Anne-Sophie DenommÃ©-Pichon

List of Publications by Year
in descending order

26	423	933447 10	839539 18
papers	citations	h-index	g-index

28	28	28	1146
all docs	docs citations	times ranked	citing authors

#	ARTICLE	IF	CITATIONS
1	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
2	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	3.8	36
3	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	2.8	34
4	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
5	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. American Journal of Human Genetics, 2021, 108, 951-961.	6.2	26
6	<scp>Nextâ€generation</scp> sequencing approaches and challenges in the diagnosis of developmental anomalies and intellectual disability. Clinical Genetics, 2020, 98, 433-444.	2.0	20
7	A novel mutation in the transmembrane 6 domain of GABBR2 leads to a Rettâ€like phenotype. Annals of Neurology, 2018, 83, 437-439.	5.3	19
8	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
9	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. Journal of Clinical Investigation, 2021, 131, .	8.2	16
10	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. Human Genetics, 2022, 141, 65-80.	3.8	14
11	Accelerated genome sequencing with controlled costs for infants in intensive care units: a feasibility study in a French hospital network. European Journal of Human Genetics, 2022, 30, 567-576.	2.8	12
12	Exome reanalysis and proteomic profiling identified TRIP4 as a novel cause of cerebellar hypoplasia and spinal muscular atrophy (PCH1). European Journal of Human Genetics, 2021, 29, 1348-1353.	2.8	10
13	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
14	High efficiency and clinical relevance of exome sequencing in the daily practice of neurogenetics. Journal of Medical Genetics, 2022, 59, 445-452.	3.2	6
15	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	6.2	6
16	Interest of exome sequencing trioâ€like strategy based on pooled parental DNA for diagnosis and translational research in rare diseases. Molecular Genetics & Genomic Medicine, 2021, 9, e1836.	1.2	5
17	SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance. Human Molecular Genetics, 2022, 31, 3325-3340.	2.9	5
18	Primrose syndrome: a phenotypic comparison of patients with a ZBTB20 missense variant versus a 3q13.31 microdeletion including ZBTB20. European Journal of Human Genetics, 2020, 28, 1044-1055.	2.8	4

#	Article	IF	Citations
19	Solving unsolved rare neurological diseasesâ€"a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	2.8	4
20	Oâ€™Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum. Journal of Medical Genetics, 2021, , jmedgenet-2020-107470.	3.2	4
21	Understanding the new <scp>BRD4</scp>â€related syndrome: Clinical and genomic delineation with an international cohort study. Clinical Genetics, 2022, 102, 117-122.	2.0	3
22	The diagnostic rate of inherited metabolic disorders by exome sequencing in a cohort of 547 individuals with developmental disorders. Molecular Genetics and Metabolism Reports, 2021, 29, 100812.	1.1	2
23	Genome-wide variant calling in reanalysis of exome sequencing data uncovered a pathogenic TUBB3 variant. European Journal of Medical Genetics, 2022, 65, 104402.	1.3	2
24	Congenital hypothyroidism and hearing loss without inner ear malformation: Think <scp>TPO</scp>. Clinical Genetics, 2021, 99, 604-606.	2.0	1
25	Toward clinical and molecular dissection of frontonasal dysplasia with facial skin polyps: From Pai syndrome to differential diagnosis through a series of 27 patients. American Journal of Medical Genetics, Part A, 2022, 188, 2036-2047.	1.2	1
26	CutePeaks: A modern viewer for Sanger trace file. Journal of Open Source Software, 2021, 6, 3457.	4.6	0