William B Dobyns

List of Publications by Year in Descending Order

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	NRF1 association with AUTS2-Polycomb mediates specific gene activation in the brain. *Molecular Cell*, **2021**, 81, 4663-4676.e8	17.6	4
397	Lissencephaly: Update on diagnostics and clinical management. *European Journal of Paediatric Neurology*, **2021**, 35, 147-152	3.8	5
396	The spectrum of brain malformations and disruptions in twins. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2690-2718	2.5	9
395	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. *Genetics in Medicine*, **2021**, 23, 1028-1040	8.1	7
394	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. *Brain*, **2021**, 144, 1435-1450	11.2	4
393	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. *American Journal of Human Genetics*, **2021**, 108, 951-961	11	4
392	Spatial and cell type transcriptional landscape of human cerebellar development. *Nature Neuroscience*, **2021**, 24, 1163-1175	25.5	14
391	Proximal variants in CCND2 associated with microcephaly, short stature, and developmental delay: A case series and review of inverse brain growth phenotypes. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2719-2738	2.5	1
390	Defining the phenotypical spectrum associated with variants in. *Journal of Medical Genetics*, **2021**, 58, 33-40	5.8	3
389	Mary Ella Mascia Pierpont: Geneticist, scientist, mentor, friend (1945-2020). *American Journal of Medical Genetics, Part A*, **2021**, 185, 319-323	2.5	
388	Cell-free DNA as a diagnostic analyte for molecular diagnosis of vascular malformations. *Genetics in Medicine*, **2021**, 23, 123-130	8.1	8
387	Biallelic Variants Are Associated With Mild Lissencephaly and Cerebellar Hypoplasia. *Neurology: Genetics*, **2021**, 7, e558	3.8	4
386	A dyadic approach to the delineation of diagnostic entities in clinical genomics. *American Journal of Human Genetics*, **2021**, 108, 8-15	11	19
385	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. *Genetics in Medicine*, **2021**, 23, 881-887	8.1	1
384	Expanding the KIF4A-associated phenotype. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3728-3739	2.5	0
383	A Variant Associated With Myoclonus and Developmental Delay: From Molecular Mechanism to Rescue Pharmacology. *Frontiers in Genetics*, **2021**, 12, 694312	4.5	1
382	The Names of Things: The 2018 Bernard Sachs Lecture. *Pediatric Neurology*, **2021**, 122, 41-49	2.9	

381	Response to Hamosh et´al. *American Journal of Human Genetics*, **2021**, 108, 1809-1810	11	
380	Multidisciplinary interaction and MCD gene discovery. The perspective of the clinical geneticist. *European Journal of Paediatric Neurology*, **2021**, 35, 27-34	3.8	0
379	Acetylsalicylic acid suppression of the PI3K pathway as a novel medical therapy for head and neck lymphatic malformations. *International Journal of Pediatric Otorhinolaryngology*, **2021**, 151, 110869	1.7	1
378	Autosomal dominant TUBB3-related syndrome: Fetal, radiologic, clinical and morphological features. *European Journal of Paediatric Neurology*, **2020**, 26, 46-60	3.8	5
377	Activating variants in PDGFRB result in a spectrum of disorders responsive to imatinib monotherapy. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1576-1591	2.5	11
376	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. *Neuron*, **2020**, 106, 404-420.e8	13.9	49
375	Pathogenic Variants in CEP85L Cause Sporadic and Familial Posterior Predominant Lissencephaly. *Neuron*, **2020**, 106, 237-245.e8	13.9	10
374	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. *European Journal of Human Genetics*, **2020**, 28, 770-782	5.3	13
373	Reply to Hsueh YP et al. *European Journal of Human Genetics*, **2020**, 28, 999	5.3	
372	Immune Evasion Strategies Used by Zika Virus to Infect the Fetal Eye and Brain. *Viral Immunology*, **2020**, 33, 22-37	1.7	6
371	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. *Brain*, **2020**, 143, 55-68	11.2	18
370	Genotype-phenotype correlation at codon 1740 of SETD2. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2037-2048	2.5	4
369	Bilateral polymicrogyria associated with dystonia: A new neurogenetic syndrome?. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2207-2213	2.5	
368	Cobblestone Malformation in LAMA2 Congenital Muscular Dystrophy (MDC1A). *Journal of Neuropathology and Experimental Neurology*, **2020**, 79, 998-1010	3.1	1
367	Recurrent constellations of embryonic malformations re-conceptualized as an overlapping group of disorders with shared pathogenesis. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2646-2661	2.5	9
366	International consensus recommendations on the diagnostic work-up for malformations of cortical development. *Nature Reviews Neurology*, **2020**, 16, 618-635	15	17
365	Biallelic loss of function variants in ATP1A2 cause hydrops fetalis, microcephaly, arthrogryposis and extensive cortical malformations. *European Journal of Medical Genetics*, **2020**, 63, 103624	2.6	8
364	Megalencephaly syndromes associated with mutations of core components of the PI3K-AKT-MTOR pathway: PIK3CA, PIK3R2, AKT3, and MTOR. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2019**, 181, 582-590	3.1	40

363	Redefining the Etiologic Landscape of Cerebellar Malformations. *American Journal of Human Genetics*, **2019**, 105, 606-615	11	40
362	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. *American Journal of Human Genetics*, **2019**, 105, 689-705	11	22
361	Postzygotic inactivating mutations of RHOA cause a mosaic neuroectodermal syndrome. *Nature Genetics*, **2019**, 51, 1438-1441	36.3	18
360	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. *American Journal of Human Genetics*, **2019**, 105, 844-853	11	15
359	Costello syndrome: Clinical phenotype, genotype, and management guidelines. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1725-1744	2.5	34
358	Somatic PDGFRB Activating Variants in Fusiform Cerebral Aneurysms. *American Journal of Human Genetics*, **2019**, 104, 968-976	11	15
357	Heterogeneous clinical phenotypes and cerebral malformations reflected by rotatin cellular dynamics. *Brain*, **2019**, 142, 867-884	11.2	14
356	De Novo Missense Variants in FBXW11 Cause Diverse Developmental Phenotypes Including Brain, Eye, and Digit Anomalies. *American Journal of Human Genetics*, **2019**, 105, 640-657	11	16
355	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. *American Journal of Human Genetics*, **2019**, 105, 413-424	11	19
354	SETD2 related overgrowth syndrome: Presentation of four new patients and review of the literature. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2019**, 181, 509-518	3.1	13
353	Spatiotemporal expansion of primary progenitor zones in the developing human cerebellum. *Science*, **2019**, 366, 454-460	33.3	34
352	Duplication 2p16 is associated with perisylvian polymicrogyria. *American Journal of Medical Genetics, Part A*, **2019**, 179, 2343-2356	2.5	
351	Genotype correlates with clinical severity in PIK3CA-associated lymphatic malformations. *JCI Insight* , **2019**, 4,	9.9	16
350	SLC35A2-CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. *Human Mutation*, **2019**, 40, 908-925	4.7	23
349	Subcortical heterotopic gray matter brain malformations: Classification study of 107 individuals. *Neurology*, **2019**, 93, e1360-e1373	6.5	21
348	Approach to overgrowth syndromes in the genome era. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2019**, 181, 483-490	3.1	7
347	Genetics of Hydrocephalus: Causal and Contributory Factors **2019**, 115-129		0
346	Analysis of 17 genes detects mutations in 81% of 811 patients with lissencephaly. *Genetics in Medicine*, **2018**, 20, 1354-1364	8.1	61

345	Joubert Syndrome **2018**, 151-157		
344	Hemimegalencephaly and Dysplastic Megalencephaly **2018**, 55-61		3
343	Structural malformations of the brain, eye, and pituitary gland in PHACE syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 48-55	2.5	8
342	Homozygous TAF8 mutation in a patient with intellectual disability results in undetectable TAF8 protein, but preserved RNA polymerase II transcription. *Human Molecular Genetics*, **2018**, 27, 2171-2186	5.6	13
341	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
340	De novo mutations in GRIN1 cause extensive bilateral polymicrogyria. *Brain*, **2018**, 141, 698-712	11.2	46
339	Walker-Warburg syndrome and tectocerebellar dysraphia: A novel association caused by a homozygous DAG1 mutation. *European Journal of Paediatric Neurology*, **2018**, 22, 525-531	3.8	9
338	Tubulinopathies continued: refining the phenotypic spectrum associated with variants in TUBG1. *European Journal of Human Genetics*, **2018**, 26, 1132-1142	5.3	15
337	Dandy–Walker Malformation, Mega Cisterna Magna, and Blake's Pouch Cyst **2018**, 141-150		
336	Primary brain calcification: an international study reporting novel variants and associated phenotypes. *European Journal of Human Genetics*, **2018**, 26, 1462-1477	5.3	37
335	The Genetic Landscape of Cerebral Steno-Occlusive Arteriopathy and Stroke in Sickle Cell Anemia. *Journal of Stroke and Cerebrovascular Diseases*, **2018**, 27, 2897-2904	2.8	3
334	Biallelic loss of human CTNNA2, encoding αN-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. *Nature Genetics*, **2018**, 50, 1093-1101	36.3	48
333	PARD3 dysfunction in conjunction with dynamic HIPPO signaling drives cortical enlargement with massive heterotopia. *Genes and Development*, **2018**, 32, 763-780	12.6	31
332	De novo and inherited private variants in MAP1B in periventricular nodular heterotopia. *PLoS Genetics*, **2018**, 14, e1007281	6	27
331	Why West? Comparisons of clinical, genetic and molecular features of infants with and without spasms. *PLoS ONE*, **2018**, 13, e0193599	3.7	18
330	Molecular Genetics and Vascular Anomalies **2018**, 21-24		
329	Rhombencephalosynapsis: Fused cerebellum, confused geneticists. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 432-439	3.1	20
328	An update on oculocerebrocutaneous (Delleman-Oorthuys) syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2018**, 178, 414-422	3.1	10

327	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. *Neuron*, **2018**, 100, 1354-1368.e5	13.9	20
326	MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. *American Journal of Human Genetics*, **2018**, 103, 1009-1021	11	30
325	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. *American Journal of Human Genetics*, **2018**, 103, 752-768	11	19
324	Autosomal-dominant early-onset spastic paraparesis with brain calcification due to IFIH1 gain-of-function. *Human Mutation*, **2018**, 39, 1076-1080	4.7	4
323	Comparison of brain MRI findings with language and motor function in the dystroglycanopathies. *Neurology*, **2017**, 88, 623-629	6.5	10
322	Biallelic mutations in the 3Œexonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. *Nature Genetics*, **2017**, 49, 457-464	36.3	43
321	Lissencephaly: Expanded imaging and clinical classification. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1473-1488	2.5	65
320	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
319	Overview of Human Brain Malformations **2017**, 179-182		
318	Malformations of Cortical Development **2017**, 218-225		2
317	encephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. *Journal of Medical Genetics*, **2017**, 54, 460-470	5.8	109
316	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. *American Journal of Human Genetics*, **2017**, 101, 552-563	11	25
315	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. *Brain*, **2017**, 140, 2610-2622	11.2	63
314	Human mutations in integrator complex subunits link transcriptome integrity to brain development. *PLoS Genetics*, **2017**, 13, e1006809	6	41
313	Disorders of Brain Size **2017**, 208-217		
312	Delineating SPTAN1 associated phenotypes: from isolated epilepsy to encephalopathy with progressive brain atrophy. *Brain*, **2017**, 140, 2322-2336	11.2	44
311	Early-Life Epilepsies and the Emerging Role of Genetic Testing. *JAMA Pediatrics*, **2017**, 171, 863-871	8.3	90
310	Characterizing the Pattern of Anomalies in Congenital Zika Syndrome for Pediatric Clinicians. *JAMA Pediatrics*, **2017**, 171, 288-295	8.3	545

309	Neuroimaging findings in Mowat-Wilson syndrome: a study of 54 patients. *Genetics in Medicine*, **2017**, 19, 691-700	8.1	28
308	Description of a new oncogenic mechanism for atypical teratoid rhabdoid tumors in patients with ring chromosome 22. *American Journal of Medical Genetics, Part A*, **2017**, 173, 245-249	2.5	4
307	Phenotypic outcomes in Mouse and Human dependent Dandy-Walker cerebellar malformation suggest shared mechanisms. *ELife*, **2017**, 6,	8.9	23
306	Catenin delta-1 (CTNND1) phosphorylation controls the mesenchymal to epithelial transition in astrocytic tumors. *Human Molecular Genetics*, **2016**, 25, 4201-4210	5.6	7
305	Genetic Basis of Brain Malformations. *Molecular Syndromology*, **2016**, 7, 220-233	1.5	95
304	Phenotype Differentiation of FOXG1 and MECP2 Disorders: A New Method for Characterization of Developmental Encephalopathies. *Journal of Pediatrics*, **2016**, 178, 233-240.e10	3.6	16
303	Recurrent de novo BICD2 mutation associated with arthrogryposis multiplex congenita and bilateral perisylvian polymicrogyria. *Neuromuscular Disorders*, **2016**, 26, 744-748	2.9	36
302	Fetal brain lesions after subcutaneous inoculation of Zika virus in a pregnant nonhuman primate. *Nature Medicine*, **2016**, 22, 1256-1259	50.5	196
301	Mutations in CRADD Result in Reduced Caspase-2-Mediated Neuronal Apoptosis and Cause Megalencephaly with a Rare Lissencephaly Variant. *American Journal of Human Genetics*, **2016**, 99, 1117-1129	11	35
300	Anatomical configurations associated with posthemorrhagic hydrocephalus among premature infants with intraventricular hemorrhage. *Neurosurgical Focus*, **2016**, 41, E5	4.2	10
299	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
298	Progress in autism research and postgenomic studies - AuthorsÕreply. *Lancet Neurology, The*, **2016**, 15, 136-137	24.1	
297	Mutations in EXOSC2 are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. *Journal of Medical Genetics*, **2016**, 53, 419-25	5.8	42
296	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. *American Journal of Human Genetics*, **2016**, 98, 579-587	11	60
295	Two Hundred Thirty-Six Children With Developmental Hydrocephalus: Causes and Clinical Consequences. *Journal of Child Neurology*, **2016**, 31, 309-20	2.5	27
294	Consensus Paper: Cerebellar Development. *Cerebellum*, **2016**, 15, 789-828	4.3	216
293	-associated developmental disorders exhibit distinct classes of mutations with variable expression and tissue distribution. *JCI Insight*, **2016**, 1,	9.9	90
292	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	21.7	293

291	Update on the ACTG1-associated Baraitser-Winter cerebrofrontofacial syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2644-51	2.5	21
290	Identification and Characterization of a Novel Constitutional PIK3CA Mutation in a Child Lacking the Typical Segmental Overgrowth of "PIK3CA-Related Overgrowth Spectrum". *Human Mutation*, **2016**, 37, 242-5	4.7	7
289	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
288	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. *Human Mutation*, **2016**, 37, 148-54	4.7	31
287	Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessive WNT1 mutations. *Journal of Medical Genetics*, **2016**, 53, 427-30	5.8	30
286	Weaver Syndrome-Associated EZH2 Protein Variants Show Impaired Histone Methyltransferase Function In Vitro. *Human Mutation*, **2016**, 37, 301-7	4.7	39
285	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
284	Astroglial-Mediated Remodeling of the Interhemispheric Midline Is Required for the Formation of the Corpus Callosum. *Cell Reports*, **2016**, 17, 735-747	10.6	45
283	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
282	Novel mutations in ATP1A3 associated with catastrophic early life epilepsy, episodic prolonged apnea, and postnatal microcephaly. *Epilepsia*, **2015**, 56, 422-30	6.4	74
281	STIL mutation causes autosomal recessive microcephalic lobar holoprosencephaly. *Human Genetics*, **2015**, 134, 45-51	6.3	26
280	De Novo Mutations in SIK1 Cause a Spectrum of Developmental Epilepsies. *American Journal of Human Genetics*, **2015**, 96, 1009	11	78
279	Recognizable cerebellar dysplasia associated with mutations in multiple tubulin genes. *Human Molecular Genetics*, **2015**, 24, 5313-25	5.6	59
278	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
277	De novo mutations in SIK1 cause a spectrum of developmental epilepsies. *American Journal of Human Genetics*, **2015**, 96, 682-90	11	31
276	Progress in autism and related disorders of brain development. *Lancet Neurology, The*, **2015**, 14, 1069-70	24.1	8
275	Malformations of cortical development and epilepsy. *Cold Spring Harbor Perspectives in Medicine*, **2015**, 5, a022392	5.4	64
274	Ultra-high-field MR imaging in polymicrogyria and epilepsy. *American Journal of Neuroradiology*, **2015**, 36, 309-16	4.4	82

273	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
272	ISDN2014_0157: Modeling human PIK3CA-related congenital brain overgrowth and epilepsy in mice. *International Journal of Developmental Neuroscience*, **2015**, 47, 46-46	2.7	1
271	Baraitser-Winter cerebrofrontofacial syndrome: delineation of the spectrum in 42 cases. *European Journal of Human Genetics*, **2015**, 23, 292-301	5.3	83
270	Variability of epilepsy, autism, brachydactyly, and other clinical features in familial and sporadic 2q37.3 deletion. *Journal of Pediatric Neurology*, **2015**, 07, 279-283	0.2	
269	Semiquantitative analysis of hypothalamic damage on MRI predicts risk for hypothalamic obesity. *Obesity*, **2015**, 23, 1226-33	8	64
268	Familial recurrences of FOXG1-related disorder: Evidence for mosaicism. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3096-102	2.5	13
267	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. *ELife*, **2015**, 4, e06602	8.9	49
266	PI3K/AKT pathway mutations cause a spectrum of brain malformations from megalencephaly to focal cortical dysplasia. *Brain*, **2015**, 138, 1613-28	11.2	211
265	Mouse models of human PIK3CA-related brain overgrowth have acutely treatable epilepsy. *ELife*, **2015**, 4,	8.9	55
264	The Developmental Brain Disorders Database (DBDB): a curated neurogenetics knowledge base with clinical and research applications. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1503-11	2.5	17
263	Autosomal recessive mutations in nuclear transport factor KPNA7 are associated with infantile spasms and cerebellar malformation. *European Journal of Human Genetics*, **2014**, 22, 587-93	5.3	19
262	A drosophila genetic resource of mutants to study mechanisms underlying human genetic diseases. *Cell*, **2014**, 159, 200-214	56.2	239
261	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
260	Mutations in CENPE define a novel kinetochore-centromeric mechanism for microcephalic primordial dwarfism. *Human Genetics*, **2014**, 133, 1023-39	6.3	71
259	Infantile hydrocephalus: a review of epidemiology, classification and causes. *European Journal of Medical Genetics*, **2014**, 57, 359-68	2.6	178
258	Malformations of cortical development: clinical features and genetic causes. *Lancet Neurology, The*, **2014**, 13, 710-26	24.1	293
257	AP1S2 is mutated in X-linked Dandy-Walker malformation with intellectual disability, basal ganglia disease and seizures (Pettigrew syndrome). *European Journal of Human Genetics*, **2014**, 22, 363-8	5.3	36
256	Mutations in KATNB1 cause complex cerebral malformations by disrupting asymmetrically dividing neural progenitors. *Neuron*, **2014**, 84, 1226-39	13.9	67

255	Congenital microcephaly and chorioretinopathy due to de novo heterozygous KIF11 mutations: five novel mutations and review of the literature. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2879-86	2.5	25
254	Epilepsy and outcome in FOXG1-related disorders. *Epilepsia*, **2014**, 55, 1292-300	6.4	39
253	Expansion of the TARP syndrome phenotype associated with de novo mutations and mosaicism. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 120-8	2.5	22
252	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
251	Mutation of FOXC1 and PITX2 induces cerebral small-vessel disease. *Journal of Clinical Investigation* , **2014**, 124, 4877-81	15.9	64
250	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
249	CDKL5 and ARX mutations in males with early-onset epilepsy. *Pediatric Neurology*, **2013**, 48, 367-77	2.9	40
248	Mutations in LAMB1 cause cobblestone brain malformation without muscular or ocular abnormalities. *American Journal of Human Genetics*, **2013**, 92, 468-74	11	75
247	The duplication 17p13.3 phenotype: analysis of 21 families delineates developmental, behavioral and brain abnormalities, and rare variant phenotypes. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1833-52	2.5	42
246	Neuropathology of brain and spinal malformations in a case of monosomy 1p36. *Acta Neuropathologica Communications*, **2013**, 1, 45	7.3	19
245	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
244	Cerebellar and posterior fossa malformations in patients with autism-associated chromosome 22q13 terminal deletion. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 131-6	2.5	50
243	Four new patients with Gomez-Lopez-Hernandez syndrome and proposed diagnostic criteria. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 320-6	2.5	22
242	Novel mutations including deletions of the entire OFD1 gene in 30 families with type 1 orofaciodigital syndrome: a study of the extensive clinical variability. *Human Mutation*, **2013**, 34, 237-47	4.7	35
241	Mutations in B3GALNT2 cause congenital muscular dystrophy and hypoglycosylation of □dystroglycan. *American Journal of Human Genetics*, **2013**, 92, 354-65	11	139
240	X Chromosome-Inactivation Patterns in 31 Individuals with PHACE Syndrome. *Molecular Syndromology*, **2013**, 4, 114-8	1.5	15
239	Expanding the differential diagnosis of fetal hydrops: an unusual prenatal presentation of megalencephaly-capillary malformation syndrome. *Prenatal Diagnosis*, **2013**, 33, 1010-2	3.2	8
238	Periventricular heterotopia in 6q terminal deletion syndrome: role of the C6orf70 gene. *Brain*, **2013** , 136, 3378-94	11.2	65

237	Overlapping cortical malformations and mutations in TUBB2B and TUBA1A. *Brain*, **2013**, 136, 536-48	11.2	115
236	Copy number variation analysis in 98 individuals with PHACE syndrome. *Journal of Investigative Dermatology*, **2013**, 133, 677-684	4.3	20
235	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
234	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
233	The "megalencephaly-capillary malformation" (MCAP) syndrome: the nomenclature of a highly recognizable multiple congenital anomaly syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2115-6	2.5	10
232	Expanding the SHOC2 mutation associated phenotype of Noonan syndrome with loose anagen hair: structural brain anomalies and myelofibrosis. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2420-30	2.5	20
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