Cyril Mignot

List of Publications by Year
in descending order

52 papers	2,778 citations	257450 24 h-index	189892 50 g-index
57 all docs	57 docs citations	57 times ranked	6518 citing authors

#	Article	IF	Citations
1	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. Nature Genetics, 2014, 46, 503-509.	21.4	490
2	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
3	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	6.2	179
4	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	6.2	171
5	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	3.2	135
6	STXBP1-related encephalopathy presenting as infantile spasms and generalized tremor in three patients. Epilepsia, 2011, 52, 1820-1827.	5.1	90
7	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	2.4	90
8	Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with PMM2-CDG (phosphomannomutase 2-congenital disorder of glycosylation) and review of the literature. Journal of Medical Genetics, 2017, 54, 843-851.	3.2	88
9	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
10	Using medical exome sequencing to identify the causes of neurodevelopmental disorders: Experience of 2 clinical units and 216 patients. Clinical Genetics, 2018, 93, 567-576.	2.0	82
11	Relapsing encephalopathy with cerebellar ataxia related to an <scp>ATP</scp>1A3 mutation. Developmental Medicine and Child Neurology, 2015, 57, 1183-1186.	2.1	78
12	The Number of Genomic Copies at the 16p11.2 Locus Modulates Language, Verbal Memory, and Inhibition. Biological Psychiatry, 2016, 80, 129-139.	1.3	78
13	WWOX-related encephalopathies: delineation of the phenotypical spectrum and emerging genotype-phenotype correlation. Journal of Medical Genetics, 2015, 52, 61-70.	3.2	74
14	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
15	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
16	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
17	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	2.4	47
18	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. Journal of Medical Genetics, 2019, 56, 526-535.	3.2	46

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19	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
20	Increased diagnostic yield in complex dystonia through exome sequencing. Parkinsonism and Related Disorders, 2020, 74, 50-56.	2.2	34
21	Relapsing encephalopathy with cerebellar ataxia are caused by variants involving p.Arg756 in ATP1A3. European Journal of Paediatric Neurology, 2019, 23, 448-455.	1.6	33
22	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
23	Reverse Phenotyping in Patients with Skin Capillary Malformations and Mosaic GNAQ or GNA11 Mutations Defines a Clinical Spectrum with Genotype-Phenotype Correlation. Journal of Investigative Dermatology, 2020, 140, 1106-1110.e2.	0.7	30
24	Clinical study of 19 patients with <scp>SCN</scp>8Aâ€related epilepsy: Two modes of onset regarding <scp>EEG</scp> and seizures. Epilepsia, 2019, 60, 845-856.	5.1	28
25	ARID1Bmutations are the major genetic cause of corpus callosum anomalies in patients with intellectual disability. Brain, 2016, 139, e64-e64.	7.6	26
26	Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability. Journal of Pediatrics, 2017, 185, 160-166.e1.	1.8	25
27	NR4A2 haploinsufficiency is associated with intellectual disability and autism spectrum disorder. Clinical Genetics, 2018, 94, 264-268.	2.0	22
28	Early-onset encephalopathy with paroxysmal movement disorders and epileptic seizures without hemiplegic attacks: About three children with novel ATP1A3 mutations. Brain and Development, 2018, 40, 768-774.	1.1	21
29	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	3.8	18
30	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. PLoS Genetics, 2018, 14, e1007671.	3.5	16
31	Monoallelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 739-748.	6.2	15
32	PAK3 mutations responsible for severe intellectual disability and callosal agenesis inhibit cell migration. Neurobiology of Disease, 2020, 136, 104709.	4.4	14
33	Clinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities. Genetics in Medicine, 2021, 23, 1484-1491.	2.4	14
34	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
35	Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. Orphanet Journal of Rare Diseases, 2016, 11, 52.	2.7	13
36	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	5.1	13

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37	Implication of folate deficiency in CYP2U1 loss of function. Journal of Experimental Medicine, 2021, 218, .	8.5	13
38	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. Brain, 2019, 142, e15-e15.	7.6	12
39	Deciphering the natural history of SCA7 in children. European Journal of Neurology, 2020, 27, 2267-2276.	3.3	12
40	Clinical and neuroimaging findings in 33 patients with <scp>MCAP</scp> syndrome: A survey to evaluate relevant endpoints for future clinical trials. Clinical Genetics, 2021, 99, 650-661.	2.0	12
41	Expanding the clinical and EEG spectrum of CNKSR2-related encephalopathy with status epilepticus during slow sleep (ESES). Clinical Neurophysiology, 2020, 131, 1030-1039.	1.5	11
42	Three novel patients with epileptic encephalopathy due to biallelic mutations in the <scp>PLCB1</scp> gene. Clinical Genetics, 2020, 97, 477-482.	2.0	9
43	Deep phenotyping unstructured data mining in an extensive pediatric database to unravel a common KCNA2 variant in neurodevelopmental syndromes. Genetics in Medicine, 2021, 23, 968-971.	2.4	9
44	Semaphorin-Plexin Signaling: From Axonal Guidance to a New X-Linked Intellectual Disability Syndrome. Pediatric Neurology, 2022, 126, 65-73.	2.1	8
45	Pseudoxanthoma elasticum overlaps hereditary spastic paraplegia type 56. Journal of Internal Medicine, 2021, 289, 709-725.	6.0	7
46	GM3 synthase deficiency in non-Amish patients. Genetics in Medicine, 2022, 24, 492-498.	2.4	7
47	SATB2-associated syndrome: characterization of skeletal features and of bone fragility in a prospective cohort of 19 patients. Orphanet Journal of Rare Diseases, 2022, 17, 100.	2.7	7
48	SCN1A-related epilepsy with recessive inheritance: Two further families. European Journal of Paediatric Neurology, 2021, 33, 121-124.	1.6	4
49	Adaptive behavior and psychiatric comorbidities in KCNB1 encephalopathy. Epilepsy and Behavior, 2022, 126, 108471.	1.7	3
50	The EPIGENE network: A French initiative to harmonize and improve the nationwide diagnosis of monogenic epilepsies. European Journal of Medical Genetics, 2022, 65, 104445.	1.3	3
51	Demyelinating motor neuropathy associated with a homozygous <scp>GPT2</scp> pathogenic variant. Muscle and Nerve, 2021, 63, E41-E44.	2.2	2
52	Pulmonary Hemorrhage Revealing Multiple Vascular Malformations in a Child with KCNT1 Developmental Epileptic Encephalopathy. Journal of Pediatrics, 2021, 237, 311-312.	1.8	0