

# Jolanta Sykut-Cegielska

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Molecular Background and Disease Prevalence of Biotinidase Deficiency in a Polish Population Data Based on the National Newborn Screening Programme. <i>Genes</i> , <b>2022</b> , 13, 802	4.2	
77	COVID-19 Pandemic and Patients with Rare Inherited Metabolic Disorders and Rare Autoinflammatory Diseases-Organizational Challenges from the Point of View of Healthcare Providers. <i>Journal of Clinical Medicine</i> , <b>2021</b> , 10,	5.1	1
76	International consensus guidelines for phosphoglucomutase 1 deficiency (PGM1-CDG): Diagnosis, follow-up, and management. <i>Journal of Inherited Metabolic Disease</i> , <b>2021</b> , 44, 148-163	5.4	15
75	Dietary Treatment from Birth to Pregnancy in a Woman with Methylmalonic Aciduria. <i>Medicina (Lithuania)</i> , <b>2021</b> , 57,	3.1	1
74	Increasing the spectrum of white matter diseases with tigroid pattern on MRI: glutaric aciduria type 1 - case report. <i>BMC Pediatrics</i> , <b>2021</b> , 21, 146	2.6	1
73	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. <i>Molecular Genetics and Metabolism</i> , <b>2021</b> , 133, 397-399	3.7	1
72	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. <i>Nature Communications</i> , <b>2021</b> , 12, 5529	17.4	3
71	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 1102-1107	8.1	24
70	The impact of COVID-19 pandemic on the diagnosis and management of inborn errors of metabolism: A global perspective. <i>Molecular Genetics and Metabolism</i> , <b>2020</b> , 131, 285-288	3.7	16
69	Proteins Structure Models in the Evaluation of Novel Variant (C.472_477del) in the Gene. <i>Diagnostics</i> , <b>2020</b> , 10,	3.8	2
68	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. <i>Annals of Neurology</i> , <b>2019</b> , 86, 116-128	9.4	20
67	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. <i>Journal of Inherited Metabolic Disease</i> , <b>2019</b> , 42, 1162-1175	5.4	17
66	International clinical guidelines for the management of phosphomannomutase 2-congenital disorders of glycosylation: Diagnosis, treatment and follow up. <i>Journal of Inherited Metabolic Disease</i> , <b>2019</b> , 42, 5-28	5.4	45
65	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders A successful strategy for clinical research of rare diseases <b>2019</b> , 42, 93		2
64	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. <i>Molecular Genetics and Metabolism</i> , <b>2019</b> , 126, 397-405	3.7	18
63	Clinical, neuroradiological, and biochemical features of SLC35A2-CDG patients. <i>Journal of Inherited Metabolic Disease</i> , <b>2019</b> , 42, 553-564	5.4	21
62	Bridging East with West of Europe A comparison of orphan drugs policies in Poland, Russia and the Netherlands. <i>Acta Poloniae Pharmaceutica</i> , <b>2018</b> , 75, 1409-1422	1.3	2

61	Medical care of patients with disorders of aromatic amino acid metabolism: a report based on the Polish National Health Fund data records. <i>Pediatric Endocrinology, Diabetes and Metabolism</i> , <b>2018</b> , 2018, 118-125	1	2
60	Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency. <i>Translational Research</i> , <b>2018</b> , 199, 62-76	11	13
59	Oral D-galactose supplementation in PGM1-CDG. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 1226-1235	8.1	41
58	Long-term clinical effects of enzyme replacement therapy in MPS II. <i>Pediatrica Polska</i> , <b>2017</b> , 92, 373-377	0.1	2
57	Clinical course and cardiovascular outcomes in patients with the long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency. <i>Cardiology Journal</i> , <b>2017</b> , 24, 101-104	1.4	1
56	Age and Gender-Related Changes in Biogenic Amine Metabolites in Cerebrospinal Fluid in Children. <i>Advances in Experimental Medicine and Biology</i> , <b>2016</b> , 878, 73-82	3.6	1
55	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. <i>Nature Communications</i> , <b>2016</b> , 7, 11600	17.4	83
54	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1 Deficiency. <i>Journal of Pediatrics</i> , <b>2016</b> , 175, 130-136.e8	3.6	37
53	The role of genetic factors and pre- and perinatal influences in the etiology of autism spectrum disorders - indications for genetic referral. <i>Psychiatria Polska</i> , <b>2016</b> , 50, 543-54	1.3	7
52	Impact of age at onset and newborn screening on outcome in organic acidurias. <i>Journal of Inherited Metabolic Disease</i> , <b>2016</b> , 39, 341-353	5.4	39
51	Patient's weight can decide about spending millions on enzyme replacement therapy in MPS II. <i>Molecular Genetics and Metabolism Reports</i> , <b>2016</b> , 6, 5-7	1.8	0
50	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , <b>2016</b> , 39, 661-672	5.4	42
49	Phenotypic features of children with neurodevelopmental diseases in relation to biogenic amines. <i>Respiratory Physiology and Neurobiology</i> , <b>2015</b> , 209, 124-32	2.8	2
48	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. <i>Journal of Inherited Metabolic Disease</i> , <b>2015</b> , 38, 1041-57	5.4	143
47	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. <i>Journal of Inherited Metabolic Disease</i> , <b>2015</b> , 38, 1059-74	5.4	135
46	Clinical Features of Lysosomal Acid Lipase Deficiency. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , <b>2015</b> , 61, 619-25	2.8	73
45	Congenital hyperinsulinism in Polish patients--how can we optimize clinical management?. <i>Endokrynologia Polska</i> , <b>2015</b> , 66, 322-8	1.1	2
44	Novel c.191C>G (p.Pro64Arg) MPV17 mutation identified in two pairs of unrelated Polish siblings with mitochondrial hepatoencephalopathy. <i>Clinical Genetics</i> , <b>2014</b> , 85, 573-7	4	7

43	Multiple phenotypes in phosphoglucomutase 1 deficiency. <i>New England Journal of Medicine</i> , <b>2014</b> , 370, 533-42	59.2	197
42	The natural history of SCO2 deficiency in 36 Polish children confirmed the genotype-phenotype correlation. <i>Mitochondrion</i> , <b>2013</b> , 13, 810-6	4.9	15
41	Left ventricular noncompaction (LVNC) and low mitochondrial membrane potential are specific for Barth syndrome. <i>Journal of Inherited Metabolic Disease</i> , <b>2013</b> , 36, 929-37	5.4	20
40	Novel cases of D-2-hydroxyglutaric aciduria with IDH1 or IDH2 mosaic mutations identified by amplicon deep sequencing. <i>Journal of Medical Genetics</i> , <b>2013</b> , 50, 754-9	5.8	15
39	Sepiapterin reductase deficiency: a treatable mimic of cerebral palsy. <i>Annals of Neurology</i> , <b>2012</b> , 71, 520-30	9.4	102
38	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 4151-61	5.6	126
37	Defining the phenotype in congenital disorder of glycosylation due to ALG1 mutations. <i>Pediatrics</i> , <b>2012</b> , 130, e1034-9	7.4	28
36	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> , <b>2011</b> , 17, CR203-9	3.2	29
35	Proton MR Spectroscopy in Patients with Leigh Syndrome. <i>Neuroradiology Journal</i> , <b>2011</b> , 24, 424-8	2	11
34	An Investigation of the Neurological and Neuropsychiatric Disturbances in Adults with Undiagnosed and/or Untreated Phenylketonuria in Poland. <i>Journal of Applied Research in Intellectual Disabilities</i> , <b>2011</b> , 24, 482-488	2.2	1
33	Urgent metabolic service improves survival in long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency detected by symptomatic identification and pilot newborn screening. <i>Journal of Inherited Metabolic Disease</i> , <b>2011</b> , 34, 185-95	5.4	23
32	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. <i>Journal of Inherited Metabolic Disease</i> , <b>2011</b> , 34, 923-7	5.4	31
31	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> , <b>2011</b> , 52, 61-6	2.5	30
30	Evaluation of somatic development in adult patients with previously undiagnosed and/or untreated phenylketonuria. <i>Medical Principles and Practice</i> , <b>2010</b> , 19, 46-50	2.1	6
29	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. <i>Brain</i> , <b>2010</b> , 133, 3210-20	11.2	76
28	A comprehensive HADHA 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> , <b>2010</b> , 33 Suppl 3, S373-7	5.4	40
27	SRD5A3 is required for converting polyprenol to dolichol and is mutated in a congenital glycosylation disorder. <i>Cell</i> , <b>2010</b> , 142, 203-17	56.2	207
26	Cross-border healthcare? The Polish experience. <i>Orphanet Journal of Rare Diseases</i> , <b>2010</b> , 5,	4.2	78

25	A homozygous mutation in the SCO2 gene causes a spinal muscular atrophy like presentation with stridor and respiratory insufficiency. <i>European Journal of Paediatric Neurology</i> , <b>2010</b> , 14, 253-60	3.8	27
24	Detection of single large-scale mitochondrial DNA deletions by MLPA technique. <i>Biochimica Et Biophysica Acta - Bioenergetics</i> , <b>2010</b> , 1797, 49-50	4.6	
23	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. <i>Human Mutation</i> , <b>2010</b> , 31, 380-90	4.7	80
22	Ophthalmological abnormalities in children with congenital disorders of glycosylation type I. <i>British Journal of Ophthalmology</i> , <b>2009</b> , 93, 350-4	5.5	48
21	High prevalence of SURF1 c.845_846delCT mutation in Polish Leigh patients. <i>European Journal of Paediatric Neurology</i> , <b>2009</b> , 13, 146-53	3.8	17
20	SURF1 missense mutations promote a mild Leigh phenotype. <i>Clinical Genetics</i> , <b>2009</b> , 76, 195-204	4	33
19	Sepiapterin reductase deficiency in a 2-year-old girl with incomplete response to treatment during short-term follow-up. <i>Journal of Inherited Metabolic Disease</i> , <b>2009</b> , 32 Suppl 1, S5-10	5.4	10
18	Measurement of functional independence level and falls-risk in individuals with undiagnosed phenylketonuria.. <i>Acta Biochimica Polonica</i> , <b>2009</b> , 56,	2	2
17	Defining the phenotype in an autosomal recessive cutis laxa syndrome with a combined congenital defect of glycosylation. <i>European Journal of Human Genetics</i> , <b>2008</b> , 16, 28-35	5.3	61
16	Clinical, biochemical and molecular findings in seven Polish patients with adenylosuccinate lyase deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2008</b> , 94, 435-442	3.7	51
15	Pericardial and abdominal fluid accumulation in congenital disorder of glycosylation type Ia. <i>Molecular Genetics and Metabolism</i> , <b>2008</b> , 94, 481-484	3.7	34
14	Orphanet Polska w europejskiej sieci jako szansa oceny sytuacji chorób rzadkich na przykładzie wrodzonych wad metabolizmu u dzieci. <i>Pediatrics Polska</i> , <b>2008</b> , 83, 704-711	0.1	
13	Wiotkość mięśniowa u niemowląt - wyzwanie diagnostyczne dla pediatri. <i>Pediatrics Polska</i> , <b>2008</b> , 83, 33-38	0.1	
12	Light and electron microscopy characteristics of the muscle of patients with SURF1 gene mutations associated with Leigh disease. <i>Journal of Clinical Pathology</i> , <b>2008</b> , 61, 460-6	3.9	23
11	Congenital disorder of glycosylation type Ix: review of clinical spectrum and diagnostic steps. <i>Journal of Inherited Metabolic Disease</i> , <b>2008</b> , 31, 450-6	5.4	28
10	Hypoxanthine-guanine phosphoribosyltransferase deficiency--the spectrum of Polish mutations. <i>Journal of Inherited Metabolic Disease</i> , <b>2008</b> , 31 Suppl 2, S447-51	5.4	8
9	Deficyt liazy adenylobursztynianowej - diagnostyka i charakterystyka kliniczna 7 polskich pacjentów. <i>Pediatrics Polska</i> , <b>2007</b> , 82, 526-532	0.1	3
8	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> , <b>2007</b> , 62, 101-5	3.2	30

7	G8363A mitochondrial DNA mutation is not a rare cause of Leigh syndrome - clinical, biochemical and pathological study of an affected child. <i>Folia Neuropathologica</i> , <b>2007</b> , 45, 187-91	2.6	9
6	Retrospective, multicentric study of 180 children with cytochrome C oxidase deficiency. <i>Pediatric Research</i> , <b>2006</b> , 59, 21-6	3.2	125
5	Trial of erythropoietin treatment in a boy with glutathione synthetase deficiency. <i>Journal of Inherited Metabolic Disease</i> , <b>2005</b> , 28, 1153-4	5.4	2
4	Seventeen novel mutations that cause profound biotinidase deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2002</b> , 77, 108-11	3.7	29
3	Diversity of clinical symptoms in A3243G mitochondrial DNA mutation (MELAS syndrome mutation). <i>Medical Science Monitor</i> , <b>2002</b> , 8, CR767-73	3.2	15
2	Clinical ascertainment of Nijmegen breakage syndrome (NBS) and prevalence of the major mutation, 657del5, in three Slav populations. <i>European Journal of Human Genetics</i> , <b>2000</b> , 8, 900-2	5.3	109
1	Partial biotinidase deficiency is usually due to the D444H mutation in the biotinidase gene. <i>Human Genetics</i> , <b>1998</b> , 102, 571-5	6.3	67