Elzbieta Jurkiewicz

List of Publications by Year
in descending order

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72	2,113	304368	253896
papers	citations	22 h-index	43 g-index

72	72	72	3312
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Antiepileptic treatment before the onset of seizures reduces epilepsy severity and risk of mental retardation in infants with tuberous sclerosis complex. European Journal of Paediatric Neurology, 2011, 15, 424-431.	0.7	254
2	Absence of an Orphan Mitochondrial Protein, C19orf12, Causes a Distinct Clinical Subtype of Neurodegeneration with Brain Iron Accumulation. American Journal of Human Genetics, 2011, 89, 543-550.	2.6	224
3	New perspective in diagnostics of mitochondrial disorders: two yearsâ€™ experience with whole-exome sequencing at a national paediatric centre. Journal of Translational Medicine, 2016, 14, 174.	1.8	176
4	Long-term effect of everolimus on epilepsy and growth in children under 3 years of age treated for subependymal giant cell astrocytoma associated with tuberous sclerosis complex. European Journal of Paediatric Neurology, 2013, 17, 479-485.	0.7	87
5	Basal ganglia lesions in children and adults. European Journal of Radiology, 2013, 82, 837-849.	1.2	79
6	Cerebral tuber count and its impact on mental outcome of patients with tuberous sclerosis complex. Epilepsia, 2011, 52, 22-27.	2.6	65
7	Effective everolimus treatment of inoperable, life-threatening subependymal giant cell astrocytoma and intractable epilepsy in a patient with tuberous sclerosis complex. European Journal of Paediatric Neurology, 2012, 16, 83-85.	0.7	62
8	Early diagnosis of tuberous sclerosis complex: a race against time. How to make the diagnosis before seizures?. Orphanet Journal of Rare Diseases, 2018, 13, 25.	1.2	60
9	EEG abnormalities preceding the epilepsy onset in tuberous sclerosis complex patients â€“ A prospective study of 5 patients. European Journal of Paediatric Neurology, 2014, 18, 458-468.	0.7	58
10	Surgical Treatment of Subependymal Giant Cell Astrocytoma inÂTuberous Sclerosis Complex Patients. Pediatric Neurology, 2014, 50, 307-312.	1.0	58
11	Long-Term MRI Cell Tracking after Intraventricular Delivery in a Patient with Global Cerebral Ischemia and Prospects for Magnetic Navigation of Stem Cells within the CSF. PLoS ONE, 2014, 9, e97631.	1.1	55
12	Blood Pressure Rhythmicity and Visceral Fat in Children With Hypertension. Hypertension, 2013, 62, 782-788.	1.3	46
13	Congenital subependymal giant cell astrocytomas in patients with tuberous sclerosis complex. Child's Nervous System, 2014, 30, 2037-2042.	0.6	45
14	Epilepsy in newborns with tuberous sclerosis complex. European Journal of Paediatric Neurology, 2014, 18, 714-721.	0.7	44
15	Congenital disorder of glycosylphosphatidylinositol (GPI)-anchor biosynthesisâ€"The phenotype of two patients with novel mutations in the PIGN and PGAP2 genes. European Journal of Paediatric Neurology, 2016, 20, 462-473.	0.7	42
16	Intracerebroventricular Transplantation of Cord Blood-Derived Neural Progenitors in a Child with Severe Global Brain Ischemic Injury. Cell Medicine, 2010, 1, 71-80.	5.0	41
17	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	2.6	40
18	Magnetic resonance imaging in the evaluation of the fetal spinal canal contents. Brain and Development, 2011, 33, 10-20.	0.6	33

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19	Contrast enhancement pattern predicts poor survival for patients with non-WNT/SHH medulloblastoma tumours. Journal of Neuro-Oncology, 2015, 123, 65-73.	1.4	27
20	Traditional and New Methods of Bone Age Assessment-An Overview. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 251-262.	0.4	27
21	MRCP Versus ERCP in the Evaluation of Chronic Pancreatitis in Children. Pancreas, 2016, 45, 1115-1119.	0.5	25
22	Cyst-like cortical tubers in patients with tuberous sclerosis complex: MR imaging with the FLAIR sequence. Pediatric Radiology, 2006, 36, 498-501.	1.1	24
23	Acute fright induces onset of symptoms in vanishing white matter diseaseâ€"Case report. European Journal of Paediatric Neurology, 2006, 10, 192-193.	0.7	24
24	Brain lesions in tuberous sclerosis complex. Review. , 2010, 48, 139-49.		24
25	Trilateral retinoblastoma: an institutional experience and review of the literature. Child's Nervous System, 2010, 26, 129-132.	0.6	22
26	Evolution and novel radiological changes of neurodegeneration associated with mutations in C19orf12. Parkinsonism and Related Disorders, 2017, 39, 71-76.	1.1	22
27	Spinal Cord Compression in Maroteaux-Lamy Syndrome: Case Report and Review of the Literature with Effects of Enzyme Replacement Therapy. Pediatric Neurosurgery, 2012, 48, 191-198.	0.4	21
28	Magnetic resonance imaging of the brain in adenylosuccinate lyase deficiency: a report of seven cases and a review of the literature. European Journal of Pediatrics, 2012, 171, 131-138.	1.3	21
29	Giant intracranial aneurysm in a 9-year-old boy with tuberous sclerosis. Pediatric Radiology, 2006, 36, 463-463.	1.1	20
30	Infantile spasms and cytomegalovirus infection: antiviral and antiepileptic treatment. Developmental Medicine and Child Neurology, 2007, 49, 684-692.	1.1	20
31	Gaucher disease due to saposin C deficiency, previously described as non-neuronopathic form â€" No positive effects after 2-years of miglustat therapy. Molecular Genetics and Metabolism, 2011, 104, 627-630.	0.5	20
32	Clinical and molecular characteristics of newly reported mitochondrial disease entity caused by biallelic PARS2 mutations. Journal of Human Genetics, 2018, 63, 473-485.	1.1	19
33	Congenital brain tumors in a series of 56 patients. Child's Nervous System, 2012, 28, 1193-1201.	0.6	18
34	Enzyme Replacement Therapy in an Attenuated Case of Mucopolysaccharidosis Type I (Scheie Syndrome): A 6.5-Year Detailed Follow-Up. Pediatric Neurology, 2012, 47, 461-465.	1.0	18
35	MR imaging, apparent diffusion coefficient and histopathological features of desmoplastic infantile tumorsâ€"own experience and review of the literature. Child's Nervous System, 2015, 31, 251-259.	0.6	18
36	Attenuated Adenylosuccinate Lyase Deficiency: A Report of One Case and a Review of the Literature. Neuropediatrics, 2014, 45, 050-055.	0.3	13

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37	Cervical Spine MRI Findings in Patients with Mucopolysaccharidosis Type II. Pediatric Neurosurgery, 2015, 50, 26-30.	0.4	13
38	Giant cerebellar cavernous malformation in 4-month-old boy. Case report and review of the literature. Neurologia I Neurochirurgia Polska, 2013, 47, 595-600.	0.6	12
39	Original article Angiocentric glioma: a rare intractable epilepsy-related tumour in children. Folia Neuropathologica, 2014, 3, 253-259.	0.5	12
40	Mild Zellweger syndrome due to a novel PEX6 mutation: correlation between clinical phenotype and in silico prediction of variant pathogenicity. Journal of Applied Genetics, 2017, 58, 475-480.	1.0	12
41	Liver Angiomyolipomas in Tuberous Sclerosis Complexâ€"Their Incidence and Course. Pediatric Neurology, 2018, 78, 20-26.	1.0	12
42	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation in the first Polish patient. Brain and Development, 2011, 33, 713-717.	0.6	11
43	Clinical and radiological pictures of two newborn babies with manifestations of chondrodysplasia punctata and review of available literature.. Polski Przeglad Radiologii I Medycyny Nuklearnej, 2013, 78, 57-64.	1.0	11
44	Leigh disease due toSCO2mutations revealed at extended autopsy. Journal of Clinical Pathology, 2015, 68, 397-399.	1.0	10
45	A Pharmacokinetics, Efficacy, and Safety Study of Gadoterate Meglumine in Pediatric Subjects Aged Younger Than 2 Years. Investigative Radiology, 2018, 53, 70-79.	3.5	10
46	Pharmacokinetics, Safety, and Efficacy of Gadopiclenol in Pediatric Patients Aged 2 to 17 Years. Investigative Radiology, 2022, 57, 510-516.	3.5	10
47	Mild Zellweger syndrome due to functionally confirmed novel PEX1 variants. Journal of Applied Genetics, 2020, 61, 87-91.	1.0	9
48	Left and Right Ventricular Morphology, Function and Myocardial Deformation in Children with Left Ventricular Non-Compaction Cardiomyopathy: A Case-Control Cardiovascular Magnetic Resonance Study. Journal of Clinical Medicine, 2022, 11, 1104.	1.0	9
49	â€œDrop attacksâ€•as first clinical symptoms in a child carrying MTTK m.8344A>G mutation. Folia Neuropathologica, 2013, 4, 347-354.	0.5	8
50	Association of Early MRI Characteristics With Subsequent Epilepsy and Neurodevelopmental Outcomes in Children With Tuberous Sclerosis Complex. Neurology, 2022, 98, .	1.5	8
51	Subependymal giant cell astrocytomas with atypical histological features mimicking malignant gliomas. , 2011, 49, 39-46.		8
52	Arterial hypertension with brachydactyly in a 15-year-old boy. Pediatric Nephrology, 2003, 18, 814-819.	0.9	7
53	MRI findings in the young infant with brainstem disconnection and extracerebral features. Report of one case and review of the literature. Brain and Development, 2010, 32, 495-498.	0.6	7
54	Leigh syndrome in individuals bearing m.9185T>C MTATP6 variant. Is hyperventilation a factor which starts its development?. Metabolic Brain Disease, 2018, 33, 191-199.	1.4	7

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55	Implementation of Computed Tomography Angiography (CTA) and Computed Tomography Perfusion (CTP) in Polish Guidelines for Determination of Cerebral Circulatory Arrest (CCA) during Brain Death/Death by Neurological Criteria (BD/DNC) Diagnosis Procedure. Journal of Clinical Medicine, 2021, 10, 4237.	1.0	7
56	Early treatment of biotinâ€“thiamineâ€“responsive basal ganglia disease improves the prognosis. Molecular Genetics and Metabolism Reports, 2021, 29, 100801.	0.4	7
57	Antenatal diagnosis of the congenital craniopharyngioma. Polish Journal of Radiology, 2010, 75, 98-102.	0.5	7
58	Segmenting pediatric optic pathway gliomas from MRI using deep learning. Computers in Biology and Medicine, 2022, 142, 105237.	3.9	7
59	MRI of a family with leukoencephalypathy with vanishing white matter. Pediatric Radiology, 2005, 35, 1027-1030.	1.1	6
60	Adenylosuccinate lyase deficiency: The first identified polish patient. Brain and Development, 2007, 29, 600-602.	0.6	6
61	Neuropathological characteristics of the brain in two patients with SLC19A3 mutations related to the biotin-thiamine-responsive basal ganglia disease. Folia Neuropathologica, 2017, 2, 146-153.	0.5	6
62	Mild phenotype of glutaric aciduria type 1 in polish patients â€“ novel data from a group of 13 cases. Metabolic Brain Disease, 2019, 34, 641-649.	1.4	6
63	Infratentorial tumors in children - value of ADC in prediction of grade of neoplasms. Polish Journal of Radiology, 2010, 75, 18-23.	0.5	6
64	Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation: High Outcome Variation between Two Siblings. Neuropediatrics, 2014, 45, 188-191.	0.3	5
65	Severe central and peripheral paraneoplastic demyelination associated with tumours of the ovaries. Child's Nervous System, 2015, 31, 1601-1606.	0.6	5
66	MR venography in children and adolescents with multiple sclerosis does not show increased prevalence of extracranial veins anomalies. European Journal of Paediatric Neurology, 2014, 18, 218-222.	0.7	4
67	Persistent multifocal atrial tachycardia in infant with encephalocraniocutaneous lipomatosis: a case report. European Journal of Pediatrics, 2014, 173, 1577-1580.	1.3	4
68	Ectopic virilising adrenocortical tumour in the spinal region in an 8Âyear-old boy: a case report and review of the literature. Italian Journal of Pediatrics, 2015, 41, 62.	1.0	4
69	Medullary cap dysplasia. Neurology, 2015, 84, 102-103.	1.5	2
70	Invitation to participate in aÂmulti-center study for validation of cerebral computed tomography angiography and computed tomography perfusion in the determination of cerebral circulatory arrest during brain death/death by neurological criteria diagnosis procedure in paediatric population below 12 years of age. Anaesthesiology Intensive Therapy, 2021, 53, 97-102.	0.4	2
71	Spinal cord lesions in children and adolescents with multiple sclerosis â€“ Magnetic resonance imaging. Neurologia I Neurochirurgia Polska, 2017, 51, 77-81.	0.6	1
72	Single Nucleotide Polymorphisms of Interleukins and Toll-like Receptors and Neuroimaging Results in Newborns with Congenital HCMV Infection. Viruses, 2021, 13, 1783.	1.5	0