# Elzbieta Jurkiewicz

## List of Publications by Citations



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| 67 papers | 1,626 citations | 21 h-index | 38 g-index |
| 72 ext. papers | 1,892 ext. citations | 3.6 avg, IF | 4.18 L-index |

| # | Paper | IF | Citations |
|---|---|---|---|
| 67 | 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, 424-31 | 3.8 | 207 |
| 66 | 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, 543-50 | 11 | 182 |
| 65 | New perspective in diagnostics of mitochondrial disorders: two yearsSexperience with whole-exome sequencing at a national paediatric centre. *Journal of Translational Medicine*, **2016**, 14, 174 | 8.5 | 123 |
| 64 | 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, 479-85 | 3.8 | 82 |
| 63 | Basal ganglia lesions in children and adults. *European Journal of Radiology*, **2013**, 82, 837-49 | 4.7 | 64 |
| 62 | 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, 83-5 | 3.8 | 57 |
| 61 | 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 | 3.7 | 50 |
| 60 | Cerebral tuber count and its impact on mental outcome of patients with tuberous sclerosis complex. *Epilepsia*, **2011**, 52, 22-7 | 6.4 | 46 |
| 59 | Surgical treatment of subependymal giant cell astrocytoma in tuberous sclerosis complex patients. *Pediatric Neurology*, **2014**, 50, 307-12 | 2.9 | 45 |
| 58 | EEG abnormalities preceding the epilepsy onset in tuberous sclerosis complex patients - a prospective study of 5 patients. *European Journal of Paediatric Neurology*, **2014**, 18, 458-68 | 3.8 | 44 |
| 57 | Intracerebroventricular Transplantation of Cord Blood-Derived Neural Progenitors in a Child With Severe Global Brain Ischemic Injury. *Cell Medicine*, **2010**, 1, 71-80 | 4.9 | 38 |
| 56 | 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 | 4.2 | 37 |
| 55 | Congenital subependymal giant cell astrocytomas in patients with tuberous sclerosis complex. *Childs Nervous System*, **2014**, 30, 2037-42 | 1.7 | 37 |
| 54 | Blood pressure rhythmicity and visceral fat in children with hypertension. *Hypertension*, **2013**, 62, 782-8 | 8.5 | 34 |
| 53 | NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. *American Journal of Human Genetics*, **2018**, 102, 460-467 | 11 | 33 |
| 52 | Epilepsy in newborns with tuberous sclerosis complex. *European Journal of Paediatric Neurology*, **2014**, 18, 714-21 | 3.8 | 32 |
| 51 | Congenital disorder of glycosylphosphatidylinositol (GPI)-anchor biosynthesis--The phenotype of two patients with novel mutations in the PIGN and PGAP2 genes. *European Journal of Paediatric Neurology*, **2016**, 20, 462-73 | 3.8 | 30 |

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| 50 | Magnetic resonance imaging in the evaluation of the fetal spinal canal contents. *Brain and Development*, **2011**, 33, 10-20 | 2.2 | 24 |
| 49 | Brain lesions in tuberous sclerosis complex. Review **2010**, 48, 139-49 | | 24 |
| 48 | Cyst-like cortical tubers in patients with tuberous sclerosis complex: MR imaging with the FLAIR sequence. *Pediatric Radiology*, **2006**, 36, 498-501 | 2.8 | 22 |
| 47 | Contrast enhancement pattern predicts poor survival for patients with non-WNT/SHH medulloblastoma tumours. *Journal of Neuro-Oncology*, **2015**, 123, 65-73 | 4.8 | 21 |
| 46 | Acute fright induces onset of symptoms in vanishing white matter disease-case report. *European Journal of Paediatric Neurology*, **2006**, 10, 192-3 | 3.8 | 20 |
| 45 | Gaucher disease due to saposin C deficiency, previously described as non-neuronopathic form--no positive effects after 2-years of miglustat therapy. *Molecular Genetics and Metabolism*, **2011**, 104, 627-30 | 3.7 | 19 |
| 44 | MR imaging, apparent diffusion coefficient and histopathological features of desmoplastic infantile tumors-own experience and review of the literature. *Childs Nervous System*, **2015**, 31, 251-9 | 1.7 | 18 |
| 43 | Giant intracranial aneurysm in a 9-year-old boy with tuberous sclerosis. *Pediatric Radiology*, **2006**, 36, 463 | 2.8 | 18 |
| 42 | MRCP Versus ERCP in the Evaluation of Chronic Pancreatitis in Children: Which Is the Better Choice?. *Pancreas*, **2016**, 45, 1115-9 | 2.6 | 18 |
| 41 | Spinal cord compression in Maroteaux-Lamy syndrome: case report and review of the literature with effects of enzyme replacement therapy. *Pediatric Neurosurgery*, **2012**, 48, 191-8 | 0.9 | 17 |
| 40 | Trilateral retinoblastoma: an institutional experience and review of the literature. *Childs Nervous System*, **2010**, 26, 129-32 | 1.7 | 17 |
| 39 | Congenital brain tumors in a series of 56 patients. *Childs Nervous System*, **2012**, 28, 1193-201 | 1.7 | 16 |
| 38 | Enzyme replacement therapy in an attenuated case of mucopolysaccharidosis type I (Scheie syndrome): a 6.5-year detailed follow-up. *Pediatric Neurology*, **2012**, 47, 461-5 | 2.9 | 16 |
| 37 | Infantile spasms and cytomegalovirus infection: antiviral and antiepileptic treatment. *Developmental Medicine and Child Neurology*, **2007**, 49, 684-92 | 3.3 | 16 |
| 36 | Evolution and novel radiological changes of neurodegeneration associated with mutations in C19orf12. *Parkinsonism and Related Disorders*, **2017**, 39, 71-76 | 3.6 | 15 |
| 35 | 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, 131-8 | 4.1 | 15 |
| 34 | Attenuated adenylosuccinate lyase deficiency: a report of one case and a review of the literature. *Neuropediatrics*, **2014**, 45, 50-5 | 1.6 | 12 |
| 33 | Clinical and molecular characteristics of newly reported mitochondrial disease entity caused by biallelic PARS2 mutations. *Journal of Human Genetics*, **2018**, 63, 473-485 | 4.3 | 11 |

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| 32 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation in the first Polish patient. *Brain and Development*, **2011**, 33, 713-7 | 2.2 | 11 |
| 31 | Liver Angiomyolipomas in Tuberous Sclerosis Complex-Their Incidence and Course. *Pediatric Neurology*, **2018**, 78, 20-26 | 2.9 | 10 |
| 30 | Angiocentric glioma: a rare intractable epilepsy-related tumour in children. *Folia Neuropathologica*, **2014**, 52, 253-9 | 2.6 | 10 |
| 29 | Leigh disease due to SCO2 mutations revealed at extended autopsy. *Journal of Clinical Pathology*, **2015**, 68, 397-9 | 3.9 | 9 |
| 28 | 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 | 2.5 | 8 |
| 27 | Cervical spine MRI findings in patients with Mucopolysaccharidosis type II. *Pediatric Neurosurgery*, **2015**, 50, 26-30 | 0.9 | 8 |
| 26 | A Pharmacokinetics, Efficacy, and Safety Study of Gadoterate Meglumine in Pediatric Subjects Aged Younger Than 2 Years. *Investigative Radiology*, **2018**, 53, 70-79 | 10.1 | 8 |
| 25 | "Drop attacks" as first clinical symptoms in a child carrying MTTK m.8344A>G mutation. *Folia Neuropathologica*, **2013**, 51, 347-54 | 2.6 | 8 |
| 24 | Giant cerebellar cavernous malformation in 4-month-old boy. Case report and review of the literature. *Neurologia I Neurochirurgia Polska*, **2013**, 47, 596-600 | 1 | 7 |
| 23 | 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, 495-8 | 2.2 | 7 |
| 22 | 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 | | 7 |
| 21 | Antenatal diagnosis of the congenital craniopharyngioma. *Polish Journal of Radiology*, **2010**, 75, 98-102 | 1.6 | 7 |
| 20 | Subependymal giant cell astrocytomas with atypical histological features mimicking malignant gliomas **2011**, 49, 39-46 | | 7 |
| 19 | Adenylosuccinate lyase deficiency: the first identified polish patient. *Brain and Development*, **2007**, 29, 600-2 | 2.2 | 6 |
| 18 | Infratentorial tumors in children - value of ADC in prediction of grade of neoplasms. *Polish Journal of Radiology*, **2010**, 75, 18-23 | 1.6 | 6 |
| 17 | Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation: high outcome variation between two siblings. *Neuropediatrics*, **2014**, 45, 188-91 | 1.6 | 5 |
| 16 | Arterial hypertension with brachydactyly in a 15-year-old boy. *Pediatric Nephrology*, **2003**, 18, 814-9 | 3.2 | 5 |
| 15 | MRI of a family with leukoencephalypathy with vanishing white matter. *Pediatric Radiology*, **2005**, 35, 1027-30 | 2.8 | 5 |

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| 14 | 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 | 3.9 | 5 |
| 13 | Traditional and New Methods of Bone Age Assessment-An Overview. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2021**, 13, 251-262 | 1.9 | 5 |
| 12 | Neuropathological characteristics of the brain in two patients with SLC19A3 mutations related to the biotin-thiamine-responsive basal ganglia disease. *Folia Neuropathologica*, **2017**, 55, 146-153 | 2.6 | 3 |
| 11 | 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, 218-22 | 3.8 | 3 |
| 10 | Mild Zellweger syndrome due to functionally confirmed novel PEX1 variants. *Journal of Applied Genetics*, **2020**, 61, 87-91 | 2.5 | 3 |
| 9 | Severe central and peripheral paraneoplastic demyelination associated with tumours of the ovaries. *Childs Nervous System*, **2015**, 31, 1601-6 | 1.7 | 2 |
| 8 | Persistent multifocal atrial tachycardia in infant with encephalocraniocutaneous lipomatosis: a case report. *European Journal of Pediatrics*, **2014**, 173, 1577-80 | 4.1 | 2 |
| 7 | Ectopic virilising adrenocortical tumour in the spinal region in an 8 year-old boy: a case report and review of the literature. *Italian Journal of Pediatrics*, **2015**, 41, 62 | 3.2 | 2 |
| 6 | Mild phenotype of glutaric aciduria type 1 in polish patients - novel data from a group of 13 cases. *Metabolic Brain Disease*, **2019**, 34, 641-649 | 3.9 | 2 |
| 5 | Spinal cord lesions in children and adolescents with multiple sclerosis - Magnetic resonance imaging. *Neurologia I Neurochirurgia Polska*, **2017**, 51, 77-81 | 1 | 1 |
| 4 | Medullary cap dysplasia: MRI and diffusion tensor imaging of a hindbrain malformation. *Neurology*, **2015**, 84, 102-3 | 6.5 | 1 |
| 3 | Segmenting pediatric optic pathway gliomas from MRI using deep learning.. *Computers in Biology and Medicine*, **2022**, 142, 105237 | 7 | 1 |
| 2 | Invitation to participate in a multi-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 | 1.7 | 0 |
| 1 | Early treatment of biotin-thiamine-responsive basal ganglia disease improves the prognosis. *Molecular Genetics and Metabolism Reports*, **2021**, 29, 100801 | 1.8 | 0 |