Karine Poirier

List of Publications by Year
in descending order

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57 papers	4,901 citations	136950 32 h-index	133252 59 g-index
60 all docs	60 docs citations	60 times ranked	5476 citing authors

#	Article	IF	Citations
1	Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647.	21.4	399
2	Mutations in Î±-Tubulin Cause Abnormal Neuronal Migration in Mice and Lissencephaly in Humans. Cell, 2007, 128, 45-57.	28.9	397
3	Mutations in the Î²-tubulin gene TUBB2B result in asymmetrical polymicrogyria. Nature Genetics, 2009, 41, 746-752.	21.4	330
4	Sporadic Infantile Epileptic Encephalopathy Caused by Mutations in PCDH19 Resembles Dravet Syndrome but Mainly Affects Females. PLoS Genetics, 2009, 5, e1000381.	3.5	304
5	The wide spectrum of tubulinopathies: what are the key features for the diagnosis?. Brain, 2014, 137, 1676-1700.	7.6	252
6	ARX, a novel Prd-class-homeobox gene highly expressed in the telencephalon, is mutated in X-linked mental retardation. Human Molecular Genetics, 2002, 11, 981-991.	2.9	248
7	Mutations in the neuronal Î²-tubulin subunit TUBB3 result in malformation of cortical development and neuronal migration defects. Human Molecular Genetics, 2010, 19, 4462-4473.	2.9	231
8	Large spectrum of lissencephaly and pachygyria phenotypes resulting from de novo missense mutations in tubulin alpha 1A (TUBA1A). Human Mutation, 2007, 28, 1055-1064.	2.5	213
9	Mutations in the Î²-Tubulin Gene TUBB5 Cause Microcephaly with Structural Brain Abnormalities. Cell Reports, 2012, 2, 1554-1562.	6.4	162
10	Mutations in Eml1 lead to ectopic progenitors and neuronal heterotopia in mouse and human. Nature Neuroscience, 2014, 17, 923-933.	14.8	137
11	GPR56-related bilateral frontoparietal polymicrogyria: further evidence for an overlap with the cobblestone complex. Brain, 2010, 133, 3194-3209.	7.6	125
12	Mutations and deletions in PCDH19 account for various familial or isolated epilepsies in females. Human Mutation, 2011, 32, E1959-E1975.	2.5	109
13	Mutations in tubulin genes are frequent causes of various foetal malformations of cortical development including microlissencephaly. Acta Neuropathologica Communications, 2014, 2, 69.	5.2	106
14	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208.	2.5	103
15	Refinement of cortical dysgeneses spectrum associated with TUBA1A mutations. Journal of Medical Genetics, 2008, 45, 647-653.	3.2	103
16	The role of ARX in cortical development. European Journal of Neuroscience, 2006, 23, 869-876.	2.6	101
17	Mutations in the HECT domain of NEDD4L lead to AKTâ€“mTOR pathway deregulation and cause periventricular nodular heterotopia. Nature Genetics, 2016, 48, 1349-1358.	21.4	101
18	New insights into genotypeâ€“phenotype correlations for the doublecortin-related lissencephaly spectrum. Brain, 2013, 136, 223-244.	7.6	99

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19	Neuropathological phenotype of a distinct form of lissencephaly associated with mutations in TUBA1A. Brain, 2008, 131, 2304-2320.	7.6	98
20	Role of the phosphoinositide phosphatase FIG4 gene in familial epilepsy with polymicrogyria. Neurology, 2014, 82, 1068-1075.	1.1	97
21	In vitro follicular growth affects oocyte imprinting establishment in mice. European Journal of Human Genetics, 2003, 11, 493-496.	2.8	91
22	Neuroanatomical distribution of ARX in brain and its localisation in GABAergic neurons. Molecular Brain Research, 2004, 122, 35-46.	2.3	85
23	Mutations in the AP1S2 gene encoding the sigma 2 subunit of the adaptor protein 1 complex are associated with syndromic X-linked mental retardation with hydrocephalus and calcifications in basal ganglia. Journal of Medical Genetics, 2007, 44, 739-744.	3.2	65
24	Expanding the spectrum of TUBA1A-related cortical dysgenesis to Polymicrogyria. European Journal of Human Genetics, 2013, 21, 381-385.	2.8	63
25	Detection of exonic copy-number changes using a highly efficient oligonucleotide-based comparative genomic hybridization-array method. Human Mutation, 2008, 29, 1083-1090.	2.5	51
26	Human lissencephaly with cerebellar hypoplasia due to mutations in TUBA1A: expansion of the foetal neuropathological phenotype. Acta Neuropathologica, 2010, 119, 779-789.	7.7	49
27	A novel mutation in the DLG3 gene encoding the synapse-associated protein 102 (SAP102) causes non-syndromic mental retardation. Neurogenetics, 2010, 11, 251-255.	1.4	49
28	Beta tubulin isoforms are not interchangeable for rescuing impaired radial migration due to Tubb3 knockdown. Human Molecular Genetics, 2014, 23, 1516-1526.	2.9	46
29	Doublecortin interacts with the ubiquitin protease DFFRX, which associates with microtubules in neuronal processes. Molecular and Cellular Neurosciences, 2005, 28, 153-164.	2.2	43
30	CSNK2B splice site mutations in patients cause intellectual disability with or without myoclonic epilepsy. Human Mutation, 2017, 38, 932-941.	2.5	41
31	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	2.5	37
32	Screening of ARX in mental retardation families: consequences for the strategy of molecular diagnosis. Neurogenetics, 2006, 7, 39-46.	1.4	36
33	Combination of infantile spasms, non-epileptic seizures and complex movement disorder: A new case of ARX-related epilepsy. Epilepsy Research, 2008, 80, 224-228.	1.6	33
34	Mosaic parental germline mutations causing recurrent forms of malformations of cortical development. European Journal of Human Genetics, 2016, 24, 611-614.	2.8	33
35	Novel IL1RAPL1 mutations associated with intellectual disability impair synaptogenesis. Human Molecular Genetics, 2015, 24, 1106-1118.	2.9	31
36	Homozygous truncating mutation of the KBP gene, encoding a KIF1B-binding protein, in a familial case of fetal polymicrogyria. Neurogenetics, 2013, 14, 215-224.	1.4	30

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37	TheARX mutations: A frequent cause of X-linked mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 727-732.	1.2	29
38	Loss of parental-specific methylation at theIGF2locus in human hepatocellular carcinoma. Journal of Pathology, 2003, 201, 473-479.	4.5	28
39	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. Brain, 2017, 140, 2597-2609.	7.6	28
40	Rare ACTG1 variants in fetal microlissencephaly. European Journal of Medical Genetics, 2015, 58, 416-418.	1.3	26
41	Loss of Function of KCNC1 is associated with intellectual disability without seizures. European Journal of Human Genetics, 2017, 25, 560-564.	2.8	26
42	Mutation screening of ASMT, the last enzyme of the melatonin pathway, in a large sample of patients with Intellectual Disability. BMC Medical Genetics, 2011, 12, 17.	2.1	25
43	Mutations in the novel gene <scp>FOPV</scp> are associated with familial autosomal dominant and nonâ€familial obliterative portal venopathy. Liver International, 2018, 38, 358-364.	3.9	25
44	ARX polyalanine expansions are highly implicated in familial cases of mental retardation with infantile epilepsy and/or hand dystonia. American Journal of Medical Genetics, Part A, 2011, 155, 98-105.	1.2	23
45	De novo TUBB2B mutation causes fetal akinesia deformation sequence with microlissencephaly: An unusual presentation of tubulinopathy. European Journal of Medical Genetics, 2016, 59, 249-256.	1.3	23
46	ARX mutation in a boy with transsphenoidal encephalocele and hypopituitarism. Clinical Genetics, 2004, 65, 503-505.	2.0	22
47	Maternal mosaicism for mutations in the ARX gene in a family with X linked mental retardation. Human Genetics, 2005, 118, 45-48.	3.8	20
48	Epileptic encephalopathy in a girl with an interstitial deletion of Xp22 comprising promoter and exon 1 of the CDKL5 gene. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 202-207.	1.7	18
49	Mosaic DCX deletion causes subcortical band heterotopia in males. Neurogenetics, 2012, 13, 367-373.	1.4	17
50	Neuropathological Hallmarks of Brain Malformations in Extreme Phenotypes Related to DYNC1H1 Mutations. Journal of Neuropathology and Experimental Neurology, 2017, 76, 195-205.	1.7	15
51	Genotype-phenotype associations for ARX gene duplication in X-linked mental retardation. Neurology, 2006, 67, 2073-2075.	1.1	14
52	The c.429_452 duplication of the ARX gene: a unique developmental-model of limb kinetic apraxia. Orphanet Journal of Rare Diseases, 2014, 9, 25.	2.7	12
53	Mutations in exon 1 of MECP2B are not a common cause of X-linked mental retardation in males. European Journal of Human Genetics, 2005, 13, 523-524.	2.8	11
54	Autism spectrum disorder recurrence, resulting of germline mosaicism for a <scp>CHD2</scp> gene missense variant. Clinical Genetics, 2017, 92, 669-670.	2.0	11

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55	Focal polymicrogyria are associated with submicroscopic chromosomal rearrangements detected by CGH microarray analysis. European Journal of Medical Genetics, 2012, 55, 527-530.	1.3	8
56	Novel role of the synaptic scaffold protein Dlgap4 in ventricular surface integrity and neuronal migration during cortical development. Nature Communications, 2022, 13, 2746.	12.8	4
57	Five novel frameshift mutations in exon 3 and 4 of the MECP2 gene identified in Rett patients: Consequences for the molecular diagnosis strategy. Human Mutation, 2001, 18, 251-252.	2.5	2