Ewa Pronicka

List of Publications by Year
in descending order

72	2,611	185998 28	197535 49
papers	citations	h-index	g-index

76	76	76	4050
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	2.6	4
2	Expanding the clinical and genetic spectrum of FDXR deficiency by functional validation of variants of uncertain significance. Human Mutation, 2021, 42, 310-319.	1.1	11
3	Analysis of vitamin D3 metabolites in survivors of infantile idiopathic hypercalcemia caused by CYP24A1 mutation or SLC34A1 mutation. Journal of Steroid Biochemistry and Molecular Biology, 2021, 208, 105824.	1.2	2
4	Long-term outcome of the survivors of infantile hypercalcaemia with CYP24A1 and SLC34A1 mutations. Nephrology Dialysis Transplantation, 2020, 36, 1484-1492.	0.4	12
5	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	1.7	17
6	The frequency of mitochondrial polymerase gamma related disorders in a large Polish population cohort. Mitochondrion, 2019, 47, 179-187.	1.6	4
7	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
8	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
9	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	1.1	22
10	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
11	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	1.2	61
12	Identification of the first in Poland CACNA1A gene mutation in familial hemiplegic migraine. Case report. Neurologia I Neurochirurgia Polska, 2017, 51, 184-189.	0.6	2
13	Hypocapnic hypothesis of Leigh disease. Medical Hypotheses, 2017, 101, 23-27.	0.8	6
14	Biallelic mutations in CYP24A1 or SLC34A1 as a cause of infantile idiopathic hypercalcemia (IIH) with vitamin D hypersensitivity: molecular study of 11 historical IIH cases. Journal of Applied Genetics, 2017, 58, 349-353.	1.0	66
15	3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. Clinica Chimica Acta, 2017, 471, 95-100.	0.5	14
16	Coexistence of mutations in keratin 10 (KRT10) and the mitochondrial genome in a patient with ichthyosis with confetti and Leber's hereditary optic neuropathy. American Journal of Medical Genetics, Part A, 2017, 173, 3093-3097.	0.7	7
17	Long-term clinical effects of enzyme replacement therapy in MPS II. Pediatria Polska, 2017, 92, 373-377.	0.1	3
18	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	2.8	63

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19	A scoring system predicting the clinical course of CLPB defect based on the foetal and neonatal presentation of 31 patients. Journal of Inherited Metabolic Disease, 2017, 40, 853-860.	1.7	27
20	Bilateral striatal necrosis caused by ADAR mutations in two siblings with dystonia and freckles-like skin changes that should be differentiated from Leigh syndrome. Folia Neuropathologica, 2016, 4, 405-409.	0.5	8
21	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 217-227.	2.6	57
22	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
23	Patient's weight can decide about spending millions on enzyme replacement therapy in MPS II. Molecular Genetics and Metabolism Reports, 2016, 6, 5-7.	0.4	1
24	NAXE Mutations Disrupt the Cellular NAD(P)HX Repair System and Cause a Lethal Neurometabolic Disorder of Early Childhood. American Journal of Human Genetics, 2016, 99, 894-902.	2.6	75
25	Autosomal-Recessive Mutations in SLC34A1 Encoding Sodium-Phosphate Cotransporter 2A Cause Idiopathic Infantile Hypercalcemia. Journal of the American Society of Nephrology: JASN, 2016, 27, 604-614.	3.0	207
26	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
27	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.	2.6	111
28	Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. Neuropediatrics, 2015, 46, 098-103.	0.3	34
29	Yeast model analysis of novel polymerase gamma variants found in patients with autosomal recessive mitochondrial disease. Human Genetics, 2015, 134, 951-966.	1.8	17
30	No Evidence for Association of SCO2 Heterozygosity with High-Grade Myopia or Other Diseases with Possible Mitochondrial Dysfunction. JIMD Reports, 2015, 27, 63-68.	0.7	7
31	Age and Gender-Related Changes in Biogenic Amine Metabolites in Cerebrospinal Fluid in Children. Advances in Experimental Medicine and Biology, 2015, 878, 73-82.	0.8	1
32	Severe phenotypes of SMARD1 associated with novel mutations of the IGHMBP2 gene and nuclear degeneration of muscle and Schwann cells. European Journal of Paediatric Neurology, 2014, 18, 183-192.	0.7	20
33	The natural history of SCO2 deficiency in 36 Polish children confirmed the genotypeâ€“phenotype correlation. Mitochondrion, 2013, 13, 810-816.	1.6	19
34	Left ventricular noncompaction (LVNC) and low mitochondrial membrane potential are specific for Barth syndrome. Journal of Inherited Metabolic Disease, 2013, 36, 929-937.	1.7	23
35	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
36	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	9.4	175

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37	Adaptation of respiratory chain biogenesis to cytochrome c oxidase deficiency caused by SURF1 gene mutations. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1114-1124.	1.8	30
38	Molecular investigations of mitochondrial deletions: Evaluating the usefulness of different genetic tests. Gene, 2012, 506, 161-165.	1.0	10
39	Drug-resistant epilepsia and fulminant valproate liver toxicity. Alpers-Huttenlocher syndrome in two children confirmed post mortem by identification of p.W748S mutation in POLG gene. Medical Science Monitor, 2011, 17, CR203-CR209.	0.5	35
40	Urgent metabolic service improves survival in longâ€chain 3â€hydroxyacylâ€CoA dehydrogenase (LCHAD) deficiency detected by symptomatic identification and pilot newborn screening. Journal of Inherited Metabolic Disease, 2011, 34, 185-195.	1.7	30
41	Post mortem identification of deoxyguanosine kinase (DGUOK) gene mutations combined with impaired glucose homeostasis and iron overload features in four infants with severe progressive liver failure. Journal of Applied Genetics, 2011, 52, 61-66.	1.0	40
42	Increased reactive oxygen species (ROS) production and low catalase level in fibroblasts of a girl with MEGDEL association (Leigh syndrome, deafness, 3-methylglutaconic aciduria). , 2011, 49, 56-63.		11
43	Oxidative stress-dependent p66Shc phosphorylation in skin fibroblasts of children with mitochondrial disorders. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 952-960.	0.5	65
44	Mutations in ZIC2 in human holoprosencephaly: description of a Novel ZIC2 specific phenotype and comprehensive analysis of 157 individuals. Journal of Medical Genetics, 2010, 47, 513-524.	1.5	75
45	A comprehensive HADHA c.1528G>C frequency study reveals high prevalence of longâ€chain 3â€hydroxyacylâ€CoA dehydrogenase deficiency in Poland. Journal of Inherited Metabolic Disease, 2010, 33, 373-377.	1.7	45
46	High prevalence of SURF1 c.845_846delCT mutation in Polish Leigh patients. European Journal of Paediatric Neurology, 2009, 13, 146-153.	0.7	17
47	Clinical course of homozygous familial hypercholesterolemia during childhood: report on 4 unrelated patients with homozygous or compound heterozygous mutations in theLDLR gene. Journal of Applied Genetics, 2008, 49, 109-113.	1.0	6
48	Clinical, biochemical and molecular findings in seven Polish patients with adenylosuccinate lyase deficiency. Molecular Genetics and Metabolism, 2008, 94, 435-442.	0.5	62
49	Elevated Carbohydrate-Deficient Transferrin (CDT) and Its Normalization on Dietary Treatment as a Useful Biochemical Test for Hereditary Fructose Intolerance and Galactosemia. Pediatric Research, 2007, 62, 101-105.	1.1	40
50	False Positive Results of Mitochondrial DNA Depletion/Deletion due to Single Nucleotide Substitutions Causing Appearance of Additional PvuII Restriction Sites. Diagnostic Molecular Pathology, 2007, 16, 116-120.	2.1	2
51	Characterization of two unusual truncating PMM2 mutations in two CDG-Ia patients. Molecular Genetics and Metabolism, 2007, 90, 408-413.	0.5	42
52	Diversity of cystathionine Î²-synthase haplotypes bearing the most common homocystinuria mutation c.833T>C: a possible role for gene conversion. Human Mutation, 2007, 28, 255-264.	1.1	20
53	Aldolase B mutations and prevalence of hereditary fructose intolerance in a Polish population. Molecular Genetics and Metabolism, 2006, 87, 376-378.	0.5	21
54	Genetic background of HSH in three Polish families and a patient with an X;9 translocation. European Journal of Human Genetics, 2006, 14, 55-62.	1.4	23

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55	Retrospective, Multicentric Study of 180 Children with Cytochrome c Oxidase Deficiency. Pediatric Research, 2006, 59, 21-26.	1.1	142
56	MRI of a family with leukoencephalypathy with vanishing white matter. Pediatric Radiology, 2005, 35, 1027-1030.	1.1	6
57	Decreased affinity for oxygen of cytochrome-coxidase in Leigh syndrome caused bySURF1mutations. American Journal of Physiology - Cell Physiology, 2004, 287, C1384-C1388.	2.1	39
58	Anthropometric characteristics of X-linked hypophosphatemia. American Journal of Medical Genetics Part A, 2004, 126A, 141-149.	2.4	9
59	The cystathionine ?-synthase (CBS) mutation c.1224-2A>C in Central Europe: Vitamin B6 nonresponsiveness and a common ancestral haplotype. Human Mutation, 2004, 24, 352-353.	1.1	16
60	Clinical heterogeneity and molecular findings in five Polish patients with glycerol kinase deficiency: investigation of two splice site mutations with computerized splice junction analysis and Xp21 gene-specific mRNA analysis. Molecular Genetics and Metabolism, 2003, 79, 149-159.	0.5	17
61	Diversity of clinical symptoms in A3243G mitochondrial DNA mutation (MELAS syndrome mutation). Medical Science Monitor, 2002, 8, CR767-73.	0.5	18
62	Abnormal Calcium Homeostasis in Fibroblasts from Patients with Leigh Disease. Biochemical and Biophysical Research Communications, 2001, 283, 687-693.	1.0	18
63	13C NMR spectroscopy: a convenient tool for detection of argininosuccinic aciduria. Journal of Pharmaceutical and Biomedical Analysis, 2001, 26, 401-408.	1.4	12
64	Dominantly inherited isolated hyperparathyroidism: a syndromic association?. Pediatric Radiology, 1999, 29, 10-15.	1.1	5
65	Distribution of mutations in the PEX gene in families with X-linked hypophosphataemic rickets (HYP). Human Molecular Genetics, 1997, 6, 539-549.	1.4	184
66	Persistent hypercalciuria and elevated 25-hydroxyvitamin D 3 in children with infantile hypercalcaemia. Pediatric Nephrology, 1997, 11, 2-6.	0.9	38
67	A Survey of the Newborn Populations in Belgium, Germany, Poland, Czech Republic, Hungary, Bulgaria, Spain, Turkey, and Japan for the G985 Variant Allele with Haplotype Analysis at the Medium Chain Acyl-CoA. Pediatric Research, 1997, 41, 201-209.	1.1	60
68	Inhibition of Phosphomannose Isomerase by Fructose 1-Phosphate: An Explanation for Defective N-Glycosylation in Hereditary Fructose Intolerance. Pediatric Research, 1996, 40, 764-766.	1.1	71
69	Pancreatic glucagon levels in infants and children with hyperinsulinemia. Journal of Pediatrics, 1995, 126, 948-950.	0.9	3
70	Abolished phosphaturic response to parathormone in adult patients with Fahr disease and its restoration after propranolol adminstration. Journal of Neurology, 1988, 235, 185-187.	1.8	6
71	Increased serum level of 1,25-dihydroxyvitamin D3 after parathyroid hormone in the normocalcemic phase of idiopathic hypercalcemia. Journal of Pediatrics, 1988, 112, 930-933.	0.9	11
72	Three cases of beta-galactosidase deficiency. Klinische Padiatrie, 1981, 193, 343-346.	0.2	5