MichÃ¨l A A P Willemsen

List of Publications by Year
in descending order

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	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
2	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
3	Lactate infusion as therapeutical intervention: a scoping review. European Journal of Pediatrics, 2022, , 1.	2.7	8
4	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
5	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
6	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
7	Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. Movement Disorders, 2021, 36, 690-703.	3.9	7
8	Clinical presentation and longâ€term followâ€up of dopamine beta hydroxylase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 554-565.	3.6	13
9	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
10	Teaching NeuroImages: Bilateral Nucleus Tractus Solitarius Lesions in Neurogenic Respiratory Failure. Neurology, 2021, , 10.1212/WNL.0000000000012614.	1.1	2
11	Nicotinamide Riboside Improves Ataxia Scores and Immunoglobulin Levels in Ataxia Telangiectasia. Movement Disorders, 2021, 36, 2951-2957.	3.9	18
12	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
13	Dysarthria in children and adults with ataxia telangiectasia. Developmental Medicine and Child Neurology, 2021, 63, 450-456.	2.1	9
14	Biochemical Studies in Fibroblasts to Interpret Variants of Unknown Significance in the ABCD1 Gene. Genes, 2021, 12, 1930.	2.4	6
15	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
16	Early diagnosis of ataxia telangiectasia in the neonatal phase: a parentsâ€™ perspective. European Journal of Pediatrics, 2020, 179, 251-256.	2.7	11
17	Classic ataxia-telangiectasia: the phenotype of long-term survivors. Journal of Neurology, 2020, 267, 830-837.	3.6	14
18	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

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19	Variable Selection in Untargeted Metabolomics and the Danger of Sparsity. Metabolites, 2020, 10, 470.	2.9	5
20	Confirmation of neurometabolic diagnoses using ageâ€dependent cerebrospinal fluid metabolomic profiles. Journal of Inherited Metabolic Disease, 2020, 43, 1112-1120.	3.6	16
21	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
22	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
23	Autosomal dominant GCH1 mutations causing spastic paraplegia at disease onset. Parkinsonism and Related Disorders, 2020, 74, 12-15.	2.2	11
24	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
25	Diagnosis and Management of Ataxia-Telangiectasia in Resource-Limited Settings. Journal of International Child Neurology Association, 2020, 1, .	0.0	1
26	Genotype, extrapyramidal features, and severity of variant ataxiaâ€telangiectasia. Annals of Neurology, 2019, 85, 170-180.	5.3	58
27	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
28	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
29	The Ketogenic Diet and Its Effect on Bone Mineral Density: A Retrospective Observational Cohort Study. Neuropediatrics, 2019, 50, 353-358.	0.6	12
30	De novo SPAST mutations may cause a complex SPG4 phenotype. Brain, 2019, 142, e31-e31.	7.6	21
31	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
32	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
33	Variable Interpretation of the Dystonia Consensus Classification Items Compromises Its Solidity. Movement Disorders, 2019, 34, 317-320.	3.9	12
34	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
35	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
36	Trajectories of motor abnormalities in milder phenotypes of ataxia telangiectasia. Neurology, 2019, 92, e19-e29.	1.1	8

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37	Dystonia in childhood: Rising networks. European Journal of Paediatric Neurology, 2018, 22, 219-220.	1.6	0
38	Stroke mimics add to the phenotypic spectrum of GLUT1 deficiency syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 668-670.	1.9	5
39	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
40	A brother and sister with intellectual disability and characteristic neuroimaging findings. European Journal of Paediatric Neurology, 2018, 22, 866-869.	1.6	9
41	Benign nocturnal alternating hemiplegia of childhood: A clinical and nomenclatural reappraisal. European Journal of Paediatric Neurology, 2018, 22, 1110-1117.	1.6	5
42	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
43	Ataxia-telangiectasia: Immunodeficiency and survival. Clinical Immunology, 2017, 178, 45-55.	3.2	72
44	A post hoc study on gene panel analysis for the diagnosis of dystonia. Movement Disorders, 2017, 32, 569-575.	3.9	59
45	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	2.8	15
46	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
47	Ataxiaâ€telangiectasia: recommendations for multidisciplinary treatment. Developmental Medicine and Child Neurology, 2017, 59, 680-689.	2.1	61
48	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	2.4	220
49	<scp>l</scp> -Dopa in dystonia. Neurology, 2017, 88, 1865-1871.	1.1	35
50	Congenital eyelid ptosis, decreased glomerular filtration, and orthostatic hypotension: Questions. Pediatric Nephrology, 2017, 32, 1169-1170.	1.7	2
51	Congenital eyelid ptosis, decreased glomerular filtration, and orthostatic hypotension: Answers. Pediatric Nephrology, 2017, 32, 1171-1174.	1.7	0
52	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
53	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
54	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

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55	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
56	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
57	Hourly analysis of cerebrospinal fluid glucose shows large diurnal fluctuations. Journal of Cerebral Blood Flow and Metabolism, 2016, 36, 899-902.	4.3	19
58	Neurometabolic disorders. Neurology: Clinical Practice, 2016, 6, 348-357.	1.6	11
59	Parental quality of life in complex paediatric neurologic disorders of unknown aetiology. European Journal of Paediatric Neurology, 2016, 20, 723-731.	1.6	12
60	Macular fibrosis complicating macular pigment deficient maculopathy in SjÃ¶grenâ€Larsson syndrome. Acta Ophthalmologica, 2016, 94, e663-e664.	1.1	3
61	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
62	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
63	Lactate and its many faces. European Journal of Paediatric Neurology, 2016, 20, 3-10.	1.6	38
64	The diagnostic pathway in complex paediatric neurology: A cost analysis. European Journal of Paediatric Neurology, 2015, 19, 233-239.	1.6	40
65	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
66	Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. Neuropediatrics, 2015, 46, 098-103.	0.6	34
67	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
68	Absence of Î±- and Î²-dystroglycan is associated with Walker-Warburg syndrome. Neurology, 2015, 84, 2177-2182.	1.1	40
69	A novel SLC2A1 mutation linking hemiplegic migraine with alternating hemiplegia of childhood. Cephalalgia, 2015, 35, 10-15.	3.9	28
70	Cerebral lipid accumulation in Chanarinâ€“Dorfman Syndrome. Molecular Genetics and Metabolism, 2015, 114, 51-54.	1.1	18
71	Multimodal imaging of the macula in hereditary and acquired lack of macular pigment. Acta Ophthalmologica, 2014, 92, 138-142.	1.1	16
72	Little folks, little myelin, and little teeth. Neurology, 2014, 83, 1884-1885.	1.1	0

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73	Cerebrospinal Fluid Analysis in the Workup of GLUT1 Deficiency Syndrome. JAMA Neurology, 2013, 70, 1440.	9.0	106
74	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
75	SjÃ¶grenâ€“Larsson syndrome in clinical practice. Journal of Inherited Metabolic Disease, 2012, 35, 955-962.	3.6	48
76	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Brain, 2010, 133, 655-670.	7.6	356
77	Tyrosine hydroxylase deficiency: a treatable disorder of brain catecholamine biosynthesis. Brain, 2010, 133, 1810-1822.	7.6	268
78	Two Greek siblings with sepiapterin reductase deficiency. Molecular Genetics and Metabolism, 2008, 94, 403-409.	1.1	23
79	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
80	Functional consequences of the autosomal dominant G272A mutation in the human GLUT1 gene. FEBS Letters, 2001, 498, 104-109.	2.8	19