Michel Aap Willemsen

List of Publications by Year
in descending order

25 papers	928 citations	623734 14 h-index	610901 24 g-index
25 all docs	25 docs citations	25 times ranked	1989 citing authors

#	Article	IF	Citations
1	Untargeted metabolomics and infrared ion spectroscopy identify biomarkers for pyridoxine-dependent epilepsy. Journal of Clinical Investigation, 2021, 131, .	8.2	33
2	Nicotinamide Riboside Improves Ataxia Scores and Immunoglobulin Levels in Ataxia Telangiectasia. Movement Disorders, 2021, 36, 2951-2957.	3.9	18
3	A detailed description of the phenotypic spectrum of North Sea Progressive Myoclonus Epilepsy in a large cohort of seventeen patients. Parkinsonism and Related Disorders, 2020, 72, 44-48.	2.2	9
4	Diagnosis and Management of Ataxia-Telangiectasia in Resource-Limited Settings. Journal of International Child Neurology Association, 2020, 1, .	0.0	1
5	Daily Functioning and Quality of Life in Patients with SjÃ¶grenâ€“Larsson Syndrome. Neuropediatrics, 2019, 50, 089-095.	0.6	4
6	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
7	Trajectories of motor abnormalities in milder phenotypes of ataxia telangiectasia. Neurology, 2019, 92, e19-e29.	1.1	8
8	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
9	Ataxia-telangiectasia: Immunodeficiency and survival. Clinical Immunology, 2017, 178, 45-55.	3.2	72
10	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	2.4	220
11	Clinical exome sequencing for cerebellar ataxia and spastic paraplegia uncovers novel geneâ€“disease associations and unanticipated rare disorders. European Journal of Human Genetics, 2016, 24, 1460-1466.	2.8	89
12	Variants inCUL4Bare Associated with Cerebral Malformations. Human Mutation, 2015, 36, 106-117.	2.5	37
13	A novel SLC2A1 mutation linking hemiplegic migraine with alternating hemiplegia of childhood. Cephalalgia, 2015, 35, 10-15.	3.9	28
14	Neuropathology in classical and variant ataxiaâ€telangiectasia. Neuropathology, 2012, 32, 234-244.	1.2	60
15	A de novo p.Asp18Asn mutation in TREX1 in a patient with Aicardiâ€“GoutiÃ¨res syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2612-2617.	1.2	35
16	Patients with SjÃ¶gren-Larsson Syndrome Lack Macular Pigment. Ophthalmology, 2010, 117, 966-971.	5.2	50
17	Ataxiaâ€Telangiectasia and mechanical ventilation: A word of caution. Pediatric Pulmonology, 2009, 44, 101-102.	2.0	14
18	Subclinical Changes in the Juvenile Crystalline Macular Dystrophy in SjÃ¶grenâ€“Larsson Syndrome Detected by Optical Coherence Tomography. Ophthalmology, 2008, 115, 870-875.	5.2	45

#	Article	IF	Citations
19	Mutations in the aldh7a1 gene cause pyridoxine-dependent seizures. Arquivos De Neuro-Psiquiatria, 2008, 66, 288-289.	0.8	3
20	Defective metabolism of Leukotriene B4 in the SjÃ¶grenâ€“Larsson Syndrome. Journal of the Neurological Sciences, 2001, 183, 61-67.	0.6	59
21	Hypoglycorrhachia: A simple clue, simply missed. Annals of Neurology, 2001, 49, 685-686.	5.3	11
22	Hypoglycorrhachia: A simple clue, simply missed. Annals of Neurology, 2001, 49, 685-686.	5.3	1
23	Juvenile macular dystrophy associated with deficient activity of fatty aldehyde dehydrogenase in SjÃ¶gren-Larsson syndrome. American Journal of Ophthalmology, 2000, 130, 782-789.	3.3	66
24	Defective inactivation of leukotriene B4 in patients with SjoÂÌˆgren-Larsson syndrome. Journal of Pediatrics, 2000, 136, 258-260.	1.8	39
25	A Novel 4 bp Deletion Mutation in the FALDH Gene Segregating in a Turkish Family with SjÃ¶grenâ€“Larsson Syndrome. Journal of Investigative Dermatology, 1999, 112, 827-828.	0.7	11