# MichÃ¨l A A P Willemsen

List of Publications by Year
in descending order



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|---|---|---|---|
| 80 papers | 3,210 citations | 218677 26 h-index | 168389 53 g-index |
| 80 all docs | 80 docs citations | 80 times ranked | 5264 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Brain, 2010, 133, 655-670. | 7.6 | 356 |
| 2 | Tyrosine hydroxylase deficiency: a treatable disorder of brain catecholamine biosynthesis. Brain, 2010, 133, 1810-1822. | 7.6 | 268 |
| 3 | A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063. | 2.4 | 220 |
| 4 | Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237. | 21.4 | 186 |
| 5 | Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. Orphanet Journal of Rare Diseases, 2017, 12, 12. | 2.7 | 172 |
| 6 | Mutations in DDHD2, Encoding an Intracellular Phospholipase A1, Cause a Recessive Form of Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1073-1081. | 6.2 | 159 |
| 7 | Health risks for ataxiaâ€telangiectasia mutated heterozygotes: a systematic review, metaâ€analysis and evidenceâ€based guideline. Clinical Genetics, 2016, 90, 105-117. | 2.0 | 143 |
| 8 | Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group. Epilepsia Open, 2020, 5, 354-365. | 2.4 | 142 |
| 9 | CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257. | 6.2 | 111 |
| 10 | Cerebrospinal Fluid Analysis in the Workup of GLUT1 Deficiency Syndrome. JAMA Neurology, 2013, 70, 1440. | 9.0 | 106 |
| 11 | De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771. | 6.2 | 96 |
| 12 | Ataxia-telangiectasia: Immunodeficiency and survival. Clinical Immunology, 2017, 178, 45-55. | 3.2 | 72 |
| 13 | MR imaging and proton MR spectroscopic studies in SjÃ¶gren-Larsson syndrome: characterization of the leukoencephalopathy. American Journal of Neuroradiology, 2004, 25, 649-57. | 2.4 | 70 |
| 14 | Ataxiaâ€telangiectasia: recommendations for multidisciplinary treatment. Developmental Medicine and Child Neurology, 2017, 59, 680-689. | 2.1 | 61 |
| 15 | A post hoc study on gene panel analysis for the diagnosis of dystonia. Movement Disorders, 2017, 32, 569-575. | 3.9 | 59 |
| 16 | Genotype, extrapyramidal features, and severity of variant ataxiaâ€telangiectasia. Annals of Neurology, 2019, 85, 170-180. | 5.3 | 58 |
| 17 | SjÃ¶grenâ€“Larsson syndrome in clinical practice. Journal of Inherited Metabolic Disease, 2012, 35, 955-962. | 3.6 | 48 |
| 18 | Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548. | 6.2 | 46 |

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|---|---|---|---|
| 19 | The diagnostic pathway in complex paediatric neurology: A cost analysis. European Journal of Paediatric Neurology, 2015, 19, 233-239. | 1.6 | 40 |
| 20 | Absence of Î±- and Î²-dystroglycan is associated with Walker-Warburg syndrome. Neurology, 2015, 84, 2177-2182. | 1.1 | 40 |
| 21 | Lactate and its many faces. European Journal of Paediatric Neurology, 2016, 20, 3-10. | 1.6 | 38 |
| 22 | <scp>l</scp> -Dopa in dystonia. Neurology, 2017, 88, 1865-1871. | 1.1 | 35 |
| 23 | Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595. | 12.8 | 35 |
| 24 | Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. Neuropediatrics, 2015, 46, 098-103. | 0.6 | 34 |
| 25 | Clinical, radiological and possible pathological overlap of cystic leukoencephalopathy without megalencephaly and Aicardi-GoutiÃ¨res syndrome. European Journal of Paediatric Neurology, 2016, 20, 604-610. | 1.6 | 29 |
| 26 | Genotypeâ€“phenotype correlations in ataxia telangiectasia patients with ATM c.3576G>A and c.8147T>C mutations. Journal of Medical Genetics, 2019, 56, 308-316. | 3.2 | 29 |
| 27 | A novel SLC2A1 mutation linking hemiplegic migraine with alternating hemiplegia of childhood. Cephalalgia, 2015, 35, 10-15. | 3.9 | 28 |
| 28 | Serum inflammatory mediators correlate with disease activity in <scp>e</scp>lectrical <scp>s</scp>tatus <scp>e</scp>pilepticus in <scp>s</scp>leep (ESES) syndrome. Epilepsia, 2016, 57, e45-50. | 5.1 | 28 |
| 29 | Disturbed brain ether lipid metabolism and histology in <scp>SjÃ¶grenâ€Larsson</scp> syndrome. Journal of Inherited Metabolic Disease, 2020, 43, 1265-1278. | 3.6 | 25 |
| 30 | Two Greek siblings with sepiapterin reductase deficiency. Molecular Genetics and Metabolism, 2008, 94, 403-409. | 1.1 | 23 |
| 31 | De novo SPAST mutations may cause a complex SPG4 phenotype. Brain, 2019, 142, e31-e31. | 7.6 | 21 |
| 32 | Myoclonus in childhood-onset neurogenetic disorders: The importance of early identification and treatment. European Journal of Paediatric Neurology, 2015, 19, 726-729. | 1.6 | 20 |
| 33 | How to proceed after â€œnegativeâ€•exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. Journal of Inherited Metabolic Disease, 2022, 45, 663-681. | 3.6 | 20 |
| 34 | Functional consequences of the autosomal dominant G272A mutation in the human GLUT1 gene. FEBS Letters, 2001, 498, 104-109. | 2.8 | 19 |
| 35 | Hourly analysis of cerebrospinal fluid glucose shows large diurnal fluctuations. Journal of Cerebral Blood Flow and Metabolism, 2016, 36, 899-902. | 4.3 | 19 |
| 36 | Dilemma of Reporting Incidental Findings in Newborn Screening Programs for SCID: Parentsâ€™ Perspective on Ataxia Telangiectasia. Frontiers in Immunology, 2019, 10, 2438. | 4.8 | 19 |

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|---|---|---|---|
| 37 | Cerebral lipid accumulation in Chanarinâ€“Dorfman Syndrome. Molecular Genetics and Metabolism, 2015, 114, 51-54. | 1.1 | 18 |
| 38 | Nicotinamide Riboside Improves Ataxia Scores and Immunoglobulin Levels in Ataxia Telangiectasia. Movement Disorders, 2021, 36, 2951-2957. | 3.9 | 18 |
| 39 | Multimodal imaging of the macula in hereditary and acquired lack of macular pigment. Acta Ophthalmologica, 2014, 92, 138-142. | 1.1 | 16 |
| 40 | Confirmation of neurometabolic diagnoses using ageâ€dependent cerebrospinal fluid metabolomic profiles. Journal of Inherited Metabolic Disease, 2020, 43, 1112-1120. | 3.6 | 16 |
| 41 | Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774. | 2.8 | 15 |
| 42 | Classic ataxia-telangiectasia: the phenotype of long-term survivors. Journal of Neurology, 2020, 267, 830-837. | 3.6 | 14 |
| 43 | B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotypeâ€“phenotype associations in the muscular dystrophy-dystroglycanopathies. Genome Medicine, 2017, 9, 118. | 8.2 | 13 |
| 44 | Clinical presentation and longâ€term followâ€up of dopamine beta hydroxylase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 554-565. | 3.6 | 13 |
| 45 | Parental quality of life in complex paediatric neurologic disorders of unknown aetiology. European Journal of Paediatric Neurology, 2016, 20, 723-731. | 1.6 | 12 |
| 46 | The Ketogenic Diet and Its Effect on Bone Mineral Density: A Retrospective Observational Cohort Study. Neuropediatrics, 2019, 50, 353-358. | 0.6 | 12 |
| 47 | Variable Interpretation of the Dystonia Consensus Classification Items Compromises Its Solidity. Movement Disorders, 2019, 34, 317-320. | 3.9 | 12 |
| 48 | Neurometabolic disorders. Neurology: Clinical Practice, 2016, 6, 348-357. | 1.6 | 11 |
| 49 | Cytidine Diphosphate-Ribitol Analysis for Diagnostics and Treatment Monitoring of Cytidine Diphosphate-l-Ribitol Pyrophosphorylase A Muscular Dystrophy. Clinical Chemistry, 2019, 65, 1295-1306. | 3.2 | 11 |
| 50 | Early diagnosis of ataxia telangiectasia in the neonatal phase: a parentsâ€™ perspective. European Journal of Pediatrics, 2020, 179, 251-256. | 2.7 | 11 |
| 51 | Autosomal dominant GCH1 mutations causing spastic paraplegia at disease onset. Parkinsonism and Related Disorders, 2020, 74, 12-15. | 2.2 | 11 |
| 52 | A brother and sister with intellectual disability and characteristic neuroimaging findings. European Journal of Paediatric Neurology, 2018, 22, 866-869. | 1.6 | 9 |
| 53 | Occurrence of symptoms in different stages of <scp>Duchenne</scp> muscular dystrophy and their impact on social participation. Muscle and Nerve, 2021, 64, 701-709. | 2.2 | 9 |
| 54 | Dysarthria in children and adults with ataxia telangiectasia. Developmental Medicine and Child Neurology, 2021, 63, 450-456. | 2.1 | 9 |

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|---|---|---|---|
| 55 | Understanding fetal factors that contribute to preterm birth: SjÃ¶gren-Larsson syndrome as a model. Journal of Perinatal Medicine, 2018, 46, 523-529. | 1.4 | 8 |
| 56 | Toward understanding tissueâ€specific symptoms in dolicholâ€phosphateâ€mannose synthesis disorders; insight from DPM3â€CDG. Journal of Inherited Metabolic Disease, 2019, 42, 984-992. | 3.6 | 8 |
| 57 | Trajectories of motor abnormalities in milder phenotypes of ataxia telangiectasia. Neurology, 2019, 92, e19-e29. | 1.1 | 8 |
| 58 | Lactate infusion as therapeutical intervention: a scoping review. European Journal of Pediatrics, 2022, , 1. | 2.7 | 8 |
| 59 | The novel P330L pathogenic variant of aromatic amino acid decarboxylase maps on the catalytic flexible loop underlying its crucial role. Cellular and Molecular Life Sciences, 2022, 79, 305. | 5.4 | 8 |
| 60 | Retinal Morphology in SjÃ¶gren-Larsson Syndrome on OCT: From Metabolic Crystalline Maculopathy to Early-Onset Macular Degeneration. Ophthalmology Retina, 2019, 3, 500-509. | 2.4 | 7 |
| 61 | Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. Movement Disorders, 2021, 36, 690-703. | 3.9 | 7 |
| 62 | The Phenotypic Spectrum of PNKP-Associated Disease and the Absence of Immunodeficiency and Cancer Predisposition in a Dutch Cohort. Pediatric Neurology, 2020, 113, 26-32. | 2.1 | 6 |
| 63 | Biochemical Studies in Fibroblasts to Interpret Variants of Unknown Significance in the ABCD1 Gene. Genes, 2021, 12, 1930. | 2.4 | 6 |
| 64 | Stroke mimics add to the phenotypic spectrum of GLUT1 deficiency syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 668-670. | 1.9 | 5 |
| 65 | Benign nocturnal alternating hemiplegia of childhood: A clinical and nomenclatural reappraisal. European Journal of Paediatric Neurology, 2018, 22, 1110-1117. | 1.6 | 5 |
| 66 | Variable Selection in Untargeted Metabolomics and the Danger of Sparsity. Metabolites, 2020, 10, 470. | 2.9 | 5 |
| 67 | BMI-z scores of boys with Duchenne muscular dystrophy already begin to increase before losing ambulation: a longitudinal exploration of BMI, corticosteroids and caloric intake. Neuromuscular Disorders, 2022, 32, 236-244. | 0.6 | 4 |
| 68 | Macular fibrosis complicating macular pigment deficient maculopathy in SjÃ¶grenâ€Larsson syndrome. Acta Ophthalmologica, 2016, 94, e663-e664. | 1.1 | 3 |
| 69 | Copy number variations as potential diagnostic and prognostic markers for CNS melanocytic neoplasms in neurocutaneous melanosis. Acta Neuropathologica, 2017, 133, 333-335. | 7.7 | 3 |
| 70 | Blood, urine and cerebrospinal fluid analysis in TH and AADC deficiency and the effect of treatment. Molecular Genetics and Metabolism Reports, 2021, 27, 100762. | 1.1 | 3 |
| 71 | Congenital eyelid ptosis, decreased glomerular filtration, and orthostatic hypotension: Questions. Pediatric Nephrology, 2017, 32, 1169-1170. | 1.7 | 2 |
| 72 | Teaching NeuroImages: Bilateral Nucleus Tractus Solitarius Lesions in Neurogenic Respiratory Failure. Neurology, 2021, , 10.1212/WNL.0000000000012614. | 1.1 | 2 |

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|---|---|---|---|
| 73 | 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 |
| 74 | Head circumference in glucose transporter 1 deficiency syndrome: Normal for individuals, abnormal as a group. European Journal of Paediatric Neurology, 2022, 38, 73-76. | 1.6 | 2 |
| 75 | Anemia in Glucose Transporter Type 1 Deficiency Syndrome: Often Expected, Rarely Encountered, and with a Fascinating Explanation. Neuropediatrics, 2017, 48, 327-328. | 0.6 | 1 |
| 76 | Hypointensity of the Basal Ganglia in Adults with Glucose Transporter Protein Type 1 Deficiency Syndrome: A Novel Magnetic Resonance Imaging Finding. Annals of Neurology, 2020, 87, 10-11. | 5.3 | 1 |
| 77 | Diagnosis and Management of Ataxia-Telangiectasia in Resource-Limited Settings. Journal of International Child Neurology Association, 2020, 1, . | 0.0 | 1 |
| 78 | Little folks, little myelin, and little teeth. Neurology, 2014, 83, 1884-1885. | 1.1 | 0 |
| 79 | Congenital eyelid ptosis, decreased glomerular filtration, and orthostatic hypotension: Answers. Pediatric Nephrology, 2017, 32, 1171-1174. | 1.7 | 0 |
| 80 | Dystonia in childhood: Rising networks. European Journal of Paediatric Neurology, 2018, 22, 219-220. | 1.6 | 0 |