# Karine Poirier

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6189540/publications.pdf

Version: 2024-02-01

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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 | 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 |
| 2 | 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 |
| 3 | Loss of Function of KCNC1 is associated with intellectual disability without seizures. European Journal of Human Genetics, 2017, 25, 560-564. | 2.8 | 26 |
| 4 | CSNK2B splice site mutations in patients cause intellectual disability with or without myoclonic epilepsy. Human Mutation, 2017, 38, 932-941. | 2.5 | 41 |
| 5 | 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 |
| 6 | 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 |
| 7 | 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 |
| 8 | 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 |
| 9 | 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 |
| 10 | Mosaic parental germline mutations causing recurrent forms of malformations of cortical development. European Journal of Human Genetics, 2016, 24, 611-614. | 2.8 | 33 |
| 11 | Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117. | 2.5 | 37 |
| 12 | Rare ACTG1 variants in fetal microlissencephaly. European Journal of Medical Genetics, 2015, 58, 416-418. | 1.3 | 26 |
| 13 | Novel IL1RAPL1 mutations associated with intellectual disability impair synaptogenesis. Human Molecular Genetics, 2015, 24, 1106-1118. | 2.9 | 31 |
| 14 | Role of the phosphoinositide phosphatase FIG4 gene in familial epilepsy with polymicrogyria. Neurology, 2014, 82, 1068-1075. | 1.1 | 97 |
| 15 | 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 |
| 16 | The wide spectrum of tubulinopathies: what are the key features for the diagnosis?. Brain, 2014, 137, 1676-1700. | 7.6 | 252 |
| 17 | Mutations in Eml1 lead to ectopic progenitors and neuronal heterotopia in mouse and human. Nature Neuroscience, 2014, 17, 923-933. | 14.8 | 137 |
| 18 | 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 |

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|---|---|---|---|
| 19 | 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 |
| 20 | Mutations in TUBG1, DYNC1H1, KIF5C and KIF2A cause malformations of cortical development and microcephaly. Nature Genetics, 2013, 45, 639-647. | 21.4 | 399 |
| 21 | 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 |
| 22 | Expanding the spectrum of TUBA1A-related cortical dysgenesis to Polymicrogyria. European Journal of Human Genetics, 2013, 21, 381-385. | 2.8 | 63 |
| 23 | New insights into genotypeâ€“phenotype correlations for the doublecortin-related lissencephaly spectrum. Brain, 2013, 136, 223-244. | 7.6 | 99 |
| 24 | 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 |
| 25 | Mosaic DCX deletion causes subcortical band heterotopia in males. Neurogenetics, 2012, 13, 367-373. | 1.4 | 17 |
| 26 | Mutations in the Î²-Tubulin Gene TUBB5 Cause Microcephaly with Structural Brain Abnormalities. Cell Reports, 2012, 2, 1554-1562. | 6.4 | 162 |
| 27 | 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 |
| 28 | 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 |
| 29 | Mutations and deletions in PCDH19 account for various familial or isolated epilepsies in females. Human Mutation, 2011, 32, E1959-E1975. | 2.5 | 109 |
| 30 | 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 |
| 31 | 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 |
| 32 | 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 |
| 33 | 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 |
| 34 | GPR56-related bilateral frontoparietal polymicrogyria: further evidence for an overlap with the cobblestone complex. Brain, 2010, 133, 3194-3209. | 7.6 | 125 |
| 35 | Sporadic Infantile Epileptic Encephalopathy Caused by Mutations in PCDH19 Resembles Dravet Syndrome but Mainly Affects Females. PLoS Genetics, 2009, 5, e1000381. | 3.5 | 304 |
| 36 | Mutations in the Î²-tubulin gene TUBB2B result in asymmetrical polymicrogyria. Nature Genetics, 2009, 41, 746-752. | 21.4 | 330 |

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|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | Refinement of cortical dysgeneses spectrum associated with TUBA1A mutations. Journal of Medical Genetics, 2008, 45, 647-653. | 3.2 | 103 |
| 40 | Neuropathological phenotype of a distinct form of lissencephaly associated with mutations in TUBA1A. Brain, 2008, 131, 2304-2320. | 7.6 | 98 |
| 41 | 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 |
| 42 | Mutations in Î±-Tubulin Cause Abnormal Neuronal Migration in Mice and Lissencephaly in Humans. Cell, 2007, 128, 45-57. | 28.9 | 397 |
| 43 | 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 |
| 44 | Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208. | 2.5 | 103 |
| 45 | The role of ARX in cortical development. European Journal of Neuroscience, 2006, 23, 869-876. | 2.6 | 101 |
| 46 | Screening of ARX in mental retardation families: consequences for the strategy of molecular diagnosis. Neurogenetics, 2006, 7, 39-46. | 1.4 | 36 |
| 47 | TheARX mutations: A frequent cause of X-linked mental retardation. American Journal of Medical Genetics, Part A, 2006, 140A, 727-732. | 1.2 | 29 |
| 48 | Genotype-phenotype associations for ARX gene duplication in X-linked mental retardation. Neurology, 2006, 67, 2073-2075. | 1.1 | 14 |
| 49 | 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 |
| 50 | 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 |
| 51 | 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 |
| 52 | ARX mutation in a boy with transsphenoidal encephalocele and hypopituitarism. Clinical Genetics, 2004, 65, 503-505. | 2.0 | 22 |
| 53 | Neuroanatomical distribution of ARX in brain and its localisation in GABAergic neurons. Molecular Brain Research, 2004, 122, 35-46. | 2.3 | 85 |
| 54 | Loss of parental-specific methylation at theIGF2locus in human hepatocellular carcinoma. Journal of Pathology, 2003, 201, 473-479. | 4.5 | 28 |

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|---|---|---|---|
| 55 | In vitro follicular growth affects oocyte imprinting establishment in mice. European Journal of Human Genetics, 2003, 11, 493-496. | 2.8 | 91 |
| 56 | 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 |
| 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 |