

Ewa Pronicka

List of Publications by Year in descending order

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72
papers

2,611
citations

185998

28
h-index

197535

49
g-index

76
all docs

76
docs citations

76
times ranked

4050
citing authors

#	ARTICLE	IF	CITATIONS
1	Autosomal-Recessive Mutations in SLC34A1 Encoding Sodium-Phosphate Cotransporter 2A Cause Idiopathic Infantile Hypercalcemia. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 604-614.	3.0	207
2	Distribution of mutations in the PEX gene in families with X-linked hypophosphataemic rickets (HYP). <i>Human Molecular Genetics</i> , 1997, 6, 539-549.	1.4	184
3	New perspective in diagnostics of mitochondrial disorders: two years'™ experience with whole-exome sequencing at a national paediatric centre. <i>Journal of Translational Medicine</i> , 2016, 14, 174.	1.8	176
4	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. <i>Nature Genetics</i> , 2012, 44, 797-802.	9.4	175
5	Retrospective, Multicentric Study of 180 Children with Cytochrome c Oxidase Deficiency. <i>Pediatric Research</i> , 2006, 59, 21-26.	1.1	142
6	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. <i>American Journal of Human Genetics</i> , 2015, 96, 245-257.	2.6	111
7	Mutations in ZIC2 in human holoprosencephaly: description of a Novel ZIC2 specific phenotype and comprehensive analysis of 157 individuals. <i>Journal of Medical Genetics</i> , 2010, 47, 513-524.	1.5	75
8	NAXE Mutations Disrupt the Cellular NAD(P)HX Repair System and Cause a Lethal Neurometabolic Disorder of Early Childhood. <i>American Journal of Human Genetics</i> , 2016, 99, 894-902.	2.6	75
9	Inhibition of Phosphomannose Isomerase by Fructose 1-Phosphate: An Explanation for Defective N-Glycosylation in Hereditary Fructose Intolerance. <i>Pediatric Research</i> , 1996, 40, 764-766.	1.1	71
10	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. <i>Journal of Applied Genetics</i> , 2017, 58, 349-353.	1.0	66
11	Oxidative stress-dependent p66Shc phosphorylation in skin fibroblasts of children with mitochondrial disorders. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , 2010, 1797, 952-960.	0.5	65
12	Progressive deafness'–dystonia due to <i>SERAC1</i> mutations: A study of 67 cases. <i>Annals of Neurology</i> , 2017, 82, 1004-1015.	2.8	63
13	Clinical, biochemical and molecular findings in seven Polish patients with adenylosuccinate lyase deficiency. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 435-442.	0.5	62
14	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 120.	1.2	61
15	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. <i>Pediatric Research</i> , 1997, 41, 201-209.	1.1	60
16	Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 217-227.	2.6	57
17	A comprehensive <i>HADHA</i> c.1528G>C frequency study reveals high prevalence of long'–chain 3'–hydroxyacyl'–CoA dehydrogenase deficiency in Poland. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 373-377.	1.7	45
18	Characterization of two unusual truncating PMM2 mutations in two CDG-Ia patients. <i>Molecular Genetics and Metabolism</i> , 2007, 90, 408-413.	0.5	42

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19	Congenital disorder of glycosylphosphatidylinositol (GPI)-anchor biosynthesisâ€”The phenotype of two patients with novel mutations in the PIGN and PGAP2 genes. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 462-473.	0.7	42
20	Elevated Carbohydrate-Deficient Transferrin (CDT) and Its Normalization on Dietary Treatment as a Useful Biochemical Test for Hereditary Fructose Intolerance and Galactosemia. <i>Pediatric Research</i> , 2007, 62, 101-105.	1.1	40
21	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. <i>Journal of Applied Genetics</i> , 2011, 52, 61-66.	1.0	40
22	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. <i>American Journal of Human Genetics</i> , 2018, 102, 460-467.	2.6	40
23	Decreased affinity for oxygen of cytochrome-c oxidase in Leigh syndrome caused by SURF1 mutations. <i>American Journal of Physiology - Cell Physiology</i> , 2004, 287, C1384-C1388.	2.1	39
24	Persistent hypercalciuria and elevated 25-hydroxyvitamin D 3 in children with infantile hypercalcaemia. <i>Pediatric Nephrology</i> , 1997, 11, 2-6.	0.9	38
25	Drug-resistant epilepsy and fulminant valproate liver toxicity. Alpers-Huttenlocher syndrome in two children confirmed post mortem by identification of p.W748S mutation in POLG gene. <i>Medical Science Monitor</i> , 2011, 17, CR203-CR209.	0.5	35
26	Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. <i>Neuropediatrics</i> , 2015, 46, 098-103.	0.3	34
27	Urgent metabolic service improves survival in longâ€”chain 3â€”hydroxyacylâ€”CoA dehydrogenase (LCHAD) deficiency detected by symptomatic identification and pilot newborn screening. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 185-195.	1.7	30
28	Adaptation of respiratory chain biogenesis to cytochrome c oxidase deficiency caused by SURF1 gene mutations. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 1114-1124.	1.8	30
29	A scoring system predicting the clinical course of CLPB defect based on the foetal and neonatal presentation of 31 patients. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 853-860.	1.7	27
30	Genetic background of HSH in three Polish families and a patient with an X;9 translocation. <i>European Journal of Human Genetics</i> , 2006, 14, 55-62.	1.4	23
31	Left ventricular noncompaction (LVNC) and low mitochondrial membrane potential are specific for Barth syndrome. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 929-937.	1.7	23
32	Clinical, biochemical, and genetic features associated with <i>VAR2</i> -related mitochondrial disease. <i>Human Mutation</i> , 2018, 39, 563-578.	1.1	22
33	Aldolase B mutations and prevalence of hereditary fructose intolerance in a Polish population. <i>Molecular Genetics and Metabolism</i> , 2006, 87, 376-378.	0.5	21
34	Diversity of cystathionine β -synthase haplotypes bearing the most common homocystinuria mutation c.833T>C: a possible role for gene conversion. <i>Human Mutation</i> , 2007, 28, 255-264.	1.1	20
35	Severe phenotypes of SMARD1 associated with novel mutations of the IGHMBP2 gene and nuclear degeneration of muscle and Schwann cells. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 183-192.	0.7	20
36	The natural history of SCO2 deficiency in 36 Polish children confirmed the genotypeâ€”phenotype correlation. <i>Mitochondrion</i> , 2013, 13, 810-816.	1.6	19

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37	Clinical and molecular characteristics of newly reported mitochondrial disease entity caused by biallelic PARS2 mutations. <i>Journal of Human Genetics</i> , 2018, 63, 473-485.	1.1	19
38	Abnormal Calcium Homeostasis in Fibroblasts from Patients with Leigh Disease. <i>Biochemical and Biophysical Research Communications</i> , 2001, 283, 687-693.	1.0	18
39	Diversity of clinical symptoms in A3243G mitochondrial DNA mutation (MELAS syndrome mutation). <i>Medical Science Monitor</i> , 2002, 8, CR767-73.	0.5	18
40	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. <i>Molecular Genetics and Metabolism</i> , 2003, 79, 149-159.	0.5	17
41	High prevalence of SURF1 c.845_846delCT mutation in Polish Leigh patients. <i>European Journal of Paediatric Neurology</i> , 2009, 13, 146-153.	0.7	17
42	Yeast model analysis of novel polymerase gamma variants found in patients with autosomal recessive mitochondrial disease. <i>Human Genetics</i> , 2015, 134, 951-966.	1.8	17
43	Leigh syndrome caused by mutations in <i>MTFMT</i> is associated with a better prognosis. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 515-524.	1.7	17
44	The cystathionine γ -synthase (CBS) mutation c.1224-2A>C in Central Europe: Vitamin B6 nonresponsiveness and a common ancestral haplotype. <i>Human Mutation</i> , 2004, 24, 352-353.	1.1	16
45	3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. <i>Clinica Chimica Acta</i> , 2017, 471, 95-100.	0.5	14
46	¹³ C NMR spectroscopy: a convenient tool for detection of argininosuccinic aciduria. <i>Journal of Pharmaceutical and Biomedical Analysis</i> , 2001, 26, 401-408.	1.4	12
47	Long-term outcome of the survivors of infantile hypercalcaemia with CYP24A1 and SLC34A1 mutations. <i>Nephrology Dialysis Transplantation</i> , 2020, 36, 1484-1492.	0.4	12
48	Increased serum level of 1,25-dihydroxyvitamin D3 after parathyroid hormone in the normocalcemic phase of idiopathic hypercalcemia. <i>Journal of Pediatrics</i> , 1988, 112, 930-933.	0.9	11
49	Expanding the clinical and genetic spectrum of FXR deficiency by functional validation of variants of uncertain significance. <i>Human Mutation</i> , 2021, 42, 310-319.	1.1	11
50	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
51	Molecular investigations of mitochondrial deletions: Evaluating the usefulness of different genetic tests. <i>Gene</i> , 2012, 506, 161-165.	1.0	10
52	Anthropometric characteristics of X-linked hypophosphatemia. <i>American Journal of Medical Genetics Part A</i> , 2004, 126A, 141-149.	2.4	9
53	Drop attacks as first clinical symptoms in a child carrying MTK m.8344A>G mutation. <i>Folia Neuropathologica</i> , 2013, 4, 347-354.	0.5	8
54	Bilateral striatal necrosis caused by ADAR mutations in two siblings with dystonia and freckles-like skin changes that should be differentiated from Leigh syndrome. <i>Folia Neuropathologica</i> , 2016, 4, 405-409.	0.5	8

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55	No Evidence for Association of SCO2 Heterozygosity with High-Grade Myopia or Other Diseases with Possible Mitochondrial Dysfunction. <i>JIMD Reports</i> , 2015, 27, 63-68.	0.7	7
56	Coexistence of mutations in keratin 10 (KRT10) and the mitochondrial genome in a patient with ichthyosis with confetti and Leber's hereditary optic neuropathy. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3093-3097.	0.7	7
57	Leigh syndrome in individuals bearing m.9185T>C MTATP6 variant. Is hyperventilation a factor which starts its development?. <i>Metabolic Brain Disease</i> , 2018, 33, 191-199.	1.4	7
58	Abolished phosphaturic response to parathormone in adult patients with Fahr disease and its restoration after propranolol administration. <i>Journal of Neurology</i> , 1988, 235, 185-187.	1.8	6
59	MRI of a family with leukoencephalopathy with vanishing white matter. <i>Pediatric Radiology</i> , 2005, 35, 1027-1030.	1.1	6
60	Clinical course of homozygous familial hypercholesterolemia during childhood: report on 4 unrelated patients with homozygous or compound heterozygous mutations in the LDLR gene. <i>Journal of Applied Genetics</i> , 2008, 49, 109-113.	1.0	6
61	Hypocapnic hypothesis of Leigh disease. <i>Medical Hypotheses</i> , 2017, 101, 23-27.	0.8	6
62	Three cases of beta-galactosidase deficiency. <i>Klinische Padiatrie</i> , 1981, 193, 343-346.	0.2	5
63	Dominantly inherited isolated hyperparathyroidism: a syndromic association?. <i>Pediatric Radiology</i> , 1999, 29, 10-15.	1.1	5
64	The frequency of mitochondrial polymerase gamma related disorders in a large Polish population cohort. <i>Mitochondrion</i> , 2019, 47, 179-187.	1.6	4
65	<i>PIGN</i>encephalopathy: Characterizing the epileptology. <i>Epilepsia</i> , 2022, 63, 974-991.	2.6	4
66	Pancreatic glucagon levels in infants and children with hyperinsulinemia. <i>Journal of Pediatrics</i> , 1995, 126, 948-950.	0.9	3
67	Long-term clinical effects of enzyme replacement therapy in MPS II. <i>Pediatrica Polska</i> , 2017, 92, 373-377.	0.1	3
68	False Positive Results of Mitochondrial DNA Depletion/Deletion due to Single Nucleotide Substitutions Causing Appearance of Additional PvuII Restriction Sites. <i>Diagnostic Molecular Pathology</i> , 2007, 16, 116-120.	2.1	2
69	Identification of the first in Poland CACNA1A gene mutation in familial hemiplegic migraine. Case report. <i>Neurologia I Neurochirurgia Polska</i> , 2017, 51, 184-189.	0.6	2
70	Analysis of vitamin D3 metabolites in survivors of infantile idiopathic hypercalcemia caused by CYP24A1 mutation or SLC34A1 mutation. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2021, 208, 105824.	1.2	2
71	Age and Gender-Related Changes in Biogenic Amine Metabolites in Cerebrospinal Fluid in Children. <i>Advances in Experimental Medicine and Biology</i> , 2015, 878, 73-82.	0.8	1
72	Patient's weight can decide about spending millions on enzyme replacement therapy in MPS II. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 6, 5-7.	0.4	1