

# Sandrine Passemard

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Neurological outcome in WDR62 primary microcephaly. <i>Developmental Medicine and Child Neurology</i> , 2022, 64, 509-517.	2.1	3
2	<i>De novo</i> coding variants in the <i>AGO1</i> gene cause a neurodevelopmental disorder with intellectual disability. <i>Journal of Medical Genetics</i> , 2022, 59, 965-975.	3.2	13
3	Differential Expression of Interferon-Alpha Protein Provides Clues to Tissue Specificity Across Type I Interferonopathies. <i>Journal of Clinical Immunology</i> , 2021, 41, 603-609.	3.8	16
4	Phenotypes and genotypes in non- consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1768.	1.2	6
5	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2021, 23, 2150-2159.	2.4	21
6	Covid-19 crisis impact on the next generation of physicians: a survey of 800 medical students. <i>BMC Medical Education</i> , 2021, 21, 529.	2.4	3
7	Digenic inheritance of human primary microcephaly delineates centrosomal and non-centrosomal pathways. <i>Human Mutation</i> , 2020, 41, 512-524.	2.5	19
8	<i>CDK5RAP2</i> primary microcephaly is associated with hypothalamic, retinal and cochlear developmental defects. <i>Journal of Medical Genetics</i> , 2020, 57, 389-399.	3.2	17
9	VPS51 biallelic variants cause microcephaly with brain malformations: A confirmatory report. <i>European Journal of Medical Genetics</i> , 2019, 62, 103704.	1.3	15
10	Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly. <i>Genetics in Medicine</i> , 2019, 21, 2043-2058.	2.4	57
11	Endoplasmic reticulum and Golgi stress in microcephaly. <i>Cell Stress</i> , 2019, 3, 369-384.	3.2	22
12	Mcp1, Mutated in Primary Microcephaly, Is Required to Complete Terminal Erythroid Differentiation. <i>Blood</i> , 2019, 134, 1192-1192.	1.4	0
13	Autosomal recessive primary microcephaly due to <i>ASPM</i> mutations: An update. <i>Human Mutation</i> , 2018, 39, 319-332.	2.5	53
14	Golgiopathies in Neurodevelopment: A New View of Old Defects. <i>Developmental Neuroscience</i> , 2018, 40, 396-416.	2.0	35
15	Phosphoglycerate dehydrogenase (PHGDH) deficiency without epilepsy mimicking primary microcephaly. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1936-1942.	1.2	8
16	Molecular and clinical delineation of 2p15p16.1 microdeletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2081-2087.	1.2	18
17	Golgi trafficking defects in postnatal microcephaly: The evidence for "Golgiopathies". <i>Progress in Neurobiology</i> , 2017, 153, 46-63.	5.7	38
18	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. <i>Human Genetics</i> , 2017, 136, 463-479.	3.8	66

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19	WDR81 mutations cause extreme microcephaly and impair mitotic progression in human fibroblasts and Drosophila neural stem cells. <i>Brain</i> , 2017, 140, 2597-2609.	7.6	28
20	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. <i>Npj Genomic Medicine</i> , 2017, 2, 32.	3.8	58
21	ARC1 Mutations Cause a Recognizable Craniofacial Syndrome Due to COPI-Mediated Transport Defects. <i>American Journal of Human Genetics</i> , 2016, 99, 451-459.	6.2	65
22	Mutations in Citron Kinase Cause Recessive Microlissencephaly with Multinucleated Neurons. <i>American Journal of Human Genetics</i> , 2016, 99, 511-520.	6.2	59
23	ZIKA virus elicits P53 activation and genotoxic stress in human neural progenitors similar to mutations involved in severe forms of genetic microcephaly and p53. <i>Cell Death and Disease</i> , 2016, 7, e2440-e2440.	6.3	88
24	Abnormal spindle-like microcephaly-associated (ASPM) mutations strongly disrupt neocortical structure but spare the hippocampus and long-term memory. <i>Cortex</i> , 2016, 74, 158-176.	2.4	32
25	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. <i>Molecular Autism</i> , 2015, 6, 19.	4.9	29
26	Dymeclin deficiency causes postnatal microcephaly, hypomyelination and reticulum-to-Golgi trafficking defects in mice and humans. <i>Human Molecular Genetics</i> , 2015, 24, 2771-2783.	2.9	25
27	Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. <i>European Journal of Human Genetics</i> , 2015, 23, 1482-1487.	2.8	62
28	Angelman syndrome and isovaleric acidemia: What is the link?. <i>Molecular Genetics and Metabolism Reports</i> , 2015, 3, 36-38.	1.1	4
29	Mutations in TUBGCP4 Alter Microtubule Organization via the $\beta$ -Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. <i>American Journal of Human Genetics</i> , 2015, 96, 666-674.	6.2	60
30	A case of <i>L</i> encephalopathy in a patient with <i>FOXG1</i> related disorder. <i>Epilepsia</i> , 2014, 55, e116-9.	5.1	11
31	Loss of $\beta$ 1 Soluble Guanylate Cyclase, the Major Nitric Oxide Receptor, Leads to Moyamoya and Achromia. <i>American Journal of Human Genetics</i> , 2014, 94, 385-394.	6.2	95
32	Microcephaly. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 111, 129-141.	1.8	94
33	A de novo 17q21.2 duplication in a boy with developmental delay and dysmorphic features. <i>European Journal of Medical Genetics</i> , 2013, 56, 226-228.	1.3	1
34	Neuroprotective Effects of Dexmedetomidine against Glutamate Agonist-induced Neuronal Cell Death Are Related to Increased Astrocyte Brain-derived Neurotrophic Factor Expression. <i>Anesthesiology</i> , 2013, 118, 1123-1132.	2.5	130
35	Genomic imbalances detected by array-CGH in patients with syndromal ocular developmental anomalies. <i>European Journal of Human Genetics</i> , 2012, 20, 527-533.	2.8	19
36	How can cobalamin injections be spaced in long-term therapy for inborn errors of vitamin B12 absorption?. <i>Molecular Genetics and Metabolism</i> , 2012, 107, 66-71.	1.1	15

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37	A new lysosomal storage disorder resembling Morquio syndrome in sibs. <i>European Journal of Medical Genetics</i> , 2012, 55, 157-162.	1.3	2
38	Haploinsufficiency of <i>SOX5</i> at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. <i>Human Mutation</i> , 2012, 33, 728-740.	2.5	85
39	A familial syndromal form of omphalocele. <i>European Journal of Medical Genetics</i> , 2011, 54, 337-340.	1.3	3
40	VIP-induced Neuroprotection of the Developing Brain. <i>Current Pharmaceutical Design</i> , 2011, 17, 1036-1039.	1.9	32
41	WDR62 is associated with the spindle pole and is mutated in human microcephaly. <i>Nature Genetics</i> , 2010, 42, 1010-1014.	21.4	255
42	Craniosynostosis: A rare complication of pycnodysostosis. <i>European Journal of Medical Genetics</i> , 2010, 53, 89-92.	1.3	21
43	Further delineation of the 17p13.3 microdeletion involving YWHAE but distal to PAFAH1B1: Four additional patients. <i>European Journal of Medical Genetics</i> , 2010, 53, 303-308.	1.3	44
44	Many roads lead to primary autosomal recessive microcephaly. <i>Progress in Neurobiology</i> , 2010, 90, 363-383.	5.7	181
45	Autosomal recessive primary microcephalies (MCPH). <i>European Journal of Paediatric Neurology</i> , 2009, 13, 458.	1.6	4
46	Embryonic depletion of serotonin affects cortical development. <i>European Journal of Neuroscience</i> , 2007, 26, 331-344.	2.6	138