Jolanta Sykut-Cegielska

List of Publications by Citations

78 papers	2,680 citations	29 h-index	51 g-index
86 ext. papers	3,094 ext. citations	5.5 avg, IF	4.07 L-index

#	Paper	IF	Citations
78	SRD5A3 is required for converting polyprenol to dolichol and is mutated in a congenital glycosylation disorder. *Cell*, **2010**, 142, 203-17	56.2	207
77	Multiple phenotypes in phosphoglucomutase 1 deficiency. *New England Journal of Medicine*, **2014**, 370, 533-42	59.2	197
76	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1041-57	5.4	143
75	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1059-74	5.4	135
74	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. *Human Molecular Genetics*, **2012**, 21, 4151-61	5.6	126
73	Retrospective, multicentric study of 180 children with cytochrome C oxidase deficiency. *Pediatric Research*, **2006**, 59, 21-6	3.2	125
72	Clinical ascertainment of Nijmegen breakage syndrome (NBS) and prevalence of the major mutation, 657del5, in three Slav populations. *European Journal of Human Genetics*, **2000**, 8, 900-2	5.3	109
71	Sepiapterin reductase deficiency: a treatable mimic of cerebral palsy. *Annals of Neurology*, **2012**, 71, 520-30	9.4	102
70	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. *Nature Communications*, **2016**, 7, 11600	17.4	83
69	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. *Human Mutation*, **2010**, 31, 380-90	4.7	80
68	Cross-border healthcare? The Polish experience. *Orphanet Journal of Rare Diseases*, **2010**, 5,	4.2	78
67	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. *Brain*, **2010**, 133, 3210-20	11.2	76
66	Clinical Features of Lysosomal Acid Lipase Deficiency. *Journal of Pediatric Gastroenterology and Nutrition*, **2015**, 61, 619-25	2.8	73
65	Partial biotinidase deficiency is usually due to the D444H mutation in the biotinidase gene. *Human Genetics*, **1998**, 102, 571-5	6.3	67
64	Defining the phenotype in an autosomal recessive cutis laxa syndrome with a combined congenital defect of glycosylation. *European Journal of Human Genetics*, **2008**, 16, 28-35	5.3	61
63	Clinical, biochemical and molecular findings in seven Polish patients with adenylosuccinate lyase deficiency. *Molecular Genetics and Metabolism*, **2008**, 94, 435-442	3.7	51
62	Ophthalmological abnormalities in children with congenital disorders of glycosylation type I. *British Journal of Ophthalmology*, **2009**, 93, 350-4	5.5	48

61	International clinical guidelines for the management of phosphomannomutase 2-congenital disorders of glycosylation: Diagnosis, treatment and follow up. *Journal of Inherited Metabolic Disease*, **2019**, 42, 5-28	5.4	45
60	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. *Journal of Inherited Metabolic Disease*, **2016**, 39, 661-672	5.4	42
59	Oral D-galactose supplementation in PGM1-CDG. *Genetics in Medicine*, **2017**, 19, 1226-1235	8.1	41
58	A comprehensive HADHA c.1528G>C frequency study reveals high prevalence of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency in Poland. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S373-7	5.4	40
57	Impact of age at onset and newborn screening on outcome in organic acidurias. *Journal of Inherited Metabolic Disease*, **2016**, 39, 341-353	5.4	39
56	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1 Deficiency. *Journal of Pediatrics*, **2016**, 175, 130-136.e8	3.6	37
55	Pericardial and abdominal fluid accumulation in congenital disorder of glycosylation type Ia. *Molecular Genetics and Metabolism*, **2008**, 94, 481-484	3.7	34
54	SURF1 missense mutations promote a mild Leigh phenotype. *Clinical Genetics*, **2009**, 76, 195-204	4	33
53	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. *Journal of Inherited Metabolic Disease*, **2011**, 34, 923-7	5.4	31
52	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, 61-6	2.5	30
51	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, 101-5	3.2	30
50	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, CR203-9	3.2	29
49	Seventeen novel mutations that cause profound biotinidase deficiency. *Molecular Genetics and Metabolism*, **2002**, 77, 108-11	3.7	29
48	Defining the phenotype in congenital disorder of glycosylation due to ALG1 mutations. *Pediatrics*, **2012**, 130, e1034-9	7.4	28
47	Congenital disorder of glycosylation type Ix: review of clinical spectrum and diagnostic steps. *Journal of Inherited Metabolic Disease*, **2008**, 31, 450-6	5.4	28
46	A homozygous mutation in the SCO2 gene causes a spinal muscular atrophy like presentation with stridor and respiratory insufficiency. *European Journal of Paediatric Neurology*, **2010**, 14, 253-60	3.8	27
45	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. *Genetics in Medicine*, **2020**, 22, 1102-1107	8.1	24
44	Urgent metabolic service improves survival in long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency detected by symptomatic identification and pilot newborn screening. *Journal of Inherited Metabolic Disease*, **2011**, 34, 185-95	5.4	23

43	Light and electron microscopy characteristics of the muscle of patients with SURF1 gene mutations associated with Leigh disease. *Journal of Clinical Pathology*, **2008**, 61, 460-6	3.9	23
42	Clinical, neuroradiological, and biochemical features of SLC35A2-CDG patients. *Journal of Inherited Metabolic Disease*, **2019**, 42, 553-564	5.4	21
41	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. *Annals of Neurology* , **2019**, 86, 116-128	9.4	20
40	Left ventricular noncompaction (LVNC) and low mitochondrial membrane potential are specific for Barth syndrome. *Journal of Inherited Metabolic Disease*, **2013**, 36, 929-37	5.4	20
39	Decreased plasma l-arginine levels in organic acidurias (MMA and PA) and decreased plasma branched-chain amino acid levels in urea cycle disorders as a potential cause of growth retardation: Options for treatment. *Molecular Genetics and Metabolism*, **2019**, 126, 397-405	3.7	18
38	Evaluation of dietary treatment and amino acid supplementation in organic acidurias and urea-cycle disorders: On the basis of information from a European multicenter registry. *Journal of Inherited Metabolic Disease*, **2019**, 42, 1162-1175	5.4	17
37	High prevalence of SURF1 c.845_846delCT mutation in Polish Leigh patients. *European Journal of Paediatric Neurology*, **2009**, 13, 146-53	3.8	17
36	The impact of COVID-19 pandemic on the diagnosis and management of inborn errors of metabolism: A global perspective. *Molecular Genetics and Metabolism*, **2020**, 131, 285-288	3.7	16
35	The natural history of SCO2 deficiency in 36 Polish children confirmed the genotype-phenotype correlation. *Mitochondrion*, **2013**, 13, 810-6	4.9	15
34	Novel cases of D-2-hydroxyglutaric aciduria with IDH1 or IDH2 mosaic mutations identified by amplicon deep sequencing. *Journal of Medical Genetics*, **2013**, 50, 754-9	5.8	15
33	International consensus guidelines for phosphoglucomutase 1 deficiency (PGM1-CDG): Diagnosis, follow-up, and management. *Journal of Inherited Metabolic Disease*, **2021**, 44, 148-163	5.4	15
32	Diversity of clinical symptoms in A3243G mitochondrial DNA mutation (MELAS syndrome mutation). *Medical Science Monitor*, **2002**, 8, CR767-73	3.2	15
31	Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency. *Translational Research*, **2018**, 199, 62-76	11	13
30	Proton MR Spectroscopy in Patients with Leigh Syndrome. *Neuroradiology Journal*, **2011**, 24, 424-8	2	11
29	Sepiapterin reductase deficiency in a 2-year-old girl with incomplete response to treatment during short-term follow-up. *Journal of Inherited Metabolic Disease*, **2009**, 32 Suppl 1, S5-10	5.4	10
28	G8363A mitochondrial DNA mutation is not a rare cause of Leigh syndrome - clinical, biochemical and pathological study of an affected child. *Folia Neuropathologica*, **2007**, 45, 187-91	2.6	9
27	Hypoxanthine-guanine phosphoribosylotransferase deficiency--the spectrum of Polish mutations. *Journal of Inherited Metabolic Disease*, **2008**, 31 Suppl 2, S447-51	5.4	8
26	Novel c.191C>G (p.Pro64Arg) MPV17 mutation identified in two pairs of unrelated Polish siblings with mitochondrial hepatoencephalopathy. *Clinical Genetics*, **2014**, 85, 573-7	4	7

25	The role of genetic factors and pre- and perinatal influences in the etiology of autism spectrum disorders - indications for genetic referral. *Psychiatria Polska*, **2016**, 50, 543-54	1.3	7
24	Evaluation of somatic development in adult patients with previously undiagnosed and/or untreated phenylketonuria. *Medical Principles and Practice*, **2010**, 19, 46-50	2.1	6
23	Deficyt liazy adenylobursztynianowej – diagnostyka i charakterystyka kliniczna 7 polskich pacjentów. *Pediatria Polska*, **2007**, 82, 526-532	0.1	3
22	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. *Nature Communications*, **2021**, 12, 5529	17.4	3
21	Phenotypic features of children with neurodevelopmental diseases in relation to biogenic amines. *Respiratory Physiology and Neurobiology*, **2015**, 209, 124-32	2.8	2
20	Long-term clinical effects of enzyme replacement therapy in MPS II. *Pediatria Polska*, **2017**, 92, 373-377	0.1	2
19	Trial of erythropoietin treatment in a boy with glutathione synthetase deficiency. *Journal of Inherited Metabolic Disease*, **2005**, 28, 1153-4	5.4	2
18	Measurement of functional independence level and falls-risk in individuals with undiagnosed phenylketonuria.. *Acta Biochimica Polonica*, **2009**, 56,	2	2
17	Bridging East with West of Europe – a comparison of orphan drugs policies in Poland, Russia and the Netherlands. *Acta Poloniae Pharmaceutica*, **2018**, 75, 1409-1422	1.3	2
16	Congenital hyperinsulinism in Polish patients--how can we optimize clinical management?. *Endokrynologia Polska*, **2015**, 66, 322-8	1.1	2
15	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders—A successful strategy for clinical research of rare diseases **2019**, 42, 93		2
14	Proteins Structure Models in the Evaluation of Novel Variant (C.472_477del) in the Gene. *Diagnostics*, **2020**, 10,	3.8	2
13	Medical care of patients with disorders of aromatic amino acid metabolism: a report based on the Polish National Health Fund data records. *Pediatric Endocrinology, Diabetes and Metabolism*, **2018**, 2018, 118-125	1	2
12	Age and Gender-Related Changes in Biogenic Amine Metabolites in Cerebrospinal Fluid in Children. *Advances in Experimental Medicine and Biology*, **2016**, 878, 73-82	3.6	1
11	An Investigation of the Neurological and Neuropsychiatric Disturbances in Adults with Undiagnosed and/or Untreated Phenylketonuria in Poland. *Journal of Applied Research in Intellectual Disabilities*, **2011**, 24, 482-488	2.2	1
10	Clinical course and cardiovascular outcomes in patients with the long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency. *Cardiology Journal*, **2017**, 24, 101-104	1.4	1
9	COVID-19 Pandemic and Patients with Rare Inherited Metabolic Disorders and Rare Autoinflammatory Diseases-Organizational Challenges from the Point of View of Healthcare Providers. *Journal of Clinical Medicine*, **2021**, 10,	5.1	1
8	Dietary Treatment from Birth to Pregnancy in a Woman with Methylmalonic Aciduria. *Medicina (Lithuania)*, **2021**, 57,	3.1	1

7	Increasing the spectrum of white matter diseases with tigroid pattern on MRI: glutaric aciduria type 1 - case report. *BMC Pediatrics*, **2021**, 21, 146	2.6	1
6	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. *Molecular Genetics and Metabolism*, **2021**, 133, 397-399	3.7	1
5	Patient's weight can decide about spending millions on enzyme replacement therapy in MPS II. *Molecular Genetics and Metabolism Reports*, **2016**, 6, 5-7	1.8	0
4	Detection of single large-scale mitochondrial DNA deletions by MLPA technique. *Biochimica Et Biophysica Acta - Bioenergetics*, **2010**, 1797, 49-50	4.6	
3	Orphanet Polska w europejskiej sieci jako szansa oceny sytuacji chorób rzadkich na przykładzie wrodzonych wad metabolizmu u dzieci. *Pediatria Polska*, **2008**, 83, 704-711	0.1	
2	Wiotkoœciowa u niemowl¹t wyzwanie diagnostyczne dla pediatry. *Pediatria Polska*, **2008**, 83, 33-38	0.1	
1	Molecular Background and Disease Prevalence of Biotinidase Deficiency in a Polish Population—Data Based on the National Newborn Screening Programme. *Genes*, **2022**, 13, 802	4.2	