Agathe Roubertie

List of Publications by Year
in descending order

37	1,402	471509 17	330143 37
papers	citations	h-index	g-index

47	47	47	1660
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Disentangling molecular and clinical stratification patterns in beta-galactosidase deficiency. Journal of Medical Genetics, 2022, 59, 377-384.	3.2	2
2	Molecular and clinical descriptions of patients with <scp>GABA_A</scp> receptor gene variants (<scp>GABRA1</scp>, <scp>GABRB2</scp>, <scp>GABRB3</scp>, <scp>GABRG2</scp>): A cohort study, review of literature, and genotypeâ€“phenotype correlation. Epilepsia, 2022, 63, 2519-2533.	5.1	23
3	Pathogenic variants in IMPG1 cause autosomal dominant and autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2021, 58, 570-578.	3.2	10
4	Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154.	1.6	13
5	Late-onset riboflavin transporter deficiency: a treatable mimic of various motor neuropathy aetiologies. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 27-35.	1.9	11
6	Optic neuropathy linked to ACAD9 pathogenic variants: A potentially riboflavin-responsive disorder?. Mitochondrion, 2021, 59, 169-174.	3.4	3
7	Gene therapy in the putamen for curing AADC deficiency and Parkinson's disease. EMBO Molecular Medicine, 2021, 13, e14712.	6.9	17
8	Cognitive impairment in children with <scp>CACNA</scp>1A mutations. Developmental Medicine and Child Neurology, 2020, 62, 330-337.	2.1	31
9	Clinical disease progression and biomarkers in Niemannâ€“Pick disease type C: a prospective cohort study. Orphanet Journal of Rare Diseases, 2020, 15, 328.	2.7	12
10	Pregnancy in MNGIE: a clinical and metabolic honeymoon. Annals of Clinical and Translational Neurology, 2020, 7, 2484-2488.	3.7	0
11	Deciphering the natural history of SCA7 in children. European Journal of Neurology, 2020, 27, 2267-2276.	3.3	12
12	FGF14â€related episodic ataxia: delineating the phenotype of Episodic Ataxia type 9. Annals of Clinical and Translational Neurology, 2020, 7, 565-572.	3.7	29
13	Hereditary spastic paraplegia and prominent sensorial involvement: think MAG mutations!. Annals of Clinical and Translational Neurology, 2019, 6, 1572-1577.	3.7	6
14	Erythrocyte Encapsulated Thymidine Phosphorylase for the Treatment of Patients with Mitochondrial Neurogastrointestinal Encephalomyopathy: Study Protocol for a Multi-Centre, Multiple Dose, Open Label Trial. Journal of Clinical Medicine, 2019, 8, 1096.	2.4	39
15	KCNT1 epilepsy with migrating focal seizures shows a temporal sequence with poor outcome, high mortality and SUDEP. Brain, 2019, 142, 2996-3008.	7.6	35
16	MyoNeuroGastroIntestinal Encephalopathy: Natural History and Means for Early Diagnosis. Gastroenterology, 2019, 156, 1525-1527.e4.	1.3	7
17	Pallidal Deep Brain Stimulation in DYT6 Dystonia: Clinical Outcome and Predictive Factors for Motor Improvement. Journal of Clinical Medicine, 2019, 8, 2163.	2.4	25
18	Quantitative analysis and <scp>EEG</scp> markers of <scp>KCNT</scp>1 epilepsy of infancy with migrating focal seizures. Epilepsia, 2019, 60, 20-32.	5.1	13

#	Article	IF	Citations
19	Maladies des ticsÂ: tics provisoires, tics chroniques et syndrome de Gilles de la Tourette. Pratique Neurologique - FMC, 2018, 9, 244-251.	0.1	0
20	Benign paroxysmal torticollis, benign paroxysmal vertigo, and benign tonic upward gaze are not benign disorders. Developmental Medicine and Child Neurology, 2018, 60, 1256-1263.	2.1	21
21	Role of neuroimaging in the diagnosis of hereditary cerebellar ataxias in childhood. Journal of Neuroradiology, 2016, 43, 176-185.	1.1	6
22	Chronic Diarrhea in l-Amino Acid Decarboxylase (AADC) Deficiency: A Prominent Clinical Finding Among a Series of Ten French Patients. JIMD Reports, 2016, 31, 85-93.	1.5	18
23	Neuroradiological findings expand the phenotype of OPA1-related mitochondrial dysfunction. Journal of the Neurological Sciences, 2015, 349, 154-160.	0.6	17
24	From splitting GLUT1 deficiency syndromes to overlapping phenotypes. European Journal of Medical Genetics, 2015, 58, 443-454.	1.3	52
25	Mutation in NDUFA13/GRIM19 leads to early onset hypotonia, dyskinesia and sensorial deficiencies, and mitochondrial complex I instability. Human Molecular Genetics, 2015, 24, 3948-3955.	2.9	42
26	Acuteâ€onset chorea, dystonia, and cardiac fibroelastoma in a child: A paraneoplastic association?. Movement Disorders, 2013, 28, 250-251.	3.9	4
27	Les vomissements cycliques de lâ€™enfant: le point de vue du neuropÃ©diatre. Archives De Pediatrie, 2012, 19, H186-H187.	1.0	0
28	Treatment for dystonia in childhood. European Journal of Neurology, 2012, 19, 1292-1299.	3.3	56
29	Benign paroxysmal vertigo of childhood: Long-term outcome. Cephalalgia, 2011, 31, 439-443.	3.9	58
30	Mouvements anormaux paroxystiques dans les carences Ã©nergÃ©tiques (PDH, GLUT1). Archives De Pediatrie, 2011, 18, H134-H135.	1.0	0
31	Developmental and benign movement disorders in childhood. Movement Disorders, 2010, 25, 1317-1334.	3.9	80
32	Speech disturbances in patients with dystonia or chorea due to neurometabolic disorders. Movement Disorders, 2010, 25, 1605-1611.	3.9	8
33	Benign paroxysmal tonic upgaze, benign paroxysmal torticollis, episodic ataxia and CACNA1A mutation in a family. Journal of Neurology, 2008, 255, 1600-1602.	3.6	77
34	Partial Epilepsy and 47,XXX Karyotype: Report of Four Cases. Pediatric Neurology, 2006, 35, 69-74.	2.1	16
35	Sepiapterin Reductase Deficiency: Clinical Presentation and Evaluation of Long-Term Therapy. Pediatric Neurology, 2006, 35, 308-313.	2.1	39
36	Treatment of early-onset dystonia: update and a new perspective. Child's Nervous System, 2000, 16, 334-340.	1.1	38

#	Article	IF	Citations
37	Treatment of DYT1-generalised dystonia by stimulation of the internal globus pallidus. Lancet, The, 2000, 355, 2220-2221.	13.7	536