# Agathe Roubertie

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6310186/publications.pdf

Version: 2024-02-01

| 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 | Treatment of DYT1-generalised dystonia by stimulation of the internal globus pallidus. Lancet, The, 2000, 355, 2220-2221. | 13.7 | 536 |
| 2 | Developmental and benign movement disorders in childhood. Movement Disorders, 2010, 25, 1317-1334. | 3.9 | 80 |
| 3 | 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 |
| 4 | Benign paroxysmal vertigo of childhood: Long-term outcome. Cephalalgia, 2011, 31, 439-443. | 3.9 | 58 |
| 5 | Treatment for dystonia in childhood. European Journal of Neurology, 2012, 19, 1292-1299. | 3.3 | 56 |
| 6 | From splitting GLUT1 deficiency syndromes to overlapping phenotypes. European Journal of Medical Genetics, 2015, 58, 443-454. | 1.3 | 52 |
| 7 | 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 |
| 8 | Sepiapterin Reductase Deficiency: Clinical Presentation and Evaluation of Long-Term Therapy. Pediatric Neurology, 2006, 35, 308-313. | 2.1 | 39 |
| 9 | 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 |
| 10 | Treatment of early-onset dystonia: update and a new perspective. Child's Nervous System, 2000, 16, 334-340. | 1.1 | 38 |
| 11 | 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 |
| 12 | Cognitive impairment in children with <scp>CACNA</scp>1A mutations. Developmental Medicine and Child Neurology, 2020, 62, 330-337. | 2.1 | 31 |
| 13 | 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 |
| 14 | 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 |
| 15 | Molecular and clinical descriptions of patients with <scp>GABA<sub>A</sub></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 |
| 16 | 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 |
| 17 | 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 |
| 18 | Neuroradiological findings expand the phenotype of OPA1-related mitochondrial dysfunction. Journal of the Neurological Sciences, 2015, 349, 154-160. | 0.6 | 17 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Gene therapy in the putamen for curing AADC deficiency and Parkinson's disease. EMBO Molecular Medicine, 2021, 13, e14712. | 6.9 | 17 |
| 20 | Partial Epilepsy and 47,XXX Karyotype: Report of Four Cases. Pediatric Neurology, 2006, 35, 69-74. | 2.1 | 16 |
| 21 | 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 |
| 22 | Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154. | 1.6 | 13 |
| 23 | 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 |
| 24 | Deciphering the natural history of SCA7 in children. European Journal of Neurology, 2020, 27, 2267-2276. | 3.3 | 12 |
| 25 | 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 |
| 26 | Pathogenic variants in IMPG1 cause autosomal dominant and autosomal recessive retinitis pigmentosa. Journal of Medical Genetics, 2021, 58, 570-578. | 3.2 | 10 |
| 27 | Speech disturbances in patients with dystonia or chorea due to neurometabolic disorders. Movement Disorders, 2010, 25, 1605-1611. | 3.9 | 8 |
| 28 | MyoNeuroGastroIntestinal Encephalopathy: Natural History and Means for Early Diagnosis. Gastroenterology, 2019, 156, 1525-1527.e4. | 1.3 | 7 |
| 29 | Role of neuroimaging in the diagnosis of hereditary cerebellar ataxias in childhood. Journal of Neuroradiology, 2016, 43, 176-185. | 1.1 | 6 |
| 30 | Hereditary spastic paraplegia and prominent sensorial involvement: think MAG mutations!. Annals of Clinical and Translational Neurology, 2019, 6, 1572-1577. | 3.7 | 6 |
| 31 | Acuteâ€onset chorea, dystonia, and cardiac fibroelastoma in a child: A paraneoplastic association?. Movement Disorders, 2013, 28, 250-251. | 3.9 | 4 |
| 32 | Optic neuropathy linked to ACAD9 pathogenic variants: A potentially riboflavin-responsive disorder?. Mitochondrion, 2021, 59, 169-174. | 3.4 | 3 |
| 33 | Disentangling molecular and clinical stratification patterns in beta-galactosidase deficiency. Journal of Medical Genetics, 2022, 59, 377-384. | 3.2 | 2 |
| 34 | Mouvements anormaux paroxystiques dans les carences Ã©nergÃ©tiques (PDH, GLUT1). Archives De Pediatrie, 2011, 18, H134-H135. | 1.0 | 0 |
| 35 | Les vomissements cycliques de lâ€™enfant: le point de vue du neuropÃ©diatre. Archives De Pediatrie, 2012, 19, H186-H187. | 1.0 | 0 |
| 36 | Maladies des ticsÂ: tics provisoires, tics chroniques et syndrome de Gilles de la Tourette. Pratique Neurologique - FMC, 2018, 9, 244-251. | 0.1 | 0 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Pregnancy in MNGIE: a clinical and metabolic honeymoon. Annals of Clinical and Translational Neurology, 2020, 7, 2484-2488. | 3.7 | 0 |