Bernhard Weschke

List of Publications by Year
in descending order

31	3,223	361296 20	414303 32
papers	citations	h-index	g-index

32	32	32	4380
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-GoutiÃ¨res syndrome and mimic congenital viral brain infection. Nature Genetics, 2006, 38, 910-916.	9.4	592
2	Mutations in antiquitin in individuals with pyridoxine-dependent seizures. Nature Medicine, 2006, 12, 307-309.	15.2	476
3	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
4	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Brain, 2010, 133, 655-670.	3.7	356
5	Tyrosine hydroxylase deficiency: a treatable disorder of brain catecholamine biosynthesis. Brain, 2010, 133, 1810-1822.	3.7	268
6	Mutations in the Gene Encoding Gap Junction Protein Î±12 (Connexin 46.6) Cause Pelizaeus-Merzbacherâ€“Like Disease. American Journal of Human Genetics, 2004, 75, 251-260.	2.6	257
7	Prevention of Epilepsy in Infants with Tuberous Sclerosis Complex in the <scp>EPISTOP</scp> Trial. Annals of Neurology, 2021, 89, 304-314.	2.8	137
8	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. Brain, 2008, 131, 747-759.	3.7	134
9	Long-term Outcome of Allogeneic Hematopoietic Stem Cell Transplantation in Patients With Juvenile Metachromatic Leukodystrophy Compared With Nontransplanted Control Patients. JAMA Neurology, 2016, 73, 1133.	4.5	94
10	Bi-allelic Mutations in EPRS, Encoding the Glutamyl-Prolyl-Aminoacyl-tRNA Synthetase, Cause a Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2018, 102, 676-684.	2.6	58
11	TSC2 pathogenic variants are predictive of severe clinical manifestations in TSC infants: results of the EPISTOP study. Genetics in Medicine, 2020, 22, 1489-1497.	1.1	51
12	GLRB is the third major gene of effect in hyperekplexia. Human Molecular Genetics, 2013, 22, 927-940.	1.4	50
13	Early Clinical Predictors of Autism Spectrum Disorder in Infants with Tuberous Sclerosis Complex: Results from the EPISTOP Study. Journal of Clinical Medicine, 2019, 8, 788.	1.0	42
14	Recurrent Stroke Due to a Novel Voltage Sensor Mutation in Ca _v 2.1 Responds to Verapamil. Stroke, 2011, 42, e14-7.	1.0	39
15	Spectrum of cerebral arteriopathies in children with arterial ischemic stroke. Neurology, 2020, 94, e2479-e2490.	1.5	34
16	Facial phenotype allows diagnosis of Mowat-Wilson syndrome in the absence of hirschsprung disease. American Journal of Medical Genetics Part A, 2004, 124A, 102-104.	2.4	30
17	Potential Risks to Stable Long-term Outcome of Allogeneic Hematopoietic Stem Cell Transplantation for Children With Cerebral X-linked Adrenoleukodystrophy. JAMA Network Open, 2018, 1, e180769.	2.8	30
18	The clinical-phenotype continuum in DYNC1H1-related disordersâ€”genomic profiling and proposal for a novel classification. Journal of Human Genetics, 2020, 65, 1003-1017.	1.1	30

#	Article	IF	Citations
19	BRAT1 mutations are associated with infantile epileptic encephalopathy, mitochondrial dysfunction, and survival into childhood. American Journal of Medical Genetics, Part A, 2016, 170, 2274-2281.	0.7	25
20	Is autism driven by epilepsy in infants with Tuberous Sclerosis Complex?. Annals of Clinical and Translational Neurology, 2020, 7, 1371-1381.	1.7	23
21	Prediction of Neurodevelopment in Infants With Tuberous Sclerosis Complex Using Early EEG Characteristics. Frontiers in Neurology, 2020, 11, 582891.	1.1	19
22	Early epileptiform EEG activity in infants with tuberous sclerosis complex predicts epilepsy and neurodevelopmental outcomes. Epilepsia, 2021, 62, 1208-1219.	2.6	19
23	Anti-NMDA receptor encephalitis presenting as atypical anorexia nervosa: an adolescent case report. European Child and Adolescent Psychiatry, 2015, 24, 1321-1324.	2.8	14
24	31Phosphorus Magnetic Resonance Spectroscopy in Late-Onset Tay-Sachs Disease. Journal of Child Neurology, 2001, 16, 377-380.	0.7	13
25	Acute Disseminated Encephalomyelitis After Human Parechovirus Infection. Pediatric Infectious Disease Journal, 2016, 35, 35-38.	1.1	12
26	A comparison study of anxiety in children undergoing brain MRI vs adults undergoing brain MRI vs children undergoing an electroencephalogram. PLoS ONE, 2019, 14, e0211552.	1.1	12
27	Acute renal failure unmasking Lesch-Nyhan disease in a patient with tuberous sclerosis complex. European Journal of Paediatric Neurology, 2016, 20, 649-651.	0.7	9
28	Association of Early MRI Characteristics With Subsequent Epilepsy and Neurodevelopmental Outcomes in Children With Tuberous Sclerosis Complex. Neurology, 2022, 98, .	1.5	8
29	Results of quantitative EEG analysis are associated with autism spectrum disorder and development abnormalities in infants with tuberous sclerosis complex. Biomedical Signal Processing and Control, 2021, 68, 102658.	3.5	7
30	Lacosamide Lowers Valproate and Levetiracetam Levels. Neuropediatrics, 2017, 48, 188-189.	0.3	6
31	Oligodendroglial transcription factor (OLIG1 and OLIG2) mutations are not associated with Pelizaeusâ€“Merzbacher-like leukodystrophy. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 365-366.	1.1	2