William B Dobyns

List of Publications by Citations

398 papers	33,137 citations	98 h-index	169 g-index
418 ext. papers	37,819 ext. citations	8.9 avg, IF	6.76 L-index

#	Paper	IF	Citations
398	Autosomal dominant cerebellar ataxia (SCA6) associated with small polyglutamine expansions in the alpha 1A-voltage-dependent calcium channel. *Nature Genetics*, **1997**, 15, 62-9	36.3	1440
397	Isolation of a Miller-Dieker lissencephaly gene containing G protein beta-subunit-like repeats. *Nature*, **1993**, 364, 717-21	50.4	906
396	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
395	Mutations in filamin 1 prevent migration of cerebral cortical neurons in human periventricular heterotopia. *Neuron*, **1998**, 21, 1315-25	13.9	729
394	A developmental and genetic classification for malformations of cortical development: update 2012. *Brain*, **2012**, 135, 1348-69	11.2	676
393	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
392	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, 359-69	36.3	567
391	Mutation of the PAX2 gene in a family with optic nerve colobomas, renal anomalies and vesicoureteral reflux. *Nature Genetics*, **1995**, 9, 358-64	36.3	551
390	A developmental and genetic classification for malformations of cortical development. *Neurology*, **2005**, 65, 1873-87	6.5	549
389	Characterizing the Pattern of Anomalies in Congenital Zika Syndrome for Pediatric Clinicians. *JAMA Pediatrics*, **2017**, 171, 288-295	8.3	545
388	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. *Nature Genetics*, **2012**, 44, 934-40	36.3	521
387	Recurrent 16p11.2 microdeletions in autism. *Human Molecular Genetics*, **2008**, 17, 628-38	5.6	515
386	G protein-coupled receptor-dependent development of human frontal cortex. *Science*, **2004**, 303, 2033-6	33.3	404
385	Diagnostic criteria for Walker-Warburg syndrome. *American Journal of Medical Genetics Part A*, **1989** , 32, 195-210		397
384	Mutations in the Na+/K+ -ATPase alpha3 gene ATP1A3 are associated with rapid-onset dystonia parkinsonism. *Neuron*, **2004**, 43, 169-75	13.9	394
383	14-3-3epsilon is important for neuronal migration by binding to NUDEL: a molecular explanation for Miller-Dieker syndrome. *Nature Genetics*, **2003**, 34, 274-85	36.3	333
382	Lymphatic and other vascular malformative/overgrowth disorders are caused by somatic mutations in PIK3CA. *Journal of Pediatrics*, **2015**, 166, 1048-54.e1-5	3.6	315

381	Malformations of cortical development: clinical features and genetic causes. *Lancet Neurology, The*, **2014**, 13, 710-26	24.1	293
380	Description of 13 Infants Born During October 2015-January 2016 With Congenital Zika Virus Infection Without Microcephaly at Birth - Brazil. *Morbidity and Mortality Weekly Report*, **2016**, 65, 1343-1348	31.7	293
379	Mutations in the cilia gene ARL13B lead to the classical form of Joubert syndrome. *American Journal of Human Genetics*, **2008**, 83, 170-9	11	284
378	PIK3CA-related overgrowth spectrum (PROS): diagnostic and testing eligibility criteria, differential diagnosis, and evaluation. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 287-95	2.5	275
377	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, 157-64	5.6	268
376	Periventricular heterotopia: phenotypic heterogeneity and correlation with Filamin A mutations. *Brain*, **2006**, 129, 1892-906	11.2	261
375	Mutations of ARX are associated with striking pleiotropy and consistent genotype-phenotype correlation. *Human Mutation*, **2004**, 23, 147-59	4.7	259
374	Lissencephaly. *JAMA - Journal of the American Medical Association*, **1993**, 270, 2838	27.4	257
373	A drosophila genetic resource of mutants to study mechanisms underlying human genetic diseases. *Cell*, **2014**, 159, 200-214	56.2	239
372	Novel submicroscopic chromosomal abnormalities detected in autism spectrum disorder. *Biological Psychiatry*, **2008**, 63, 1111-7	7.9	238
371	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
370	Lissencephaly and the molecular basis of neuronal migration. *Human Molecular Genetics*, **2003**, 12 Spec No 1, R89-96	5.6	234
369	Spinocerebellar ataxia type 6: gaze-evoked and vertical nystagmus, Purkinje cell degeneration, and variable age of onset. *Annals of Neurology*, **1997**, 42, 933-50	9.4	233
368	WDR62 is associated with the spindle pole and is mutated in human microcephaly. *Nature Genetics*, **2010**, 42, 1010-4	36.3	229
367	A developmental and genetic classification for midbrain-hindbrain malformations. *Brain*, **2009**, 132, 3199-230	11.2	227
366	Consensus Paper: Cerebellar Development. *Cerebellum*, **2016**, 15, 789-828	4.3	216
365	PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. *Brain*, **2015**, 138, 1613-28	11.2	211
364	SRD5A3 is required for converting polyprenol to dolichol and is mutated in a congenital glycosylation disorder. *Cell*, **2010**, 142, 203-17	56.2	207

363	Mutations of CASK cause an X-linked brain malformation phenotype with microcephaly and hypoplasia of the brainstem and cerebellum. *Nature Genetics*, **2008**, 40, 1065-7	36.3	207
362	The phenotypic spectrum of rapid-onset dystonia-parkinsonism (RDP) and mutations in the ATP1A3 gene. *Brain*, **2007**, 130, 828-35	11.2	204
361	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	11	200
360	Syndromes with lissencephaly. I: Miller-Dieker and Norman-Roberts syndromes and isolated lissencephaly. *American Journal of Medical Genetics Part A*, **1984**, 18, 509-26		197
359	Fetal brain lesions after subcutaneous inoculation of Zika virus in a pregnant nonhuman primate. *Nature Medicine*, **2016**, 22, 1256-1259	50.5	196
358	Abnormal development of the human cerebral cortex: genetics, functional consequences and treatment options. *Trends in Neurosciences*, **2008**, 31, 154-62	13.3	196
357	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		194
356	FOXC1 is required for normal cerebellar development and is a major contributor to chromosome 6p25.3 Dandy-Walker malformation. *Nature Genetics*, **2009**, 41, 1037-42	36.3	191
355	ISPD loss-of-function mutations disrupt dystroglycan O-mannosylation and cause Walker-Warburg syndrome. *Nature Genetics*, **2012**, 44, 575-80	36.3	183
354	Practice parameter: Evaluation of the child with microcephaly (an evidence-based review): report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. *Neurology*, **2009**, 73, 887-97	6.5	182
353	Molar tooth sign of the midbrain-hindbrain junction: occurrence in multiple distinct syndromes. *American Journal of Medical Genetics Part A*, **2004**, 125A, 125-34; discussion 117		182
352	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, 918-30	11	182
351	De novo mutations in the actin genes ACTB and ACTG1 cause Baraitser-Winter syndrome. *Nature Genetics*, **2012**, 44, 440-4, S1-2	36.3	181
350	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	5.8	179
349	Human malformations of the midbrain and hindbrain: review and proposed classification scheme. *Molecular Genetics and Metabolism*, **2003**, 80, 36-53	3.7	179
348	Infantile hydrocephalus: a review of epidemiology, classification and causes. *European Journal of Medical Genetics*, **2014**, 57, 359-68	2.6	178
347	Clinical and imaging heterogeneity of polymicrogyria: a study of 328 patients. *Brain*, **2010**, 133, 1415-27	11.2	177
346	Heterozygous deletion of the linked genes ZIC1 and ZIC4 is involved in Dandy-Walker malformation. *Nature Genetics*, **2004**, 36, 1053-5	36.3	174

345	Mutations of the catalytic subunit of RAB3GAP cause Warburg Micro syndrome. *Nature Genetics*, **2005**, 37, 221-3	36.3	173
344	Association of MTOR Mutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. *JAMA Neurology*, **2016**, 73, 836-845	17.2	166
343	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. *Brain*, **2011**, 134, 143-56	11.2	161
342	Characterization of mutations in the gene doublecortin in patients with double cortex syndrome. *Annals of Neurology*, **1999**, 45, 146-53	9.4	157
341	Megalencephaly-capillary malformation (MCAP) and megalencephaly-polydactyly-polymicrogyria-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, 269-91	2.5	154
340	Targeted loss of Arx results in a developmental epilepsy mouse model and recapitulates the human phenotype in heterozygous females. *Brain*, **2009**, 132, 1563-76	11.2	151
339	A revision of the lissencephaly and Miller-Dieker syndrome critical regions in chromosome 17p13.3. *Human Molecular Genetics*, **1997**, 6, 147-55	5.6	151
338	Malformations of cortical development and epilepsy. *Dialogues in Clinical Neuroscience*, **2008**, 10, 47-62	5.7	144
337	TUBA1A mutations cause wide spectrum lissencephaly (smooth brain) and suggest that multiple neuronal migration pathways converge on alpha tubulins. *Human Molecular Genetics*, **2010**, 19, 2817-27	5.6	142
336	Miller-Dieker syndrome: lissencephaly and monosomy 17p. *Journal of Pediatrics*, **1983**, 102, 552-8	3.6	141
335	Mutations in B3GALNT2 cause congenital muscular dystrophy and hypoglycosylation of □dystroglycan. *American Journal of Human Genetics*, **2013**, 92, 354-65	11	139
334	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, 157-95		134
333	Mutation analysis of the DCX gene and genotype/phenotype correlation in subcortical band heterotopia. *European Journal of Human Genetics*, **2001**, 9, 5-12	5.3	129
332	Subcortical band heterotopia in rare affected males can be caused by missense mutations in DCX (XLIS) or LIS1. *Human Molecular Genetics*, **1999**, 8, 1757-60	5.6	127
331	Expansion of the first PolyA tract of ARX causes infantile spasms and status dystonicus. *Neurology*, **2007**, 69, 427-33	6.5	125
330	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	11	125
329	Loss-of-function mutations in RAB18 cause Warburg micro syndrome. *American Journal of Human Genetics*, **2011**, 88, 499-507	11	123
328	Cerebello□culo□enal syndromes including Arima, Senior-L□en and COACH syndromes: More than just variants of Joubert syndrome. *American Journal of Medical Genetics Part A*, **1999**, 86, 459-469		123

327	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
326	Genetic links between brain development and brain evolution. *Nature Reviews Genetics*, **2005**, 6, 581-90	30.1	121
325	X-linked lissencephaly with abnormal genitalia as a tangential migration disorder causing intractable epilepsy: proposal for a new term, "interneuronopathy". *Journal of Child Neurology*, **2005**, 20, 392-7	2.5	119
324	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		116
323	Lissencephaly with cerebellar hypoplasia (LCH): a heterogeneous group of cortical malformations. *Neuropediatrics*, **2001**, 32, 256-63	1.6	116
322	Overlapping cortical malformations and mutations in TUBB2B and TUBA1A. *Brain*, **2013**, 136, 536-48	11.2	115
321	Genetic and biologic classification of infantile spasms. *Pediatric Neurology*, **2011**, 45, 355-67	2.9	115
320	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, 421-32		114
319	Megalencephaly syndromes and activating mutations in the PI3K-AKT pathway: MPPH and MCAP. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2013**, 163C, 122-30	3.1	113
318	Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. *Brain*, **2012**, 135, 1370-86	11.2	113
317	Polymicrogyria and deletion 22q11.2 syndrome: window to the etiology of a common cortical malformation. *American Journal of Medical Genetics, Part A*, **2006**, 140, 2416-25	2.5	113
316	AHI1 gene mutations cause specific forms of Joubert syndrome-related disorders. *Annals of Neurology*, **2006**, 59, 527-34	9.4	111
315	X-linked lissencephaly with absent corpus callosum and ambiguous genitalia. *American Journal of Medical Genetics Part A*, **1999**, 86, 331-337		111
314	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
313	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
312	Inheritance of most X-linked traits is not dominant or recessive, just X-linked. *American Journal of Medical Genetics Part A*, **2004**, 129A, 136-43		108
311	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, 8102-6	11.5	108
310	COL4A1 mutations cause ocular dysgenesis, neuronal localization defects, and myopathy in mice and Walker-Warburg syndrome in humans. *PLoS Genetics*, **2011**, 7, e1002062	6	104

309	Genotype-phenotype analysis of human frontoparietal polymicrogyria syndromes. *Annals of Neurology*, **2005**, 58, 680-7	9.4	103
308	New chromosomal syndrome: Miller-Dieker syndrome and monosomy 17p13. *Human Genetics*, **1984**, 67, 193-200	6.3	103
307	Mutations in 3 genes (MKS3, CC2D2A and RPGRIP1L) cause COACH syndrome (Joubert syndrome with congenital hepatic fibrosis). *Journal of Medical Genetics*, **2010**, 47, 8-21	5.8	101
306	AHI1 mutations cause both retinal dystrophy and renal cystic disease in Joubert syndrome. *Journal of Medical Genetics*, **2006**, 43, 334-9	5.8	101
305	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. *Nature Genetics*, **2014**, 46, 510-515	36.3	100
304	Clinical and molecular basis of classical lissencephaly: Mutations in the LIS1 gene (PAFAH1B1). *Human Mutation*, **2002**, 19, 4-15	4.7	100
303	Familial cavernous malformations of the central nervous system and retina. *Annals of Neurology*, **1987**, 21, 578-83	9.4	100
302	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, 354-64	11	98
301	PRKDC mutations in a SCID patient with profound neurological abnormalities. *Journal of Clinical Investigation*, **2013**, 123, 2969-80	15.9	98
300	A locus for bilateral perisylvian polymicrogyria maps to Xq28. *American Journal of Human Genetics*, **2002**, 70, 1003-8	11	97
299	Genetic Basis of Brain Malformations. *Molecular Syndromology*, **2016**, 7, 220-233	1.5	95
298	Genotypically defined lissencephalies show distinct pathologies. *Journal of Neuropathology and Experimental Neurology*, **2005**, 64, 847-57	3.1	94
297	Albright hereditary osteodystrophy and del(2) (q37.3) in four unrelated individuals. *American Journal of Medical Genetics Part A*, **1995**, 58, 1-7		91
296	Early-Life Epilepsies and the Emerging Role of Genetic Testing. *JAMA Pediatrics*, **2017**, 171, 863-871	8.3	90
295	Mechanisms of interhemispheric transfer and patterns of cognitive function in acallosal patients of normal intelligence. *Archives of Neurology*, **1992**, 49, 271-7		90
294	-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. *JCI Insight*, **2016**, 1,	9.9	90
293	Genetic and neuroradiological heterogeneity of double cortex syndrome. *Annals of Neurology*, **2000**, 47, 265-269	9.4	89
292	Phenotypic spectrum associated with CASK loss-of-function mutations. *Journal of Medical Genetics*, **2011**, 48, 741-51	5.8	88

291	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, 403-9		88
290	Congenital Zika virus infection as a silent pathology with loss of neurogenic output in the fetal brain. *Nature Medicine*, **2018**, 24, 368-374	50.5	85
289	Neuroimaging findings in macrocephaly-capillary malformation: a longitudinal study of 17 patients. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 2981-3008	2.5	85
288	Subcortical band heterotopia (SBH) in males: clinical, imaging and genetic findings in comparison with females. *Brain*, **2002**, 125, 2507-22	11.2	85
287	Microlissencephaly: a heterogeneous malformation of cortical development. *Neuropediatrics*, **1998**, 29, 113-9	1.6	84
286	Baraitser-Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. *European Journal of Human Genetics*, **2015**, 23, 292-301	5.3	83
285	Ultra-high-field MR imaging in polymicrogyria and epilepsy. *American Journal of Neuroradiology*, **2015**, 36, 309-16	4.4	82
284	Rapid-onset dystonia-parkinsonism: linkage to chromosome 19q13. *Annals of Neurology*, **1999**, 46, 176-82	9.4	82
283	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, 1161-8	2.5	81
282	Clinical nosologic and genetic aspects of Joubert and related syndromes. *Journal of Child Neurology* , **1999**, 14, 660-6; discussion 669-72	2.5	81
281	Clinical spectrum of SIX3-associated mutations in holoprosencephaly: correlation between genotype, phenotype and function. *Journal of Medical Genetics*, **2009**, 46, 389-98	5.8	80
280	De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. *American Journal of Human Genetics*, **2015**, 96, 1009	11	78
279	De novo mutations in the beta-tubulin gene TUBB2A cause simplified gyral patterning and infantile-onset epilepsy. *American Journal of Human Genetics*, **2014**, 94, 634-41	11	77
278	The molecular landscape of ASPM mutations in primary microcephaly. *Journal of Medical Genetics*, **2009**, 46, 249-53	5.8	76
277	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
276	Mutations in LAMB1 cause cobblestone brain malformation without muscular or ocular abnormalities. *American Journal of Human Genetics*, **2013**, 92, 468-74	11	75
275	Flores hominid: new species or microcephalic dwarf?. *The Anatomical Record Part A: Discoveries in Molecular, Cellular, and Evolutionary Biology*, **2006**, 288, 1123-45		75
274	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, 2237-47	2.5	75

273	Novel mutations in ATP1A3 associated with catastrophic early life epilepsy, episodic prolonged apnea, and postnatal microcephaly. *Epilepsia*, **2015**, 56, 422-30	6.4	74
272	Mutations in CENPE define a novel kinetochore-centromeric mechanism for microcephalic primordial dwarfism. *Human Genetics*, **2014**, 133, 1023-39	6.3	71
271	MEF2C Haploinsufficiency features consistent hyperkinesis, variable epilepsy, and has a role in dorsal and ventral neuronal developmental pathways. *Neurogenetics*, **2013**, 14, 99-111	3	69
270	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephaly-capillary malformation syndrome. *Nature Genetics*, **2013**, 45, 556-62	36.3	69
269	Identification of a duplication of Xq28 associated with bilateral periventricular nodular heterotopia. *American Journal of Human Genetics*, **1997**, 61, 379-87	11	69
268	CHMP1A encodes an essential regulator of BMI1-INK4A in cerebellar development. *Nature Genetics*, **2012**, 44, 1260-4	36.3	68
267	Association and mutation analyses of 16p11.2 autism candidate genes. *PLoS ONE*, **2009**, 4, e4582	3.7	68
266	Linkage and physical mapping of X-linked lissencephaly/SBH (XLIS): a gene causing neuronal migration defects in human brain. *Human Molecular Genetics*, **1997**, 6, 555-62	5.6	68
265	Mutations in KATNB1 cause complex cerebral malformations by disrupting asymmetrically dividing neural progenitors. *Neuron*, **2014**, 84, 1226-39	13.9	67
264	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, 1216-20	5.3	66
263	A developmental classification of malformations of the brainstem. *Annals of Neurology*, **2007**, 62, 625-39	9.4	66
262	Lissencephaly: Expanded imaging and clinical classification. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1473-1488	2.5	65
261	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. *Brain*, **2013**, 136, 3378-94	11.2	65
260	Bilateral periventricular nodular heterotopia with mental retardation and frontonasal malformation. *Neurology*, **1998**, 51, 499-503	6.5	65
259	Malformations of cortical development and epilepsy. *Cold Spring Harbor Perspectives in Medicine*, **2015**, 5, a022392	5.4	64
258	Semiquantitative analysis of hypothalamic damage on MRI predicts risk for hypothalamic obesity. *Obesity*, **2015**, 23, 1226-33	8	64
257	Periventricular nodular heterotopia with overlying polymicrogyria. *Brain*, **2005**, 128, 2811-21	11.2	64
256	Mutation of FOXC1 and PITX2 induces cerebral small-vessel disease. *Journal of Clinical Investigation*, **2014**, 124, 4877-81	15.9	64

255	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. *Brain*, **2017**, 140, 2610-2622	11.2	63
254	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, 1238-45	5.3	62
253	The Neurogenetics of Lissencephaly. *Neurologic Clinics*, **1989**, 7, 89-105	4.5	62
252	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. *Genetics in Medicine*, **2018**, 20, 1354-1364	8.1	61
251	Autosomal dominant optic nerve colobomas, vesicoureteral reflux, and renal anomalies. *American Journal of Medical Genetics Part A*, **1995**, 59, 204-8		61
250	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. *American Journal of Human Genetics*, **2016**, 98, 579-587	11	60
249	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. *Human Molecular Genetics*, **2015**, 24, 5313-25	5.6	59
248	Characterisation of mutations of the phosphoinositide-3-kinase regulatory subunit, PIK3R2, in perisylvian polymicrogyria: a next-generation sequencing study. *Lancet Neurology, The*, **2015**, 14, 1182-95	24.1	58
247	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	6	58
246	Pontocerebellar hypoplasia type 6: A British case with PEHO-like features. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2079-84	2.5	58
245	Cellular and clinical impact of haploinsufficiency for genes involved in ATR signaling. *American Journal of Human Genetics*, **2007**, 81, 77-86	11	57
244	Megalencephaly and perisylvian polymicrogyria with postaxial polydactyly and hydrocephalus: a rare brain malformation syndrome associated with mental retardation and seizures. *Neuropediatrics* , **2004**, 35, 353-9	1.6	57
243	Identification of a novel recessive RELN mutation using a homozygous balanced reciprocal translocation. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 939-44	2.5	56
242	Variable phenotype of rapid-onset dystonia-parkinsonism. *Movement Disorders*, **1996**, 11, 151-6	7	56
241	Further comments on the lissencephaly syndromes. *American Journal of Medical Genetics Part A*, **1985**, 22, 197-211		56
240	Mouse models of human PIK3CA-related brain overgrowth have acutely treatable epilepsy. *ELife*, **2015**, 4,	8.9	55
239	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
238	Nonsyndromic mental retardation and cryptogenic epilepsy in women with doublecortin gene mutations. *Annals of Neurology*, **2003**, 54, 30-7	9.4	54

237	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	11	53
236	Deletion 16p13.11 uncovers NDE1 mutations on the non-deleted homolog and extends the spectrum of severe microcephaly to include fetal brain disruption. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1523-30	2.5	53
235	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	11	52
234	The clinical patterns and molecular genetics of lissencephaly and subcortical band heterotopia. *Epilepsia*, **2010**, 51 Suppl 1, 5-9	6.4	52
233	Identification of genomic loci contributing to agenesis of the corpus callosum. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2145-59	2.5	51
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