

# Nicola Longo

## List of Publications by Citations

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136  
papers

4,094  
citations

33  
h-index

59  
g-index

152  
ext. papers

4,913  
ext. citations

4.6  
avg, IF

5.5  
L-index

#	Paper	IF	Citations
136	Disorders of carnitine transport and the carnitine cycle. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2006</b> , 142C, 77-85	3.1	320
135	Carnitine transport and fatty acid oxidation. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , <b>2016</b> , 1863, 2422-35	4.9	315
134	Impaired PIEZO1 function in patients with a novel autosomal recessive congenital lymphatic dysplasia. <i>Nature Communications</i> , <b>2015</b> , 6, 8329	17.4	166
133	Mutations in ABCD4 cause a new inborn error of vitamin B12 metabolism. <i>Nature Genetics</i> , <b>2012</b> , 44, 1152-5	36.3	157
132	Efficacy of sapropterin dihydrochloride in increasing phenylalanine tolerance in children with phenylketonuria: a phase III, randomized, double-blind, placebo-controlled study. <i>Journal of Pediatrics</i> , <b>2009</b> , 154, 700-7	3.6	142
131	A Delphi clinical practice protocol for the management of very long chain acyl-CoA dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2009</b> , 96, 85-90	3.7	126
130	Single-dose, subcutaneous recombinant phenylalanine ammonia lyase conjugated with polyethylene glycol in adult patients with phenylketonuria: an open-label, multicentre, phase 1 dose-escalation trial. <i>Lancet, The</i> , <b>2014</b> , 384, 37-44	4.0	109
129	Global Analysis of Plasma Lipids Identifies Liver-Derived Acylcarnitines as a Fuel Source for Brown Fat Thermogenesis. <i>Cell Metabolism</i> , <b>2017</b> , 26, 509-522.e6	24.6	105
128	Genotype-phenotype correlation in inherited severe insulin resistance. <i>Human Molecular Genetics</i> , <b>2002</b> , 11, 1465-75	5.6	104
127	Disorders of bipterin metabolism. <i>Journal of Inherited Metabolic Disease</i> , <b>2009</b> , 32, 333-42	5.4	99
126	Guanidinoacetate methyltransferase (GAMT) deficiency: outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. <i>Molecular Genetics and Metabolism</i> , <b>2014</b> , 111, 16-25	3.7	77
125	Expanded newborn screening identifies maternal primary carnitine deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2007</b> , 90, 441-5	3.7	74
124	Disorders of creatine transport and metabolism. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2011</b> , 157C, 72-8	3.1	71
123	Glutaric acidemia type 1. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2006</b> , 142C, 86-94	3.1	71
122	Primary and secondary alterations of neonatal carnitine metabolism. <i>Seminars in Perinatology</i> , <b>1999</b> , 23, 152-61	3.3	69
121	Metabolic changes associated with hyperammonemia in patients with propionic acidemia. <i>Molecular Genetics and Metabolism</i> , <b>2006</b> , 88, 123-30	3.7	66
120	Phenotype and genotype variation in primary carnitine deficiency. <i>Genetics in Medicine</i> , <b>2001</b> , 3, 387-92	8.1	62

119	Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. <i>Hepatology</i> , <b>2013</b> , 57, 2171-9	11.2	61
118	Functional analysis of mutations in the OCTN2 transporter causing primary carnitine deficiency: lack of genotype-phenotype correlation. <i>Human Mutation</i> , <b>2000</b> , 16, 401-7	4.7	61
117	Cardiomyopathy and carnitine deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2008</b> , 94, 162-6	3.7	53
116	Defective urinary carnitine transport in heterozygotes for primary carnitine deficiency. <i>Genetics in Medicine</i> , <b>1998</b> , 1, 34-9	8.1	52
115	Validation of dye-binding/high-resolution thermal denaturation for the identification of mutations in the SLC22A5 gene. <i>Human Mutation</i> , <b>2005</b> , 25, 306-13	4.7	51
114	Glutaric acidemia type 1: outcomes before and after expanded newborn screening. <i>Molecular Genetics and Metabolism</i> , <b>2012</b> , 106, 430-8	3.7	49
113	A missense mutation in the OCTN2 gene associated with residual carnitine transport activity. <i>Human Mutation</i> , <b>2000</b> , 15, 238-45	4.7	49
112	Genotype-phenotype correlation in primary carnitine deficiency. <i>Human Mutation</i> , <b>2012</b> , 33, 118-23	4.7	45
111	Progressive decline in insulin levels in Rabson-Mendenhall syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>1999</b> , 84, 2623-9	5.6	45
110	Safety and efficacy of velaglucerase alfa in Gaucher disease type 1 patients previously treated with imiglucerase. <i>American Journal of Hematology</i> , <b>2013</b> , 88, 172-8	7.1	43
109	Glycosylation of the OCTN2 carnitine transporter: study of natural mutations identified in patients with primary carnitine deficiency. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2011</b> , 1812, 312-20	6.9	38
108	Primary Carnitine Deficiency and Newborn Screening for Disorders of the Carnitine Cycle. <i>Annals of Nutrition and Metabolism</i> , <b>2016</b> , 68 Suppl 3, 5-9	4.5	38
107	Abnormal sodium stimulation of carnitine transport in primary carnitine deficiency. <i>Journal of Biological Chemistry</i> , <b>2000</b> , 275, 20782-6	5.4	36
106	Pharmacological rescue of carnitine transport in primary carnitine deficiency. <i>Human Mutation</i> , <b>2006</b> , 27, 513-23	4.7	35
105	Functional and molecular studies in primary carnitine deficiency. <i>Human Mutation</i> , <b>2017</b> , 38, 1684-1699	4.7	34
104	Long-term safety and efficacy of sapropterin: the PKUDOS registry experience. <i>Molecular Genetics and Metabolism</i> , <b>2015</b> , 114, 557-63	3.7	32
103	Pegvaliase for the treatment of phenylketonuria: A pivotal, double-blind randomized discontinuation Phase 3 clinical trial. <i>Molecular Genetics and Metabolism</i> , <b>2018</b> , 124, 20-26	3.7	32
102	Feasibility of newborn screening for guanidinoacetate methyltransferase (GAMT) deficiency. <i>Journal of Inherited Metabolic Disease</i> , <b>2014</b> , 37, 231-6	5.4	32

101	Response to therapy in carnitine/acylcarnitine translocase (CACT) deficiency due to a novel missense mutation. <i>American Journal of Medical Genetics Part A</i> , <b>2004</b> , 126A, 150-5		32
100	Biochemical findings in common inborn errors of metabolism. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2006</b> , 142C, 64-76	3.1	31
99	Mitochondrial encephalopathy. <i>Neurologic Clinics</i> , <b>2003</b> , 21, 817-31	4.5	30
98	Correlation of age-specific phenylalanine levels with intellectual outcome in patients with phenylketonuria. <i>Journal of Inherited Metabolic Disease</i> , <b>2011</b> , 34, 963-71	5.4	29
97	Functional characterization of the carnitine transporter defective in primary carnitine deficiency. <i>Archives of Biochemistry and Biophysics</i> , <b>1999</b> , 364, 99-106	4.1	29
96	Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 1851-1867	8.1	29
95	Homozygous nonsense mutation in the MCEE gene and siRNA suppression of methylmalonyl-CoA epimerase expression: a novel cause of mild methylmalonic aciduria. <i>Molecular Genetics and Metabolism</i> , <b>2006</b> , 88, 327-33	3.7	28
94	Increased glucose transport by human fibroblasts with a heritable defect in insulin binding. <i>Metabolism: Clinical and Experimental</i> , <b>1989</b> , 38, 690-7	12.7	27
93	Biochemical abnormalities in Pearson syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167A, 621-8	2.5	26
92	Effect of insulin on the activity of amino acid transport systems in cultured human fibroblasts. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , <b>1985</b> , 844, 216-23	4.9	26
91	Effect of extracellular potassium on amino acid transport and membrane potential in fetal human fibroblasts. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , <b>1986</b> , 854, 240-50	3.8	26
90	Progressive cerebral vascular degeneration with mitochondrial encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , <b>2008</b> , 146A, 361-7	2.5	25
89	Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. <i>Genetics in Medicine</i> , <b>2015</b> , 17, 561-8	8.1	23
88	Functional domains in the carnitine transporter OCTN2, defective in primary carnitine deficiency. <i>Journal of Biological Chemistry</i> , <b>2003</b> , 278, 47776-84	5.4	23
87	Succinyl-CoA:3-ketoacid transferase (SCOT) deficiency in a new patient homozygous for an R217X mutation. <i>Journal of Inherited Metabolic Disease</i> , <b>2004</b> , 27, 691-2	5.4	23
86	6q subtelomeric deletion: is there a recognizable syndrome?. <i>Clinical Dysmorphology</i> , <b>2004</b> , 13, 103-106	0.9	23
85	SLC35A2-CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. <i>Human Mutation</i> , <b>2019</b> , 40, 908-925	4.7	23
84	Creatine transporter deficiency in two half-brothers. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 1979-83	2.5	22

83	Paternal uniparental disomy of chromosome 14: confirmation of a clinically-recognizable phenotype. <i>American Journal of Medical Genetics Part A</i> , <b>2004</b> , 130A, 88-91		22
82	Effect of dietary lysine restriction and arginine supplementation in two patients with pyridoxine-dependent epilepsy. <i>Molecular Genetics and Metabolism</i> , <b>2016</b> , 118, 167-172	3.7	21
81	Primary Carnitine Deficiency Presents Atypically with Long QT Syndrome: A Case Report. <i>JIMD Reports</i> , <b>2012</b> , 2, 87-90	1.9	21
80	Evidence-based treatment of guanidinoacetate methyltransferase (GAMT) deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2013</b> , 110, 255-62	3.7	20
79	Tyrosine residues affecting sodium stimulation of carnitine transport in the OCTN2 carnitine/organic cation transporter. <i>Journal of Biological Chemistry</i> , <b>2004</b> , 279, 7247-53	5.4	20
78	Hyperammonaemia in classic organic acidemias: a review of the literature and two case histories. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 219	4.2	20
77	Anaplerotic therapy in propionic acidemia. <i>Molecular Genetics and Metabolism</i> , <b>2017</b> , 122, 51-59	3.7	18
76	Long-term safety and efficacy of pegvaliase for the treatment of phenylketonuria in adults: combined phase 2 outcomes through PAL-003 extension study. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 108	4.2	17
75	Diagnosis, Treatment, and Clinical Outcome of Patients with Mitochondrial Trifunctional Protein/Long-Chain 3-Hydroxy Acyl-CoA Dehydrogenase Deficiency. <i>JIMD Reports</i> , <b>2017</b> , 31, 63-71	1.9	17
74	Long-term velaglucerase alfa treatment in children with Gaucher disease type 1 naïve to enzyme replacement therapy or previously treated with imiglucerase. <i>Molecular Genetics and Metabolism</i> , <b>2016</b> , 117, 164-71	3.7	16
73	Clinical and biochemical outcome of patients with very long-chain acyl-CoA dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2019</b> , 127, 64-73	3.7	15
72	Biochemical changes and clinical outcomes in 34 patients with classic galactosemia. <i>Journal of Inherited Metabolic Disease</i> , <b>2018</b> , 41, 197-208	5.4	15
71	Recurrent liver failure in a 25-year-old female. <i>Liver Transplantation</i> , <b>2010</b> , 16, 1049-53	4.5	15
70	Insulin stimulates the Na <sup>+</sup> ,K <sup>(+)</sup> -ATPase and the Na <sup>+</sup> /K <sup>+</sup> /Cl <sup>-</sup> cotransporter of human fibroblasts. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , <b>1996</b> , 1281, 38-44	3.8	15
69	Phenylketonuria. <i>Nature Reviews Disease Primers</i> , <b>2021</b> , 7, 36	51.1	15
68	Loss-of-Function Mutations in ELMO2 Cause Intraosseous Vascular Malformation by Impeding RAC1 Signaling. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 299-317	11	15
67	Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to $\epsilon$ -aminoadipic semialdehyde dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , <b>2021</b> , 44, 178-192	5.4	15
66	Long-term developmental progression in infants and young children taking sapropterin for phenylketonuria: a two-year analysis of safety and efficacy. <i>Genetics in Medicine</i> , <b>2015</b> , 17, 365-73	8.1	14

65	Induction, titration, and maintenance dosing regimen in a phase 2 study of pegvaliase for control of blood phenylalanine in adults with phenylketonuria. <i>Molecular Genetics and Metabolism</i> , <b>2018</b> , 125, 217-227	3.7	14
64	Glucose transport by cultured human fibroblasts: regulation by phorbol esters and insulin. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , <b>1992</b> , 1104, 24-30	3.8	14
63	Laboratory diagnosis of creatine deficiency syndromes: a technical standard and guideline of the American College of Medical Genetics and Genomics. <i>Genetics in Medicine</i> , <b>2017</b> , 19, 256-263	8.1	13
62	A novel method for simultaneous quantification of alpha-amino adipic semialdehyde/piperidine-6-carboxylate and pipercolic acid in plasma and urine. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , <b>2016</b> , 1017-1018, 145-152	3.2	13
61	Phase I clinical evaluation of CNSA-001 (sepiapterin), a novel pharmacological treatment for phenylketonuria and tetrahydrobiopterin deficiencies, in healthy volunteers. <i>Molecular Genetics and Metabolism</i> , <b>2019</b> , 126, 406-412	3.7	12
60	Perturbation of Na <sup>+</sup> and K <sup>+</sup> gradients in human fibroblasts incubated in unsupplemented saline solutions. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , <b>1986</b> , 860, 1-8	3.8	12
59	Clinical and biochemical outcomes of patients with medium-chain acyl-CoA dehydrogenase deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2020</b> , 129, 13-19	3.7	12
58	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , <b>2020</b> , 6,	14.3	12
57	Creatine transporter deficiency: Novel mutations and functional studies. <i>Molecular Genetics and Metabolism Reports</i> , <b>2016</b> , 8, 20-3	1.8	12
56	Once- versus twice-daily dosing of eliglustat in adults with Gaucher disease type 1: The Phase 3, randomized, double-blind EDGE trial. <i>Molecular Genetics and Metabolism</i> , <b>2018</b> , 123, 347-356	3.7	11
55	Safety and efficacy of glycerol phenylbutyrate for management of urea cycle disorders in patients aged 2 months to 2 years. <i>Molecular Genetics and Metabolism</i> , <b>2017</b> , 122, 46-53	3.7	11
54	Effects of triheptanoin (UX007) in patients with long-chain fatty acid oxidation disorders: Results from an open-label, long-term extension study. <i>Journal of Inherited Metabolic Disease</i> , <b>2021</b> , 44, 253-263	5.4	11
53	Neonatal Screening for Primary Carnitine Deficiency: Lessons Learned from the Faroe Islands. <i>International Journal of Neonatal Screening</i> , <b>2017</b> , 3, 1	2.6	10
52	Developing a National Registry for conditions identifiable through newborn screening. <i>Genetics in Medicine</i> , <b>2009</b> , 11, 176-82	8.1	10
51	Pegvaliase for the treatment of phenylketonuria: Results of the phase 2 dose-finding studies with long-term follow-up. <i>Molecular Genetics and Metabolism</i> , <b>2020</b> , 130, 239-246	3.7	9
50	Pegvaliase: Immunological profile and recommendations for the clinical management of hypersensitivity reactions in patients with phenylketonuria treated with this enzyme substitution therapy. <i>Molecular Genetics and Metabolism</i> , <b>2019</b> , 128, 84-91	3.7	9
49	A primer on newborn screening. <i>Advances in Neonatal Care</i> , <b>2004</b> , 4, 306-17	2	9
48	Extrapolation of Variant Phase in Mitochondrial Short-Chain Enoyl-CoA Hydratase (ECHS1) Deficiency. <i>JIMD Reports</i> , <b>2019</b> , 43, 103-109	1.9	9

47	Self-reported treatment-associated symptoms among patients with urea cycle disorders participating in glycerol phenylbutyrate clinical trials. <i>Molecular Genetics and Metabolism</i> , <b>2015</b> , 116, 29-34	3.7	8
46	Glycerol phenylbutyrate for the maintenance treatment of patients with deficiencies in enzymes of the urea cycle. <i>Expert Opinion on Orphan Drugs</i> , <b>2017</b> , 5, 999-1010	1.1	8
45	Insulin increases the turnover rate of Na <sup>+</sup> -K <sup>+</sup> -ATPase in human fibroblasts. <i>American Journal of Physiology - Cell Physiology</i> , <b>2001</b> , 280, C912-9	5.4	8
44	Insulin-receptor autophosphorylation and kinase activity are constitutively increased in fibroblasts cultured from a patient with heritable insulin-resistance. <i>Biochemical and Biophysical Research Communications</i> , <b>1990</b> , 167, 1229-34	3.4	8
43	Cytotoxic edema and diffusion restriction as an early pathoradiologic marker in canavan disease: case report and review of the literature. <i>Orphanet Journal of Rare Diseases</i> , <b>2016</b> , 11, 169	4.2	8
42	National Institutes of Health (NIH) review of evidence in phenylalanine hydroxylase deficiency (phenylketonuria) and recommendations/guidelines for therapy from the American College of Medical Genetics (ACMG) and Genetics Metabolic Dietitians International (GMDI). <i>Molecular Genetics and Metabolism</i> , <b>2014</b> , 112, 65-71	3.7	7
41	Creatine Transporter Deficiency: Screening of Males with Neurodevelopmental Disorders and Neurocognitive Characterization of a Case. <i>Journal of Developmental and Behavioral Pediatrics</i> , <b>2016</b> , 37, 322-6	2.4	7
40	Gene expression in human cells with mutant insulin receptors. <i>Biochemical and Biophysical Research Communications</i> , <b>2003</b> , 307, 1013-20	3.4	6
39	Insulin-induced gene 33 mRNA expression in Chinese hamster ovary cells is insulin receptor dependent <b>2000</b> , 77, 432-444		6
38	Prenatal analysis of the insulin receptor gene in a family with leprechaunism. <i>Prenatal Diagnosis</i> , <b>1995</b> , 15, 1070-4	3.2	6
37	Parkes Weber syndrome associated with two somatic pathogenic variants in. <i>Journal of Physical Education and Sports Management</i> , <b>2020</b> , 6,	2.8	6
36	Wide tolerance to amino acids substitutions in the OCTN1 ergothioneine transporter. <i>Biochimica Et Biophysica Acta - General Subjects</i> , <b>2016</b> , 1860, 1334-42	4	5
35	Long-term safety and efficacy of glycerol phenylbutyrate for the management of urea cycle disorder patients. <i>Molecular Genetics and Metabolism</i> , <b>2019</b> , 127, 336-345	3.7	5
34	The effect of valinomycin in fibroblasts from patients with fatty acid oxidation disorders. <i>Biochemical and Biophysical Research Communications</i> , <b>2013</b> , 437, 637-41	3.4	5
33	Newborn screening and inborn errors of metabolism. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2011</b> , 157C, 1-2	3.1	4
32	Role of arginine 86 of the insulin receptor in insulin binding and activation of glucose transport. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , <b>1998</b> , 1402, 86-94	4.9	4
31	Phosphocyclocreatine is the dominant form of cyclocreatine in control and creatine transporter deficiency patient fibroblasts. <i>Pharmacology Research and Perspectives</i> , <b>2019</b> , 7, e00525	3.1	4
30	Pharmacokinetics of glycerol phenylbutyrate in pediatric patients 2 months to 2 years of age with urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , <b>2018</b> , 125, 251-257	3.7	4

29	Exploratory study of the effect of one week of orally administered CNSA-001 (sepiapterin) on CNS levels of tetrahydrobiopterin, dihydrobiopterin and monoamine neurotransmitter metabolites in healthy volunteers. <i>Molecular Genetics and Metabolism Reports</i> , <b>2019</b> , 21, 100500	1.8	3
28	A benefit-risk analysis of pegvaliase for the treatment of phenylketonuria: A study of patientsS preferences. <i>Molecular Genetics and Metabolism Reports</i> , <b>2019</b> , 21, 100507	1.8	3
27	Retrospective analysis of 19 patients with 6-Pyruvoyl Tetrahydropterin Synthase Deficiency: Prolactin levels inversely correlate with growth. <i>Molecular Genetics and Metabolism</i> , <b>2020</b> , 131, 380-389	3.7	3
26	Effect of genotype on galactose-1-phosphate in classic galactosemia patients. <i>Molecular Genetics and Metabolism</i> , <b>2018</b> , 125, 258-265	3.7	3
25	Response to chen et Al.: carnitine uptake defect (primary carnitine deficiency): risk in genotype-phenotype correlation. <i>Human Mutation</i> , <b>2013</b> , 34, 656	4.7	3
24	Glycerol phenylbutyrate efficacy and safety from an open label study in pediatric patients under 2 months of age with urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , <b>2021</b> , 132, 19-26	3.7	3
23	Laboratory evaluation of homocysteine remethylation disorders and classic homocystinuria: Long-term follow-up using a cohort of 123 patients. <i>Clinica Chimica Acta</i> , <b>2020</b> , 509, 126-134	6.2	2
22	Inborn errors of metabolism: new challenges with expanded newborn screening programs. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2006</b> , 142C, 61-3	3.1	2
21	Restriction fragment length polymorphisms of the insulin receptor gene in families with insulin resistance and leprechaunism. <i>American Journal of the Medical Sciences</i> , <b>1989</b> , 298, 366-70	2.2	2
20	Acute Presentation and Management of the Encephalopathic Child With an Undiagnosed Inborn Error of Metabolism. <i>Journal of Emergency Medicine</i> , <b>2019</b> , 56, e5-e8	1.5	2
19	Dietary management and major clinical events in patients with long-chain fatty acid oxidation disorders enrolled in a phase 2 triheptanoin study. <i>Clinical Nutrition ESPEN</i> , <b>2021</b> , 41, 293-298	1.3	2
18	Long-term preservation of intellectual functioning in sapropterin-treated infants and young children with phenylketonuria: A seven-year analysis. <i>Molecular Genetics and Metabolism</i> , <b>2021</b> , 132, 119-127	3.7	2
17	Creatine metabolism in patients with urea cycle disorders. <i>Molecular Genetics and Metabolism Reports</i> , <b>2021</b> , 29, 100791	1.8	2
16	Timing of therapy and neurodevelopmental outcomes in 18 families with pyridoxine-dependent epilepsy.. <i>Molecular Genetics and Metabolism</i> , <b>2022</b> , 135, 350-356	3.7	2
15	Biomarkers for Drug Development in Propionic and Methylmalonic Acidemias.. <i>Journal of Inherited Metabolic Disease</i> , <b>2022</b> ,	5.4	1
14	Newborn Screening and Inborn Errors of Metabolism <b>2012</b> , 2045-2082		1
13	Prospective identification by neonatal screening of patients with guanidinoacetate methyltransferase deficiency. <i>Molecular Genetics and Metabolism</i> , <b>2021</b> , 134, 60-64	3.7	1
12	Noncoding sequence variants define a novel regulatory element in the first intron of the N-acetylglutamate synthase gene. <i>Human Mutation</i> , <b>2021</b> , 42, 1624-1636	4.7	1



11	A non-interventional observational study to identify and validate clinical outcome assessments for adults with phenylketonuria for use in clinical trials. <i>Molecular Genetics and Metabolism Reports</i> , <b>2021</b> , 29, 100810	1.8	o
10	Pharmacokinetic, pharmacodynamic, and immunogenic rationale for optimal dosing of pegvaliase, a PEGylated bacterial enzyme, in adult patients with phenylketonuria. <i>Clinical and Translational Science</i> , <b>2021</b> , 14, 1894-1905	4.9	o
9	Pathophysiology and management of classic galactosemic primary ovarian insufficiency.. <i>Reproduction and Fertility</i> , <b>2021</b> , 2, R67-R84	1.1	o
8	Round Table Discussion. <i>Annals of Nutrition and Metabolism</i> , <b>2016</b> , 68 Suppl 3, 21-23	4.5	
7	Newborn Screening for Metabolic Disorders <b>2012</b> , 163-197		
6	P-14: Mutational analysis of the insulin receptor: Arginine 86 plays an essential role in insulin binding and stimulation of glucose transport. <i>Experimental and Clinical Endocrinology and Diabetes</i> , <b>1996</b> , 104, 78-80	2.3	
5	Glycine transport by cultured human fibroblasts. <i>Biochemical and Biophysical Research Communications</i> , <b>1988</b> , 152, 617-22	3.4	
4	The OCTN2 carnitine transporter and fatty acid oxidation <b>2003</b> , 161-174		
3	Metabolic Lysosomal Enzyme Probes. <i>FASEB Journal</i> , <b>2013</b> , 27, 576.1	0.9	
2	Gene Therapy for Inherited Metabolic Diseases <b>2022</b> , 97-109		
1	Medical and Nutrition Management of Phenylketonuria: Pegvaliase <b>2022</b> , 153-167		