Jolanta Sykut-Cegielska

List of Publications by Year
in descending order

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79 papers	3,338 citations	147566 31 h-index	149479 56 g-index
86 all docs	86 docs citations	86 times ranked	4429 citing authors

#	Article	IF	Citations
1	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	13.5	253
2	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542.	13.9	236
3	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. Journal of Inherited Metabolic Disease, 2015, 38, 1041-1057.	1.7	186
4	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074.	1.7	175
5	Gene identification in the congenital disorders of glycosylation type I by whole-exome sequencing. Human Molecular Genetics, 2012, 21, 4151-4161.	1.4	147
6	Retrospective, Multicentric Study of 180 Children with Cytochrome c Oxidase Deficiency. Pediatric Research, 2006, 59, 21-26.	1.1	142
7	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, 900-902.	1.4	130
8	Sepiapterin reductase deficiency: A Treatable Mimic of Cerebral Palsy. Annals of Neurology, 2012, 71, 520-530.	2.8	125
9	ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. Nature Communications, 2016, 7, 11600.	5.8	110
10	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. Human Mutation, 2010, 31, 380-390.	1.1	108
11	Clinical Features of Lysosomal Acid Lipase Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2015, 61, 619-625.	0.9	92
12	International clinical guidelines for the management of phosphomannomutase 2â€congenital disorders of glycosylation: Diagnosis, treatment and follow up. Journal of Inherited Metabolic Disease, 2019, 42, 5-28.	1.7	91
13	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. Brain, 2010, 133, 3210-3220.	3.7	87
14	Partial biotinidase deficiency is usually due to the D444H mutation in the biotinidase gene. Human Genetics, 1998, 102, 571-575.	1.8	78
15	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.	1.4	67
16	Clinical, biochemical and molecular findings in seven Polish patients with adenylosuccinate lyase deficiency. Molecular Genetics and Metabolism, 2008, 94, 435-442.	0.5	62
17	Impact of age at onset and newborn screening on outcome in organic acidurias. Journal of Inherited Metabolic Disease, 2016, 39, 341-353.	1.7	60
18	Ophthalmological abnormalities in children with congenital disorders of glycosylation type I. British Journal of Ophthalmology, 2009, 93, 350-354.	2.1	56

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19	Oral D-galactose supplementation in PGM1-CDG. Genetics in Medicine, 2017, 19, 1226-1235.	1.1	55
20	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.	1.7	52
21	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. Journal of Inherited Metabolic Disease, 2011, 34, 923-927.	1.7	50
22	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
23	Pericardial and abdominal fluid accumulation in Congenital Disorder of Glycosylation type Ia. Molecular Genetics and Metabolism, 2008, 94, 481-484.	0.5	43
24	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1ÂDeficiency. Journal of Pediatrics, 2016, 175, 130-136.e8.	0.9	43
25	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	2.8	42
26	Clinical and biochemical improvement with galactose supplementation in SLC35A2-CDG. Genetics in Medicine, 2020, 22, 1102-1107.	1.1	42
27	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
28	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
29	SURF1 missense mutations promote a mild Leigh phenotype. Clinical Genetics, 2009, 76, 195-204.	1.0	37
30	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
31	Defining the Phenotype in Congenital Disorder of Glycosylation Due to ALG1 Mutations. Pediatrics, 2012, 130, e1034-e1039.	1.0	35
32	Seventeen novel mutations that cause profound biotinidase deficiency. Molecular Genetics and Metabolism, 2002, 77, 108-111.	0.5	33
33	Congenital disorder of glycosylation type Ix: Review of clinical spectrum and diagnostic steps. Journal of Inherited Metabolic Disease, 2008, 31, 450-456.	1.7	32
34	Clinical, neuroradiological, and biochemical features of SLC35A2â€CDG patients. Journal of Inherited Metabolic Disease, 2019, 42, 553-564.	1.7	32
35	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.	0.5	31
36	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

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37	Evaluation of dietary treatment and amino acid supplementation in organic acidurias and ureaâ€cycle disorders: On the basis of information from a European multicenter registry. Journal of Inherited Metabolic Disease, 2019, 42, 1162-1175.	1.7	30
38	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, 253-260.	0.7	28
39	International consensus guidelines for phosphoglucomutase 1 deficiency (<scp>PGM1â€CDG</scp>): Diagnosis, followâ€up, and management. Journal of Inherited Metabolic Disease, 2021, 44, 148-163.	1.7	27
40	Light and electron microscopy characteristics of the muscle of patients withSURF1gene mutations associated with Leigh disease. Journal of Clinical Pathology, 2008, 61, 460-466.	1.0	26
41	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.	0.5	26
42	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
43	Intact transferrin and total plasma glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase-I deficiency. Translational Research, 2018, 199, 62-76.	2.2	22
44	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	5.8	21
45	The natural history of SCO2 deficiency in 36 Polish children confirmed the genotypeâ€“phenotype correlation. Mitochondrion, 2013, 13, 810-816.	1.6	19
46	Novel cases of D-2-hydroxyglutaric aciduria withIDH1orIDH2mosaic mutations identified by amplicon deep sequencing. Journal of Medical Genetics, 2013, 50, 754-759.	1.5	19
47	Diversity of clinical symptoms in A3243G mitochondrial DNA mutation (MELAS syndrome mutation). Medical Science Monitor, 2002, 8, CR767-73.	0.5	18
48	High prevalence of SURF1 c.845_846delCT mutation in Polish Leigh patients. European Journal of Paediatric Neurology, 2009, 13, 146-153.	0.7	17
49	Sepiapterin reductase deficiency in a 2â€yearâ€old girl with incomplete response to treatment during shortâ€term followâ€up. Journal of Inherited Metabolic Disease, 2009, 32, 5-10.	1.7	14
50	Proton MR Spectroscopy in Patients with Leigh Syndrome. Neuroradiology Journal, 2011, 24, 424-428.	0.6	11
51	The role of genetic factors and pre- and perinatal influences in the etiology of autism spectrum disorders â€“ indications for genetic referral. Psychiatria Polska, 2016, 50, 543-554.	0.2	11
52	Hypoxanthine-guanine phosphoribosylotransferase deficiencyâ€”The spectrum of Polish mutations. Journal of Inherited Metabolic Disease, 2008, 31, 447-451.	1.7	9
53	COVID-19 Pandemic and Patients with Rare Inherited Metabolic Disorders and Rare Autoinflammatory Diseasesâ€”Organizational Challenges from the Point of View of Healthcare Providers. Journal of Clinical Medicine, 2021, 10, 4862.	1.0	9
54	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.	0.5	9

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55	Evaluation of Somatic Development in Adult Patients with Previously Undiagnosed and/or Untreated Phenylketonuria. Medical Principles and Practice, 2010, 19, 46-50.	1.1	7
56	Novel c.<scp>191C</scp>>G (p.<scp>Pro64Arg</scp>) <scp>MPV17</scp> mutation identified in two pairs of unrelated Polish siblings with mitochondrial hepatoencephalopathy. Clinical Genetics, 2014, 85, 573-577.	1.0	7
57	Increasing the spectrum of white matter diseases with tigroid pattern on MRI: glutaric aciduria type 1 â€“ case report. BMC Pediatrics, 2021, 21, 146.	0.7	4
58	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”a successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93.	1.7	4
59	Trial of erythropoietin treatment in a boy with glutathione synthetase deficiency. Journal of Inherited Metabolic Disease, 2005, 28, 1153-1154.	1.7	3
60	Deficyt liazy adenylobursztynianowej â€“ diagnostyka i charakterystyka kliniczna 7 polskich pacjentÃ³w. Pediatria Polska, 2007, 82, 526-532.	0.1	3
61	Long-term clinical effects of enzyme replacement therapy in MPS II. Pediatria Polska, 2017, 92, 373-377.	0.1	3
62	Proteins Structure Models in the Evaluation of Novel Variant (C.472_477del) in the MOCS2 Gene. Diagnostics, 2020, 10, 821.	1.3	3
63	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. Molecular Genetics and Metabolism, 2021, 133, 397-399.	0.5	3
64	Measurement of functional independence level and falls-risk in individuals with undiagnosed phenylketonuria.. Acta Biochimica Polonica, 2009, 56, .	0.3	3
65	Molecular Background and Disease Prevalence of Biotinidase Deficiency in a Polish Populationâ€”Data Based on the National Newborn Screening Programme. Genes, 2022, 13, 802.	1.0	3
66	Phenotypic features of children with neurodevelopmental diseases in relation to biogenic amines. Respiratory Physiology and Neurobiology, 2015, 209, 124-132.	0.7	2
67	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, 24, 118-125.	0.3	2
68	Bridging East with West of Europe â€“ a comparison of orphan drugs policies in Poland, Russia and the Netherlands. Acta Poloniae Pharmaceutica, 2018, 75, 1409-1422.	0.3	2
69	Wrodzony hiperinsulinizm â€” prÃ³ba optymalizacji diagnostyki i leczenia u polskich pacjentÃ³w. Endokrynologia Polska, 2015, 66, 322-328.	0.3	2
70	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.	1.3	1
71	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
72	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

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73	Dietary Treatment from Birth to Pregnancy in a Woman with Methylmalonic Aciduria. Medicina (Lithuania), 2021, 57, 128.	0.8	1
74	Clinical course and cardiovascular outcomes in patients with the long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency. Cardiology Journal, 2017, 24, 101-104.	0.5	1
75	PSY26 COST OF TYROSINAEMIA TYPE ONE IN POLAND IN 2006. Value in Health, 2008, 11, A637.	0.1	0
76	Detection of single large-scale mitochondrial DNA deletions by MLPA technique. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 49-50.	0.5	0
77	Cross-border healthcare? The Polish experience. Orphanet Journal of Rare Diseases, 2010, 5, .	1.2	0
78	Dyslipidemia and Sustained Transaminase Elevations from Early Childhood are Common in Lysosomal Acid Lipase Deficiency. Journal of Clinical Lipidology, 2014, 8, 306-307.	0.6	0
79	Dyslipidemia and sustained elevations in transaminases from early childhood are common in lysosomal acid lipase deficiency. Atherosclerosis, 2014, 235, e55-e56.	0.4	0