Vincent Cantagrel

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4875697/publications.pdf

Version: 2024-02-01

31 papers	2,220 citations	331670 21 h-index	454955 30 g-index
34 all docs	34 docs citations	34 times ranked	4786 citing authors

#	Article	IF	Citations
1	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	6.2	352
2	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	28.9	253
3	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	28.9	228
4	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
5	Mutations in the Glycosylphosphatidylinositol Gene PIGL Cause CHIME Syndrome. American Journal of Human Genetics, 2012, 90, 685-688.	6.2	114
6	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	21.4	111
7	Faulty Initiation of Proteoglycan Synthesis Causes Cardiac and Joint Defects. American Journal of Human Genetics, 2011, 89, 15-27.	6.2	108
8	Mutations in LAMB1 Cause Cobblestone Brain Malformation without Muscular or Ocular Abnormalities. American Journal of Human Genetics, 2013, 92, 468-474.	6.2	96
9	AMPD2 Regulates GTP Synthesis and Is Mutated in a Potentially Treatable Neurodegenerative Brainstem Disorder. Cell, 2013, 154, 505-517.	28.9	94
10	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. Brain, 2010, 133, 3210-3220.	7.6	87
11	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013.	7.6	67
12	From glycosylation disorders to dolichol biosynthesis defects: a new class of metabolic diseases. Journal of Inherited Metabolic Disease, 2011, 34, 859-867.	3.6	65
13	Disruption of a new X linked gene highly expressed in brain in a family with two mentally retarded males. Journal of Medical Genetics, 2004, 41, 736-742.	3.2	60
14	Opposite Modulation of RAC1 by Mutations in TRIO Is Associated with Distinct, Domain-Specific Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 338-355.	6.2	58
15	Identification of a novel ARL13B variant in a Joubert syndrome-affected patient with retinal impairment and obesity. European Journal of Human Genetics, 2015, 23, 621-627.	2.8	48
16	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
17	High N-glycan multiplicity is critical for neuronal adhesion and sensitizes the developing cerebellum to N-glycosylation defect. ELife, 2018, 7, .	6.0	43
18	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	2.5	34

#	Article	IF	Citations
19	Truncation ofNHEJ1 in a patient with polymicrogyria. Human Mutation, 2007, 28, 356-364.	2.5	33
20	Utility of whole exome sequencing for the early diagnosis of pediatric-onset cerebellar atrophy associated with developmental delay in an inbred population. Orphanet Journal of Rare Diseases, 2016, 11, 57.	2.7	31
21	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
22	MINPP1 prevents intracellular accumulation of the chelator inositol hexakisphosphate and is mutated in Pontocerebellar Hypoplasia. Nature Communications, 2020, 11, 6087.	12.8	28
23	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
24	Inherited glycosylphosphatidylinositol defects cause the rare Emm-negative blood phenotype and developmental disorders. Blood, 2021, 137, 3660-3669.	1.4	18
25	Spatiotemporal expression in mouse brain of Kiaa2022, a gene disrupted in two patients with severe mental retardation. Gene Expression Patterns, 2009, 9, 423-429.	0.8	17
26	Recurrent KIF5C mutation leading to frontal pachygyria without microcephaly. Neurogenetics, 2016, 17, 79-82.	1.4	17
27	Recessive PRDM13 mutations cause fatal perinatal brainstem dysfunction with cerebellar hypoplasia and disrupt Purkinje cell differentiation. American Journal of Human Genetics, 2022, 109, 909-927.	6.2	10
28	Normal glycosylation screening does not rule out SRD5A3-CDG. European Journal of Human Genetics, 2011, 19, 1019-1019.	2.8	7
29	16p13.11p11.2 triplication syndrome: a new recognizable genomic disorder characterized by optical genome mapping and whole genome sequencing. European Journal of Human Genetics, 2022, , .	2.8	5
30	Suleiman-El-Hattab syndrome: a histone modification disorder caused by TASP1 deficiency. Human Molecular Genetics, 2022, 31, 3083-3094.	2.9	3
31	Biallelic loss of <scp> EMC10 </scp> leads to mild to severe intellectual disability. Annals of Clinical and Translational Neurology, 0, , .	3.7	1