Anna Tylki-SzymaÅ„ska

List of Publications by Year
in descending order

165 papers	5,812 citations	81900 39 h-index	91884 69 g-index
168 all docs	168 docs citations	168 times ranked	5243 citing authors

#	Article	IF	Citations
1	Therapeutic goals in the treatment of Gaucher disease. Seminars in Hematology, 2004, 41, 4-14.	3.4	418
2	Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. Orphanet Journal of Rare Diseases, 2015, 10, 36.	2.7	239
3	Characterization of Fabry Disease in 352 Pediatric Patients in the Fabry Registry. Pediatric Research, 2008, 64, 550-555.	2.3	235
4	Elevated plasma glucosylsphingosine in Gaucher disease: relation to phenotype, storage cell markers, and therapeutic response. Blood, 2011, 118, e118-e127.	1.4	224
5	Gaucher disease type 1: Revised recommendations on evaluations and monitoring for adult patients. Seminars in Hematology, 2004, 41, 15-22.	3.4	215
6	Mucopolysaccharidosis type II: European recommendations for the diagnosis and multidisciplinary management of a rare disease. Orphanet Journal of Rare Diseases, 2011, 6, 72.	2.7	183
7	Genistein-mediated inhibition of glycosaminoglycan synthesis as a basis for gene expression-targeted isoflavone therapy for mucopolysaccharidoses. European Journal of Human Genetics, 2006, 14, 846-852.	2.8	161
8	Safety and Efficacy of Enzyme Replacement Therapy with Agalsidase Beta: An International, Open-label Study in Pediatric Patients with Fabry Disease. Journal of Pediatrics, 2008, 152, 563-570.e1.	1.8	126
9	European expert consensus statement on therapeutic goals in Fabry disease. Molecular Genetics and Metabolism, 2018, 124, 189-203.	1.1	122
10	Adenylosuccinate lyase deficiency. Journal of Inherited Metabolic Disease, 2015, 38, 231-242.	3.6	119
11	Neuronopathic Gaucher disease: demographic and clinical features of 131 patients enrolled in the International Collaborative Gaucher Group Neurological Outcomes Subregistry. Journal of Inherited Metabolic Disease, 2010, 33, 339-346.	3.6	105
12	Mucopolysaccharidosis type <scp>III</scp> (<scp>S</scp>anfilippo syndrome) and misdiagnosis of idiopathic developmental delay, attention deficit/hyperactivity disorder or autism spectrum disorder. Acta Paediatrica, International Journal of Paediatrics, 2013, 102, 462-470.	1.5	102
13	Genistin-rich soy isoflavone extract in substrate reduction therapy for Sanfilippo syndrome: An open-label, pilot study in 10 pediatric patients. Current Therapeutic Research, 2008, 69, 166-179.	1.2	92
14	Saposin C mutations in Gaucher disease patients resulting in lysosomal lipid accumulation, saposin C deficiency, but normal prosaposin processing and sorting. Human Molecular Genetics, 2010, 19, 2987-2997.	2.9	89
15	Force Majeure: Therapeutic measures in response to restricted supply of imiglucerase (Cerezyme) for patients with Gaucher disease. Blood Cells, Molecules, and Diseases, 2010, 44, 41-47.	1.4	88
16	Nonâ€neuronopathic Gaucher disease due to saposin C deficiency. Clinical Genetics, 2007, 72, 538-542.	2.0	86
17	Idursulfase treatment of Hunter syndrome in children younger than 6 years: Results from the Hunter Outcome Survey. Genetics in Medicine, 2011, 13, 102-109.	2.4	86
18	Management goals for type 1 Gaucher disease: An expert consensus document from the European working group on Gaucher disease. Blood Cells, Molecules, and Diseases, 2018, 68, 203-208.	1.4	82

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19	Thyroid hormone resistance syndrome due to mutations in the thyroid hormone receptor Î± gene (THRA). Journal of Medical Genetics, 2015, 52, 312-316.	3.2	80
20	Enzyme Replacement Therapy for Fabry Disease. Drugs, 2009, 69, 2179-2205.	10.9	79
21	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.	3.2	75
22	Occurrence, distribution, and phenotype of arylsulfatase A mutations in patients with metachromatic leukodystrophy. American Journal of Medical Genetics Part A, 1997, 69, 335-340.	2.4	73
23	Identification of 31 novel mutations in the N-acetylgalactosamine-6-sulfatase gene reveals excessive allelic heterogeneity among patients with Morquio A syndrome. Human Mutation, 1997, 10, 223-232.	2.5	69
24	Update of the pompe disease mutation database with 60 novel GAA sequence variants and additional studies on the functional effect of 34 previously reported variants. Human Mutation, 2012, 33, 1161-1165.	2.5	67
25	IDUA mutational profiling of a cohort of 102 European patients with mucopolysaccharidosis type I: identification and characterization of 35 novel Î±-L-iduronidase (IDUA) alleles. Human Mutation, 2011, 32, E2189-E2210.	2.5	66
26	Clinical, biochemical and molecular findings in seven Polish patients with adenylosuccinate lyase deficiency. Molecular Genetics and Metabolism, 2008, 94, 435-442.	1.1	62
27	Evaluation of a low dose, after a standard therapeutic dose, of agalsidase beta during enzyme replacement therapy in patients with Fabry disease. Genetics in Medicine, 2009, 11, 256-264.	2.4	56
28	Efficacy of recombinant human Î±-L-iduronidase (laronidase) on restricted range of motion of upper extremities in mucopolysaccharidosis typeÂI patients. Journal of Inherited Metabolic Disease, 2010, 33, 151-157.	3.6	54
29	CRIM-negative infantile Pompe disease: characterization of immune responses in patients treated with ERT monotherapy. Genetics in Medicine, 2015, 17, 912-918.	2.4	54
30	Aminoacylase 1 deficiency associated with autistic behavior. Journal of Inherited Metabolic Disease, 2010, 33, 211-214.	3.6	53
31	Two-year follow-up of Sanfilippo Disease patients treated with a genistein-rich isoflavone extract: Assessment of effects on cognitive functions and general status of patients. Medical Science Monitor, 2011, 17, CR196-CR202.	1.1	51
32	Safety and efficacy of velaglucerase alfa in Gaucher disease type 1 patients previously treated with imiglucerase. American Journal of Hematology, 2013, 88, 172-178.	4.1	50
33	The effect of enzyme replacement therapy on clinical outcomes in paediatric patients with Fabry disease â€“ A systematic literature review by a European panel of experts. Molecular Genetics and Metabolism, 2019, 126, 212-223.	1.1	50
34	Ten years of the Hunter Outcome Survey (HOS): insights, achievements, and lessons learned from a global patient registry. Orphanet Journal of Rare Diseases, 2017, 12, 82.	2.7	48
35	Clinical outcomes in idursulfase-treated patients with mucopolysaccharidosis type II: 3-year data from the hunter outcome survey (HOS). Orphanet Journal of Rare Diseases, 2017, 12, 161.	2.7	48
36	Improvement in the range of joint motion in seven patients with mucopolysaccharidosis type II during experimental gene expressionâ€targeted isoflavone therapy (GET IT). American Journal of Medical Genetics, Part A, 2011, 155, 2257-2262.	1.2	46

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37	Long-term hematological, visceral, and growth outcomes in children with Gaucher disease type 3 treated with imiglucerase in the International Collaborative Gaucher Group Gaucher Registry. Molecular Genetics and Metabolism, 2017, 120, 47-56.	1.1	45
38	Female Fabry disease patients and X-chromosome inactivation. Gene, 2018, 641, 259-264.	2.2	44
39	Mitochondrial dysfunction in fibroblasts derived from patients with Niemann-Pick type C disease. Archives of Biochemistry and Biophysics, 2016, 593, 50-59.	3.0	43
40	Characterization of Early Disease Status in Treatment-Naive Male Paediatric Patients with Fabry Disease Enrolled in a Randomized Clinical Trial. PLoS ONE, 2015, 10, e0124987.	2.5	42
41	Cardiovascular manifestations of mucopolysaccharidosis type VI (Maroteauxâ€“Lamy syndrome). International Journal of Cardiology, 2012, 158, 6-11.	1.7	41
42	Correlation between severity of mucopolysaccharidoses and combination of the residual enzyme activity and efficiency of glycosaminoglycan synthesis. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 743-749.	1.5	38
43	Prevalence rates of mucopolysaccharidoses in Poland. Journal of Applied Genetics, 2015, 56, 205-210.	1.9	37
44	Changes in serum chitotriosidase activity with cessation of replacement enzyme (cerebrosidase) administration in Gaucher disease. Clinical Biochemistry, 2000, 33, 147-149.	1.9	35
45	Anthropometric data of 14 patients with mucopolysaccharidosis I: Retrospective analysis and efficacy of recombinant human Î±-l-iduronidase (laronidase). Molecular Genetics and Metabolism, 2010, 99, 10-17.	1.1	35
46	Growth pattern and growth prediction of body height in children with mucopolysaccharidosis type II. Acta Paediatrica, International Journal of Paediatrics, 2011, 100, 456-460.	1.5	35
47	Comprehensive longâ€term efficacy and safety of recombinant human alphaâ€mannosidase (velmanase) Tj ETQq1 1 0.784314 rgBT /Ov 1225-1233.	3.6	35
48	Efficacy and safety of Velmanase alfa in the treatment of patients with alphaâ€mannosidosis: results from the core and extension phase analysis of a phase III multicentre, doubleâ€blind, randomised, placeboâ€controlled trial. Journal of Inherited Metabolic Disease, 2018, 41, 1215-1223.	3.6	34
49	Characteristics of type I Gaucher disease associated with persistent thrombocytopenia after treatment with imiglucerase for 4â€“5Âyears. British Journal of Haematology, 2012, 158, 528-538.	2.5	33
50	Serum chitotriosidase activity in gaucher patients on enzyme replacement therapy (ERT). Clinical Biochemistry, 1998, 31, 417-420.	1.9	31
51	Mutations of the iduronate-2-sulfatase gene in 12 Polish. Patients with mucopolysaccharidosis type II (Hunter syndrome). Human Mutation, 1995, 5, 97-100.	2.5	30
52	Homologous nonallelic recombinations between the iduronate-sulfatase gene and pseudogene cause various intragenic deletions and inversions in patients with mucopolysaccharidosis type II. European Journal of Human Genetics, 1998, 6, 492-500.	2.8	30
53	Characterization of neuronopathic Gaucher disease among ethnic Poles. Genetics in Medicine, 2006, 8, 8-15.	2.4	30
54	Transaldolase deficiency in two new patients with a relative mild phenotype. Molecular Genetics and Metabolism, 2009, 97, 15-17.	1.1	30

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55	Biomarkers for the mucopolysaccharidoses: Discovery and clinical utility. Molecular Genetics and Metabolism, 2012, 106, 395-402.	1.1	30
56	Growth patterns in children with mucopolysaccharidosis I and II. World Journal of Pediatrics, 2015, 11, 226-231.	1.8	30
57	A multicenter, open-label study evaluating safety and clinical outcomes in children (1.4â€“7.5 years) with Hunter syndrome receiving idursulfase enzyme replacement therapy. Genetics in Medicine, 2014, 16, 435-441.	2.4	29
58	Clinical variability in mucolipidosis III (pseudo-Hurler polydystrophy). American Journal of Medical Genetics Part A, 2002, 108, 214-218.	2.4	27
59	Mutations c.459+1G>A and p.P426L in the ARSA gene: Prevalence in metachromatic leukodystrophy patients from European countries. Molecular Genetics and Metabolism, 2005, 86, 353-359.	1.1	27
60	Fourâ€year followâ€up of chronic neuronopathic Gaucher disease in Europeans using a modified severity scoring tool. Journal of Inherited Metabolic Disease, 2011, 34, 1053-1059.	3.6	26
61	Effect of rapid cessation of enzyme replacement therapy: A report of 5 cases and a review of the literature. Molecular Genetics and Metabolism, 2012, 107, 508-512.	1.1	26
62	Molecular analysis of mucopolysaccharidosis type VI in Poland, Belarus, Lithuania and Estonia. Molecular Genetics and Metabolism, 2012, 105, 237-243.	1.1	26
63	Mucopolysaccharidosis type II in females and response to enzyme replacement therapy. American Journal of Medical Genetics, Part A, 2012, 158A, 450-454.	1.2	26
64	Clinical, biochemical, and molecular overview of transaldolase deficiency and evaluation of the endocrine function: Update of 34 patients. Journal of Inherited Metabolic Disease, 2019, 42, 147-158.	3.6	26
65	Cholesterol as a factor regulating intracellular localization of annexin A6 in Niemannâ€“Pick type C human skin fibroblasts. Archives of Biochemistry and Biophysics, 2010, 493, 221-233.	3.0	25
66	Low-dose agalsidase beta treatment in male pediatric patients with Fabry disease: A 5-year randomized controlled trial. Molecular Genetics and Metabolism, 2019, 127, 86-94.	1.1	25
67	Elevated sulfatide excretion in compound heterozygotes of metachromatic leukodystrophy and ASA-pseudodeficiency allele. Clinical Biochemistry, 1997, 30, 325-331.	1.9	23
68	Juvenile onset acid maltase deficiency presenting as a rigid spine syndrome. Neuromuscular Disorders, 2006, 16, 282-285.	0.6	23
69	Abnormalities in the hair morphology of patients with some but not all types of mucopolysaccharidoses. European Journal of Pediatrics, 2008, 167, 203-209.	2.7	23
70	Mucopolysaccharidosis Type VI (Maroteauxâ€“Lamy syndrome) with a predominantly cardiac phenotype. Molecular Genetics and Metabolism, 2011, 104, 695-699.	1.1	23
71	Mucopolysaccharidosis type <scp>VI</scp> in <scp>R</scp>ussia, <scp>K</scp>azakhstan, and Central and Eastern <scp>E</scp>urope. Pediatrics International, 2014, 56, 520-525.	0.5	22
72	Rapid deterioration of a patient with mucopolysaccharidosis type I during interruption of enzyme replacement therapy. American Journal of Medical Genetics, Part A, 2007, 143A, 1925-1927.	1.2	21

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73	Spinal Cord Compression in Maroteaux-Lamy Syndrome: Case Report and Review of the Literature with Effects of Enzyme Replacement Therapy. Pediatric Neurosurgery, 2012, 48, 191-198.	0.7	21
74	Neurologic Presentation, Diagnostics, and Therapeutic Insights in a Severe Case of Adenylosuccinate Lyase Deficiency. Journal of Child Neurology, 2012, 27, 645-649.	1.4	21
75	Magnetic resonance imaging of the brain in adenylosuccinate lyase deficiency: a report of seven cases and a review of the literature. European Journal of Pediatrics, 2012, 171, 131-138.	2.7	21
76	Gaucher disease due to saposin C deficiency, previously described as non-neuronopathic form â€" No positive effects after 2-years of miglustat therapy. Molecular Genetics and Metabolism, 2011, 104, 627-630.	1.1	20
77	Impaired dynamics of the late endosome/lysosome compartment in human Niemannâ€"Pick type C skin fibroblasts carrying mutation in NPC1 gene. Molecular BioSystems, 2012, 8, 1197.	2.9	20
78	Gastroenterological Complications of Anderson-Fabry Disease. Current Pharmaceutical Design, 2013, 19, 6009-6013.	1.9	20
79	Enzyme Replacement Therapy in an Attenuated Case of Mucopolysaccharidosis Type I (Scheie Syndrome): A 6.5-Year Detailed Follow-Up. Pediatric Neurology, 2012, 47, 461-465.	2.1	18
80	Chronic visceral acid sphingomyelinase deficiency (Niemann-Pick disease type B) in 16 Polish patients: long-term follow-up. Orphanet Journal of Rare Diseases, 2019, 14, 55.	2.7	18
81	Nephrological abnormalities in patients with transaldolase deficiency. Nephrology Dialysis Transplantation, 2012, 27, 3224-3227.	0.7	17
82	Restricted joint range of motion in patients with MPS II: correlation with height, age and functional status. Acta Paediatrica, International Journal of Paediatrics, 2012, 101, e183-8.	1.5	17
83	Enzyme replacement therapy: lessons learned and emerging questions. Expert Opinion on Orphan Drugs, 2015, 3, 293-305.	0.8	17
84	A further report of Brachmannâ€de Lange syndrome in two sibs with normal parents. Clinical Genetics, 1995, 47, 324-327.	2.0	16
85	Xanthine dehydrogenase deficiency with novel sequence variations presenting as rheumatoid arthritis in a 78-year-old patient. Journal of Inherited Metabolic Disease, 2010, 33, 21-24.	3.6	16
86	Mucopolysaccharidosis type VI: A predominantly cardiac phenotype associated with homozygosity for p.R152W mutation in the ARSB gene. American Journal of Medical Genetics, Part A, 2013, 161, 1291-1299.	1.2	16
87	Tyrosinemia type III in an asymptomatic girl. Molecular Genetics and Metabolism Reports, 2015, 5, 48-50.	1.1	16
88	Anemia in Patients With Resistance to Thyroid Hormone Î±: A Role for Thyroid Hormone Receptor Î± in Human Erythropoiesis. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 3517-3525.	3.6	16
89	Early onset lysosomal acid lipase deficiency presenting as secondary hemophagocytic lymphohistiocytosis: Two infants treated with sebelipase alfa. Clinics and Research in Hepatology and Gastroenterology, 2018, 42, e77-e82.	1.5	16
90	NGLY1 deficiency: Novel patient, review of the literature and diagnostic algorithm. JIMD Reports, 2020, 51, 82-88.	1.5	16

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91	Precocious Puberty in Three Boys with Sanfilippo A (Mucopolysaccharidosis III A). Journal of Pediatric Endocrinology and Metabolism, 1995, 8, 291-3.	0.9	14
92	Clinical and molecular characteristics of two transaldolase-deficient patients. European Journal of Pediatrics, 2014, 173, 1679-1682.	2.7	14
93	Effect of rapid cessation of enzyme replacement therapy: A report of 5 more cases. Molecular Genetics and Metabolism, 2014, 111, 212-213.	1.1	14
94	Analysis of voice quality in patients with late-onset Pompe disease. Orphanet Journal of Rare Diseases, 2016, 11, 99.	2.7	14
95	The Effect of Recombinant Human Iduronate-2-Sulfatase (Idursulfase) on Growth in Young Patients with Mucopolysaccharidosis Type II. PLoS ONE, 2014, 9, e85074.	2.5	14
96	Mucopolysaccharidosis type VI: A cardiologist's guide to diagnosis and treatment. International Journal of Cardiology, 2013, 167, 1-10.	1.7	13
97	Attenuated Adenylosuccinate Lyase Deficiency: A Report of One Case and a Review of the Literature. Neuropediatrics, 2014, 45, 050-055.	0.6	13
98	Cervical Spine MRI Findings in Patients with Mucopolysaccharidosis Type II. Pediatric Neurosurgery, 2015, 50, 26-30.	0.7	13
99	Plasma chitotriosidase activity versus plasma glucosylsphingosine in wide spectrum of Gaucher disease phenotypes â€“ A statistical insight. Molecular Genetics and Metabolism, 2018, 123, 495-500.	1.1	13
100	Clinical, biochemical and histological analysis of seven patients with cholesteryl ester storage disease. Pediatrics International, 1997, 39, 643-646.	0.5	12
101	Outcomes of oral biotin treatment in patients with biotinidase deficiency â€” Twenty years follow-up. Molecular Genetics and Metabolism Reports, 2015, 5, 33-35.	1.1	12
102	Can Macrosomia or Large for Gestational Age Be Predictive of Mucopolysaccharidosis Type I, II and VI?. Pediatrics and Neonatology, 2016, 57, 181-187.	0.9	12
103	Mild Zellweger syndrome due to a novel PEX6 mutation: correlation between clinical phenotype and in silico prediction of variant pathogenicity. Journal of Applied Genetics, 2017, 58, 475-480.	1.9	12
104	Practical Suggestions in Diagnosing Metachromatic Leukodystrophy in Probands andin Testing Family Members. European Neurology, 1998, 40, 67-70.	1.4	11
105	Attenuated osteoarticular phenotype of type VI mucopolysaccharidosis: a report of four patients and a review of the literature. Clinical Rheumatology, 2014, 33, 725-731.	2.2	11
106	Long-Term Systematic Monitoring of Four Polish Transaldolase Deficient Patients. JIMD Reports, 2018, 42, 79-87.	1.5	11
107	Easyâ€toâ€use algorithm would provide faster diagnoses for mucopolysaccharidosis type I and enable patients to receive earlier treatment. Acta Paediatrica, International Journal of Paediatrics, 2018, 107, 1402-1408.	1.5	11
108	Serum very long-chain fatty acids (VLCFA) levels as predictive biomarkers of diseases severity and probability of survival in peroxisomal disorders. PLoS ONE, 2020, 15, e0238796.	2.5	11

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109	Type III D mucopolysaccharidosis (Sanfilippo D): Clinical course and symptoms. Pediatrics International, 1998, 40, 492-494.	0.5	10
110	Serum hyaluronidase aberrations in metabolic and morphogenetic disorders. Glycoconjugate Journal, 2005, 22, 395-400.	2.7	10
111	Gaucher disease and dysgammaglobulinemia: A report of 61 patients, including 18 with GD type III. Blood Cells, Molecules, and Diseases, 2011, 46, 85-87.	1.4	10
112	Gastrointestinal Phenotype of Fabry Disease in a Patient with Pseudoobstruction Syndrome. JIMD Reports, 2011, 4, 25-28.	1.5	10
113	International working group identifies need for newborn screening for mucopolysaccharidosis type I but states that existing hurdles must be overcome. Acta Paediatrica, International Journal of Paediatrics, 2018, 107, 2059-2065.	1.5	10
114	Neuraminidase deficiency presenting as a nephrosialidosis: The first case detected in Poland. Pediatrics International, 1996, 38, 529-532.	0.5	9
115	Atypical microbial infections of digestive tract may contribute to diarrhea in mucopolysaccharidosis patients: a MPS I case study. BMC Pediatrics, 2005, 5, 9.	1.7	9
116	Recommendations on Reintroduction of Agalsidase Beta for Patients with Fabry Disease in Europe, Following a Period of Shortage. JIMD Reports, 2012, 8, 51-56.	1.5	9
117	Late form of Pompe disease with glycogen storage in peripheral nerves axons. Journal of the Neurological Sciences, 2011, 301, 59-62.	0.6	8
118	Changes in hair morphology as a biomarker in gene expression-targeted isoflavone therapy for Sanfilippo disease. Gene, 2012, 504, 292-295.	2.2	8
119	Source document verification in the Mucopolysaccharidosis Type I Registry. Pharmacoepidemiology and Drug Safety, 2012, 21, 749-752.	1.9	8
120	Voice alterations in patients with Morquio A syndrome. Journal of Applied Genetics, 2018, 59, 73-80.	1.9	8
121	Novel data on growth phenotype and causative genotypes in 29 patients with Morquio (Morquio-Brailsford) syndrome from Central-Eastern Europe. Journal of Applied Genetics, 2019, 60, 163-174.	1.9	8
122	Lipidoses detected in Poland through 1993. Pediatric Neurology, 1994, 11, 295-297.	2.1	7
123	Investigations of micro-organic brain damage (MOBD) in heterozygotes of metachromatic leukodystrophy. American Journal of Medical Genetics Part A, 2002, 110, 315-319.	2.4	7
124	Molecular bases of metachromatic leukodystrophy in Polish patients. Journal of Human Genetics, 2010, 55, 394-396.	2.3	7
125	Interaction of AnxA6 with isolated and artificial lipid microdomains; importance of lipid composition and calcium content. Molecular BioSystems, 2013, 9, 668.	2.9	7
126	Ultrasonographic Features of Hip Joints in Mucopolysaccharidoses Type I and II. PLoS ONE, 2015, 10, e0123792.	2.5	7

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127	Human pulmonary artery endothelial cells in the model of mucopolysaccharidosis VI present a prohypertensive phenotype. Molecular Genetics and Metabolism Reports, 2015, 3, 11-17.	1.1	7
128	Bioimpedance Analysis as a Method to Evaluate the Proportion of Fatty and Muscle Tissues in Progressive Myopathy in Pompe Disease. JIMD Reports, 2015, 26, 45-51.	1.5	7
129	Follow-up analysis of voice quality in patients with late-onset Pompe disease. Orphanet Journal of Rare Diseases, 2018, 13, 189.	2.7	7
130	Diagnostic Algorithm for Cholesteryl Ester Storage Disease. Journal of Pediatric Gastroenterology and Nutrition, 2018, 67, 452-457.	1.8	7
131	Long Term Follow-Up of Polish Patients with Isovaleric Aciduria. Clinical and Molecular Delineation of Isovaleric Aciduria. Diagnostics, 2020, 10, 738.	2.6	7
132	Gaucher disease diagnosed after bone marrow trephine biopsy — a report of two cases. Folia Histochemica Et Cytobiologica, 2011, 49, 352-356.	1.5	7
133	Mild phenotype of glutaric aciduria type 1 in polish patients â€“ novel data from a group of 13 cases. Metabolic Brain Disease, 2019, 34, 641-649.	2.9	6
134	Over 20-Year Follow-up of Patients with Hepatic Glycogen Storage Diseases: Single-Center Experience. Diagnostics, 2020, 10, 297.	2.6	6
135	Prevalence of Arylsulfatase A Pseudodeficiency Allele in Metachromatic Leukodystrophy Patients from Poland. European Neurology, 2000, 44, 104-107.	1.4	5
136	The prevalence and diagnosis of lysosomal storage diseases in Poland. European Journal of Pediatrics, 2001, 160, 261-262.	2.7	5
137	Natural history of Polish patients with mucopolysaccharidosis type VI. Open Medicine (Poland), 2011, 6, 163-171.	1.3	5
138	Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation: High Outcome Variation between Two Siblings. Neuropediatrics, 2014, 45, 188-191.	0.6	5
139	Liver Involvement in Congenital Disorders of Glycosylation and Deglycosylation. Frontiers in Pediatrics, 2021, 9, 696918.	1.9	5
140	Schimke immuno-osseous dysplasia: two cases. Pediatric Radiology, 2003, 33, 216-218.	2.0	4
141	A Twelve-Year Follow-Up Study on a Case of Early-Onset Parkinsonism Preceding Clinical Manifestation of Gaucher Disease. JIMD Reports, 2011, 3, 53-57.	1.5	4
142	Original article Monitoring of very long-chain fatty acids levels in X-linked adrenoleukodystrophy, treated with haematopoietic stem cell transplantation and Lorenzoâ€™s Oil. Folia Neuropathologica, 2014, 2, 159-163.	1.2	4
143	The First Metabolome Analysis in Children with Epilepsy and ALG13-CDG Resulting from c.320A>G Variant. Children, 2021, 8, 251.	1.5	4
144	Quality of life among polish Fabry patients â€” a cross-sectional study quality of life among polish Fabry patients. Open Medicine (Poland), 2011, 6, 741-749.	1.3	3

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145	Apolipoprotein E genotype and LRP1 polymorphisms in patients with different clinical types of metachromatic leukodystrophy. Gene, 2013, 526, 176-181.	2.2	3
146	Gynecomastia in MPS IIIA boys: Related to treatment or precocious puberty?. Molecular Genetics and Metabolism, 2014, 111, 61-62.	1.1	3
147	Monitoring of dipeptidyl peptidase-IV (DPP-IV) activity in patients with mucopolysaccharidoses types I and II on enzyme replacement therapy â€" Results of a pilot study. Clinical Biochemistry, 2016, 49, 458-462.	1.9	3
148	Activation of mammalian terget of rapamycin kinase and glycogen synthase kinaseâ€3Î² accompanies abnormal accumulation of cholesterol in fibroblasts from Niemannâ€Pick type C patients. Journal of Cellular Biochemistry, 2019, 120, 6580-6588.	2.6	3
149	Effect of glycerol trioleate oil milk formula administration on very long chain fatty acid levels and clinical course in a patient with Zellweger syndrome. European Journal of Pediatrics, 1995, 154, 867-867.	2.7	2
150	Infantile sialic acid storage disease (ISSD): Report of the first case detected in Poland. Pediatrics International, 2003, 45, 199-200.	0.5	2
151	Demonstration of glucoseâ€6â€phosphate hydrogen 5 enrichment from deuterated water by transaldolaseâ€mediated exchange alone. Magnetic Resonance in Medicine, 2016, 75, 1781-1786.	3.0	2
152	Newborn presentation of Niemannâ€“Pick disease type C â€“ Difficulties and limitations of diagnostic methods. Pediatrics and Neonatology, 2018, 59, 317-318.	0.9	2
153	Modeling Morquio A Syndrome: An Anthropometric Study of Body Characteristics and Stature. Diagnostics, 2020, 10, 116.	2.6	2
154	Transferrin isoform analysis from dried blood spots and serum samples by gel isoelectric focusing for screening congenital disorders of glycosylation. Acta Biochimica Polonica, 2021, 68, 139-142.	0.5	2
155	A case described as translocation 15;15 revised: maternal 15 UPD, resulting from isochromosome 15, in a PWS patient. European Journal of Medical Genetics, 2005, 48, 207-209.	1.3	1
156	Musculoskeletal manifestations of mucopolysaccharidosis type VI and effects of enzyme replacement therapy. Open Medicine (Poland), 2012, 7, 154-162.	1.3	1
157	Bone metabolism in patients with mucopolysaccharidosis type II. Reumatologia, 2014, 6, 354-361.	1.1	1
158	Do Not Miss the (Genetic) Diagnosis of Gaucher Syndrome: A Narrative Review on Diagnostic Clues and Management in Severe Prenatal and Perinatal-Lethal Sporadic Cases. Journal of Clinical Medicine, 2021, 10, 4890.	2.4	1
159	Structural Analysis of the Effect of Asn107Ser Mutation on Alg13 Activity and Alg13-Alg14 Complex Formation and Expanding the Phenotypic Variability of ALG13-CDG. Biomolecules, 2022, 12, 398.	4.0	1
160	Choroba spichrzania estrÃ³w cholesterolu. Pediatria Polska, 2013, 88, 69-74.	0.2	0
161	Choroba Wolmana. Pediatria Polska, 2013, 88, 64-68.	0.2	0
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