Sandrine Passemard

List of Publications by Year
in descending order

46 papers	2,052 citations	279798 23 h-index	243625 44 g-index
48 all docs	48 docs citations	48 times ranked	4564 citing authors

#	Article	IF	Citations
1	WDR62 is associated with the spindle pole and is mutated in human microcephaly. Nature Genetics, 2010, 42, 1010-1014.	21.4	255
2	Many roads lead to primary autosomal recessive microcephaly. Progress in Neurobiology, 2010, 90, 363-383.	5.7	181
3	Embryonic depletion of serotonin affects cortical development. European Journal of Neuroscience, 2007, 26, 331-344.	2.6	138
4	Neuroprotective Effects of Dexmedetomidine against Glutamate Agonist-induced Neuronal Cell Death Are Related to Increased Astrocyte Brain-derived Neurotrophic Factor Expression. Anesthesiology, 2013, 118, 1123-1132.	2.5	130
5	Loss of Î±1Î²1 Soluble Guanylate Cyclase, the Major Nitric Oxide Receptor, Leads to Moyamoya and Achalasia. American Journal of Human Genetics, 2014, 94, 385-394.	6.2	95
6	Microcephaly. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 111, 129-141.	1.8	94
7	ZIKA virus elicits P53 activation and genotoxic stress in human neural progenitors similar to mutations involved in severe forms of genetic microcephaly and p53. Cell Death and Disease, 2016, 7, e2440-e2440.	6.3	88
8	Haploinsufficiency ofSOX5at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. Human Mutation, 2012, 33, 728-740.	2.5	85
9	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	3.8	66
10	ARCN1 Mutations Cause a Recognizable Craniofacial Syndrome Due to COPI-Mediated Transport Defects. American Journal of Human Genetics, 2016, 99, 451-459.	6.2	65
11	Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. European Journal of Human Genetics, 2015, 23, 1482-1487.	2.8	62
12	Mutations in TUBGCP4 Alter Microtubule Organization via the Î³-Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. American Journal of Human Genetics, 2015, 96, 666-674.	6.2	60
13	Mutations in Citron Kinase Cause Recessive Microlissencephaly with Multinucleated Neurons. American Journal of Human Genetics, 2016, 99, 511-520.	6.2	59
14	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
15	Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly. Genetics in Medicine, 2019, 21, 2043-2058.	2.4	57
16	Autosomal recessive primary microcephaly due to ASPM mutations: An update. Human Mutation, 2018, 39, 319-332.	2.5	53
17	Further delineation of the 17p13.3 microdeletion involving YWHAE but distal to PAFAH1B1: Four additional patients. European Journal of Medical Genetics, 2010, 53, 303-308.	1.3	44
18	Golgi trafficking defects in postnatal microcephaly: The evidence for â€œGolgipathiesâ€• Progress in Neurobiology, 2017, 153, 46-63.	5.7	38

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19	Golgipathies in Neurodevelopment: A New View of Old Defects. Developmental Neuroscience, 2018, 40, 396-416.	2.0	35
20	VIP-induced Neuroprotection of the Developing Brain. Current Pharmaceutical Design, 2011, 17, 1036-1039.	1.9	32
21	Abnormal spindle-like microcephaly-associated (ASPM) mutations strongly disrupt neocortical structure but spare the hippocampus and long-term memory. Cortex, 2016, 74, 158-176.	2.4	32
22	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. Molecular Autism, 2015, 6, 19.	4.9	29
23	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
24	Dymeclin deficiency causes postnatal microcephaly, hypomyelination and reticulum-to-Golgi trafficking defects in mice and humans. Human Molecular Genetics, 2015, 24, 2771-2783.	2.9	25
25	Endoplasmic reticulum and Golgi stress in microcephaly. Cell Stress, 2019, 3, 369-384.	3.2	22
26	Craniosynostosis: A rare complication of pycnodysostosis. European Journal of Medical Genetics, 2010, 53, 89-92.	1.3	21
27	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	2.4	21
28	Genomic imbalances detected by array-CGH in patients with syndromal ocular developmental anomalies. European Journal of Human Genetics, 2012, 20, 527-533.	2.8	19
29	Digenic inheritance of human primary microcephaly delineates centrosomal and nonâ€centrosomal pathways. Human Mutation, 2020, 41, 512-524.	2.5	19
30	Molecular and clinical delineation of 2p15p16.1 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2081-2087.	1.2	18
31	CDK5RAP2 primary microcephaly is associated with hypothalamic, retinal and cochlear developmental defects. Journal of Medical Genetics, 2020, 57, 389-399.	3.2	17
32	Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. Journal of Clinical Immunology, 2021, 41, 603-609.	3.8	16
33	How can cobalamin injections be spaced in long-term therapy for inborn errors of vitamin B12 absorption?. Molecular Genetics and Metabolism, 2012, 107, 66-71.	1.1	15
34	VPS51 biallelic variants cause microcephaly with brain malformations: A confirmatory report. European Journal of Medical Genetics, 2019, 62, 103704.	1.3	15
35	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	3.2	13
36	A case of <scp>L</scp>ennoxâ€<scp>G</scp>astaut syndrome in a patient with <scp>FOXG</scp>1â€related disorder. Epilepsia, 2014, 55, e116-9.	5.1	11

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37	Phosphoglycerate dehydrogenase (PHGDH) deficiency without epilepsy mimicking primary microcephaly. American Journal of Medical Genetics, Part A, 2017, 173, 1936-1942.	1.2	8
38	Phenotypes and genotypes in nonâ€consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	1.2	6
39	Autosomal recessive primary microcephalies (MCPH). European Journal of Paediatric Neurology, 2009, 13, 458.	1.6	4
40	Angelman syndrome and isovaleric acidemia: What is the link?. Molecular Genetics and Metabolism Reports, 2015, 3, 36-38.	1.1	4
41	A familial syndromal form of omphalocele. European Journal of Medical Genetics, 2011, 54, 337-340.	1.3	3
42	Neurological outcome in WDR62 primary microcephaly. Developmental Medicine and Child Neurology, 2022, 64, 509-517.	2.1	3
43	Covid-19 crisis impact on the next generation of physicians: a survey of 800 medical students. BMC Medical Education, 2021, 21, 529.	2.4	3
44	A new lysosomal storage disorder resembling Morquio syndrome in sibs. European Journal of Medical Genetics, 2012, 55, 157-162.	1.3	2
45	A de novo 17q21.2 duplication in a boy with developmental delay and dysmorphic features. European Journal of Medical Genetics, 2013, 56, 226-228.	1.3	1
46	Mcph1, Mutated in Primary Microcephaly, Is Required to Complete Terminal Erythroid Differentiation. Blood, 2019, 134, 1192-1192.	1.4	0