acta Neuropathologica</i> , 2006, 111, 489-496.	3.9	22
200	Somatic Mutation in Pediatric Neurological Diseases. <i>Pediatric Neurology</i> , 2018, 87, 20-22.	1.0	22
201	Clonal mixing, clonal restriction, and specification of cell types in the developing rat olfactory bulb. <i>Development</i> , 1999, 126, 106-118.		21
202	Brain Evolution and Uniqueness in the Human Genome. <i>Cell</i> , 2006, 126, 1033-1035.	13.5	21
203	A novel form of lethal microcephaly with simplified gyral pattern and brain stem hypoplasia. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2761-2767.	0.7	20
204	Identification of a novel CNTNAP1 mutation causing arthrogryposis multiplex congenita with cerebral and cerebellar atrophy. <i>European Journal of Medical Genetics</i> , 2017, 60, 245-249.	0.7	20
205	Early role for a Na <sup>+</sup> ,K <sup>+</sup> -ATPase ( <i>ATP1A3</i> ) in brain development. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	20
206	Posterior Neocortex-Specific Regulation of Neuronal Migration by CEP85L Identifies Maternal Centriole-Dependent Activation of CDK5. <i>Neuron</i> , 2020, 106, 246-255.e6.	3.8	19
207	Rates and Patterns of Clonal Oncogenic Mutations in the Normal Human Brain. <i>Cancer Discovery</i> , 2022, 12, 172-185.	7.7	19
208	Somatic Mutations in Cerebral Cortical Malformations. <i>New England Journal of Medicine</i> , 2014, 371, 2037-2038.	13.9	18
209	Evolutionary Changes in Transcriptional Regulation: Insights into Human Behavior and Neurological Conditions. <i>Annual Review of Neuroscience</i> , 2018, 41, 185-206.	5.0	18
210	SFI1 promotes centriole duplication by recruiting USP9X to stabilize the microcephaly protein STIL. <i>Journal of Cell Biology</i> , 2019, 218, 2185-2197.	2.3	18
211	APP gene copy number changes reflect exogenous contamination. <i>Nature</i> , 2020, 584, E20-E28.	13.7	18
212	Reelin is expressed in the accessory olfactory system, but is not a guidance cue for vomeronasal axons. <i>Developmental Brain Research</i> , 2003, 140, 303-307.	2.1	17
213	Whole-genome analysis reveals the contribution of non-coding de novo transposon insertions to autism spectrum disorder. <i>Mobile DNA</i> , 2021, 12, 28.	1.3	17
214	Mutation in <i>PQBP1</i> is associated with periventricular heterotopia. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2888-2890.	0.7	16
215	Mapping form and function in the human brain: the emerging field of functional neuroimaging in cortical malformations. <i>Epilepsy and Behavior</i> , 2003, 4, 618-625.	0.9	15
216	PaSD-qc: quality control for single cell whole-genome sequencing data using power spectral density estimation. <i>Nucleic Acids Research</i> , 2018, 46, e20-e20.	6.5	14

#	ARTICLE	IF	CITATIONS
217	Polymicrogyria is Associated With Pathogenic Variants in PTEN. <i>Annals of Neurology</i> , 2020, 88, 1153-1164.	2.8	14
218	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
219	A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. <i>Genetics in Medicine</i> , 2021, 23, 1158-1162.	1.1	13
220	De novo variants in <i>TCF7L2</i> are associated with a syndromic neurodevelopmental disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2384-2390.	0.7	13
221	A YAC Contig in Xq22.3q23, fromDXS287toDXS8088, Spanning the Brain-Specific Genes doublecortin(DCX) andPAK3. <i>Genomics</i> , 1998, 52, 214-218.	1.3	12
222	Isolation of Cerebrospinal Fluid from Rodent Embryos for use with Dissected Cerebral Cortical Explants. <i>Journal of Visualized Experiments</i> , 2013, , e50333.	0.2	12
223	Homozygous deletions implicate non-coding epigenetic marks in Autism spectrum disorder. <i>Scientific Reports</i> , 2020, 10, 14045.	1.6	12
224	MIPP-Seq: ultra-sensitive rapid detection and validation of low-frequency mosaic mutations. <i>BMC Medical Genomics</i> , 2021, 14, 47.	0.7	12
225	<i>PDCD6IP</i> , encoding a regulator of the ESCRT complex, is mutated in microcephaly. <i>Clinical Genetics</i> , 2020, 98, 80-85.	1.0	11
226	Orgo-Seq integrates single-cell and bulk transcriptomic data to identify cell type specific-driver genes associated with autism spectrum disorder. <i>Nature Communications</i> , 2022, 13, .	5.8	11
227	Peter Huttenlocher (1931–2013). <i>Nature</i> , 2013, 502, 172-172.	13.7	10
228	The polymicrogyria-associated GPR56 promoter preferentially drives gene expression in developing GABAergic neurons in common marmosets. <i>Scientific Reports</i> , 2020, 10, 21516.	1.6	10
229	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. <i>Molecular Psychiatry</i> , 2021, 26, 1706-1718.	4.1	10
230	A novel 2q37 microdeletion containing human neural progenitors genes including <i>STK25</i> results in severe developmental delay, epilepsy, and microcephaly. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2808-2816.	0.7	9
231	Application of single cell genomics to focal epilepsies: A call to action. <i>Brain Pathology</i> , 2021, 31, e12958.	2.1	8
232	Somatic Mosaicism and Neurological Diseases. , 2016, , 179-199.		7
233	Biallelic loss-of-function variants in WDR11 are associated with microcephaly and intellectual disability. <i>European Journal of Human Genetics</i> , 2021, 29, 1663-1668.	1.4	7
234	LISsen up!. <i>Nature Genetics</i> , 1998, 19, 307-308.	9.4	6

#	ARTICLE	IF	CITATIONS
235	Making a Notch in the Evolution of the Human Cortex. <i>Developmental Cell</i> , 2018, 45, 548-550.	3.1	6
236	Somatic copy number variants in neuropsychiatric disorders. <i>Current Opinion in Genetics and Development</i> , 2021, 68, 9-17.	1.5	6
237	Variants in ADD1 cause intellectual disability, corpus callosum dysgenesis, and ventriculomegaly in humans. <i>Genetics in Medicine</i> , 2022, 24, 319-331.	1.1	6
238	Mapping of the mouse <i>hyh</i> gene to a YAC/BAC contig on proximal Chromosome 7. <i>Mammalian Genome</i> , 2002, 13, 239-244.	1.0	5
239	Studies of the candidate genes in X-linked congenital cerebellar hypoplasia. <i>Journal of Neurology</i> , 1999, 246, 1177-1180.	1.8	4
240	A Novel Signaling Mechanism in Brain Development. <i>Pediatric Research</i> , 2004, 56, 309-310.	1.1	4
241	Genetic and neuroradiological heterogeneity of double cortex syndrome. <i>Annals of Neurology</i> , 2000, 47, 265-269.	2.8	4
242	Reelin/Dab1 Signaling in the Developing Cerebral Cortex. , 2008, , 89-105.		4
243	Impaired catabolism of free oligosaccharides due to MAN2C1 variants causes a neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2022, 109, 345-360.	2.6	4
244	Rare genetic causes of lissencephaly may implicate microtubule-based transport in the pathogenesis of cortical dysplasias. <i>Epilepsia</i> , 2010, 51, 67-67.	2.6	3
245	Cover Image, Volume 176A, Number 2, February 2018. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, i.	0.7	3
246	Rainer W. Guillery and the genetic analysis of brain development. <i>European Journal of Neuroscience</i> , 2019, 49, 900-908.	1.2	3
247	Jettison-MS of Nucleic Acid Species. <i>Journal of the American Society for Mass Spectrometry</i> , 2020, 31, 1641-1646.	1.2	2
248	DNA Adductomics by mass tag prelabeling. <i>Rapid Communications in Mass Spectrometry</i> , 2021, 35, e9095.	0.7	2
249	Duplication 2p16 is associated with perisylvian polymicrogyria. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2343-2356.	0.7	1
250	Focal cortical dysplasia. , 2020, , 285-307.		1
251	Clonal mixing, clonal restriction, and specification of cell types in the developing rat olfactory bulb. <i>Journal of Comparative Neurology</i> , 1999, 403, 106-118.	0.9	1
252	The Genetic Basis of Human Cerebral Cortical Malformations. , 2006, , 1073-1079.		1

#	ARTICLE	IF	CITATIONS
253	Cell Fate and Cell Migration in the Developing Cerebral Cortex. , 1999, , 529-547.		1
254	Congenital disorders of cerebral cortical development. , 2002, , 177-194.		0
255	Response to "The Role of Cytomegalovirus in Schizencephaly" by Spalice et al.. American Journal of Medical Genetics, Part A, 2011, 155, 1769-1769.	0.7	0
256	Genetic Disorders of Cerebral Cortical Development. , 2013, , 1-26.		0
257	Reply. Annals of Neurology, 2014, 75, 326-326.	2.8	0
258	Impaired Viability, Platelet Survival, Morphology and Function in Mice Lacking Filamin A.. Blood, 2006, 108, 391-391.	0.6	0
259	Microcephalies and DNA Repair. , 2008, , 109-120.		0