Carlo Dionisi-Vici

List of Publications by Year
in descending order

289	13,811	19657 61	31849 101
papers	citations	h-index	g-index

307	307	307	14029
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Suggested guidelines for the diagnosis and management of urea cycle disorders. Orphanet Journal of Rare Diseases, 2012, 7, 32.	2.7	596
2	Mutations of SURF-1 in Leigh Disease Associated with Cytochrome c Oxidase Deficiency. American Journal of Human Genetics, 1998, 63, 1609-1621.	6.2	504
3	Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130.	2.7	482
4	Clinical Features of 52 Neonates with Hyperinsulinism. New England Journal of Medicine, 1999, 340, 1169-1175.	27.0	308
5	Suggested guidelines for the diagnosis and management of urea cycle disorders: First revision. Journal of Inherited Metabolic Disease, 2019, 42, 1192-1230.	3.6	277
6	Cobalamin C defect: natural history, pathophysiology, and treatment. Journal of Inherited Metabolic Disease, 2011, 34, 127-135.	3.6	242
7	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	21.4	231
8	Lysinuric protein intolerance (LPI): A multi organ disease by far more complex than a classic urea cycle disorder. Molecular Genetics and Metabolism, 2012, 106, 12-17.	1.1	221
9	Methylmalonic and propionic aciduria. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2006, 142C, 104-112.	1.6	214
10	Guidelines for diagnosis and management of the cobalaminâ€related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 21-48.	3.6	206
11	â€˜Classicalâ€™ organic acidurias, propionic aciduria, methylmalonic aciduria and isovaleric aciduria: Long-term outcome and effects of expanded newborn screening using tandem mass spectrometry. Journal of Inherited Metabolic Disease, 2006, 29, 383-389.	3.6	199
12	Inborn errors of metabolism in the Italian pediatric population: A national retrospective survey. Journal of Pediatrics, 2002, 140, 321-329.	1.8	196
13	Ethylmalonic Encephalopathy Is Caused by Mutations in ETHE1, a Gene Encoding a Mitochondrial Matrix Protein. American Journal of Human Genetics, 2004, 74, 239-252.	6.2	192
14	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. Journal of Inherited Metabolic Disease, 2015, 38, 1041-1057.	3.6	186
15	Recommendations for the management of tyrosinaemia type 1. Orphanet Journal of Rare Diseases, 2013, 8, 8.	2.7	182
16	SUCLA2 mutations are associated with mild methylmalonic aciduria, Leigh-like encephalomyopathy, dystonia and deafness. Brain, 2007, 130, 862-874.	7.6	180
17	Impaired Skin Fibroblast Carnitine Uptake in Primary Systemic Carnitine Deficiency Manifested by Childhood Carnitine-Responsive Cardiomyopathy. Pediatric Research, 1990, 28, 247-255.	2.3	179
18	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.	3.6	175

#	Article	IF	Citations
19	Phosphomannose Isomerase Deficiency: A Carbohydrate-Deficient Glycoprotein Syndrome with Hepatic-Intestinal Presentation. American Journal of Human Genetics, 1998, 62, 1535-1539.	6.2	167
20	Extracorporeal dialysis in neonatal hyperammonemia: modalities and prognostic indicators. Pediatric Nephrology, 2001, 16, 862-867.	1.7	167
21	EPI-743 reverses the progression of the pediatric mitochondrial diseaseâ€"Genetically defined Leigh Syndrome. Molecular Genetics and Metabolism, 2012, 107, 383-388.	1.1	163
22	Clinical and molecular features of mitochondrial DNA depletion due to mutations in deoxyguanosine kinase. Human Mutation, 2008, 29, 330-331.	2.5	144
23	The ketogenic diet in children, adolescents and young adults with refractory epilepsy: an Italian multicentric experience. Epilepsy Research, 2002, 48, 221-227.	1.6	134
24	Clinical presentation and outcome in a series of 88 patients with the cblC defect. Journal of Inherited Metabolic Disease, 2014, 37, 831-840.	3.6	133
25	MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. Brain, 2013, 136, 872-881.	7.6	130
26	Fatal infantile liver failure associated with mitochondrial DNA depletion. Journal of Pediatrics, 1992, 121, 896-901.	1.8	123
27	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511.	1.6	119
28	Guidelines for the diagnosis and management of methylmalonic acidaemia and propionic acidaemia: FirstÂrevision. Journal of Inherited Metabolic Disease, 2021, 44, 566-592.	3.6	118
29	Tyrosine hydroxylase deficiency causes progressive encephalopathy and dopa-nonresponsive dystonia. Annals of Neurology, 2003, 54, S56-S65.	5.3	117
30	The Mitochondrial Ornithine Transporter. Journal of Biological Chemistry, 2003, 278, 32778-32783.	3.4	117
31	A Gene on Chromosome 11q23 Coding for a Putative Glucose- 6-Phosphate Translocase Is Mutated in Glycogen-Storage Disease Types Ib and Ic. American Journal of Human Genetics, 1998, 63, 976-983.	6.2	116
32	GM1 gangliosidosis and Morquio B disease: An update on genetic alterations and clinical findings. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 782-790.	3.8	115
33	Novel OCTN2 mutations: No genotypeâ€"phenotype correlations: Early carnitine therapy prevents cardiomyopathy. American Journal of Medical Genetics Part A, 2002, 111, 271-284.	2.4	110
34	Cross-sectional study of 168 patients with hepatorenal tyrosinaemia and implications for clinical practice. Orphanet Journal of Rare Diseases, 2014, 9, 107.	2.7	110
35	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
36	Genotype-Phenotype Relationship in Human ATP6i-Dependent Autosomal Recessive Osteopetrosis. American Journal of Pathology, 2003, 162, 57-68.	3.8	97

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37	Agenesis of the corpus callosum, combined immunodeficiency, bilateral cataract, and hypopigmentation in two brothers. American Journal of Medical Genetics Part A, 1988, 29, 1-8.	2.4	96
38	Plasma Levels of Homocysteine and Cysteine Increased in Pediatric NAFLD and Strongly Correlated with Severity of Liver Damage. International Journal of Molecular Sciences, 2014, 15, 21202-21214.	4.1	84
39	Citrin deficiency, a perplexing global disorder. Molecular Genetics and Metabolism, 2009, 96, 44-49.	1.1	81
40	Mitochondrial trifunctional protein deficiency: A severe fatty acid oxidation disorder with cardiac and neurologic involvement. Journal of Pediatrics, 2003, 142, 684-689.	1.8	80
41	Spectrum of MMACHC mutations in Italian and Portuguese patients with combined methylmalonic aciduria and homocystinuria, cblC type. Molecular Genetics and Metabolism, 2008, 93, 475-480.	1.1	80
42	The T9176G mtDNA mutation severely affects ATP production and results in Leigh syndrome. Neurology, 2001, 56, 687-690.	1.1	79
43	Succinateâ€CoA ligase deficiency due to mutations in SUCLA2 and SUCLG1: phenotype and genotype correlations in 71 patients. Journal of Inherited Metabolic Disease, 2016, 39, 243-252.	3.6	79
44	Diagnosis of sphingolipidoses: a new simultaneous measurement of lysosphingolipids by LC-MS/MS. Clinical Chemistry and Laboratory Medicine, 2017, 55, 403-414.	2.3	78
45	Consensus recommendation for a diagnostic guideline for acid sphingomyelinase deficiency. Genetics in Medicine, 2017, 19, 967-974.	2.4	77
46	Progressive neuropathy and recurrent myoglobinuria in a child with long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency. Journal of Pediatrics, 1991, 118, 744-746.	1.8	74
47	Associations among genotype, clinical phenotype, and intracellular localization of trafficking proteins in ARC syndrome. Human Mutation, 2012, 33, 1656-1664.	2.5	74
48	A new syndrome with ethylmalonic aciduria and normal fatty acid oxidation in fibroblasts. Journal of Pediatrics, 1994, 124, 79-86.	1.8	73
49	Recurrent fatal pulmonary alveolar proteinosis after heart-lung transplantation in a child with lysinuric protein intolerance. Journal of Pediatrics, 2004, 145, 268-272.	1.8	73
50	Mild trifunctional protein deficiency is associated with progressive neuropathy and myopathy and suggests a novel genotype-phenotype correlation.. Journal of Clinical Investigation, 1998, 102, 1193-1199.	8.2	71
51	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. Journal of Inherited Metabolic Disease, 2013, 36, 43-53.	3.6	70
52	New clinical phenotype of branched-chain acyl-CoA oxidation defect. Lancet, The, 1991, 338, 1522-1523.	13.7	69
53	Mutation analysis in 16 patients with mtDNA depletion. Human Mutation, 2003, 21, 453-454.	2.5	69
54	A severe variant of childhood ataxia with central hypomyelination/vanishing white matter leukoencephalopathy related to EIF21B5 mutation. Neurology, 2002, 59, 1966-1968.	1.1	67

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55	Fulminant Leigh syndrome and sudden unexpected death in a family with the T9176C mutation of the mitochondrial ATPase 6 gene. Journal of Inherited Metabolic Disease, 1998, 21, 2-8.	3.6	66
56	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
57	Prediction of outcome in isolated methylmalonic acidurias: combined use of clinical and biochemical parameters. Journal of Inherited Metabolic Disease, 2009, 32, 630-639.	3.6	65
58	The hyperornithinemiaâ€“hyperammonemia-homocitrullinuria syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 29.	2.7	65
59	Liver transplant in ethylmalonic encephalopathy: a new treatment for an otherwise fatal disease. Brain, 2016, 139, 1045-1051.	7.6	65
60	Fatal infantile leukodystrophy. Neurology, 2001, 57, 265-270.	1.1	64
61	Molecular and functional analysis ofSLC25A20 mutations causing carnitine-acylcarnitine translocase deficiency. Human Mutation, 2004, 24, 312-320.	2.5	63
62	Hypoparathyroidism in mitochondrial trifunctional protein deficiency. Journal of Pediatrics, 1996, 129, 159-162.	1.8	62
63	Hyperinsulinism and Hyperammonemia Syndrome: Report of Twelve Unrelated Patients. Pediatric Research, 2001, 50, 353-357.	2.3	62
64	A new simple and rapid LCâ€“ESI-MS/MS method for quantification of plasma oxysterols as dimethylaminobutyrate esters. Its successful use for the diagnosis of Niemannâ€“Pick type C disease. Clinica Chimica Acta, 2014, 437, 93-100.	1.1	62
65	Evaluation of plasma cholestane-3Î²,5Î±,6Î²-triol and 7-ketocholesterol in inherited disorders related to cholesterol metabolism. Journal of Lipid Research, 2016, 57, 361-367.	4.2	60
66	Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. PLoS ONE, 2019, 14, e0214250.	2.5	59
67	Peripheral sensory-motor polyneuropathy, pigmentary retinopathy, and fatal cardiomyopathy in long-chain 3-hydroxy-acyl-CoA dehydrogenase deficiency. European Journal of Pediatrics, 1992, 151, 121-126.	2.7	57
68	Identification of novel mutations in theSLC25A15gene in hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome: A clinical, molecular, and functional study. Human Mutation, 2009, 30, 741-748.	2.5	57
69	Clinical and molecular findings in hyperornithinemia-hyperammonemia-homocitrullinuria syndrome. Neurology, 2001, 57, 911-914.	1.1	56
70	Medical Management and Dialysis Therapy for the Infant With an Inborn Error of Metabolism. Seminars in Nephrology, 2008, 28, 477-480.	1.6	56
71	Recommendations for clinical monitoring of patients with acid sphingomyelinase deficiency (ASMD). Molecular Genetics and Metabolism, 2019, 126, 98-105.	1.1	56
72	Vici syndrome: a review. Orphanet Journal of Rare Diseases, 2016, 11, 21.	2.7	55

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73	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	3.6	55
74	Microheterogeneity of Serum Glycoproteins in Patients with Chronic Alcohol Abuse Compared with Carbohydrate-deficient Glycoprotein Syndrome Type I. Clinical Chemistry, 1999, 45, 1408-1413.	3.2	54
75	Long term follow-up to evaluate the efficacy of miglustat treatment in Italian patients with Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2015, 10, 22.	2.7	54
76	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	3.6	53
77	Immunodeficiency in Vici syndrome: A heterogeneous phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 434-439.	1.2	52
78	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.	3.6	52
79	Clinical pattern, mutations and in vitro residual activity in 33 patients with severe 5, 10 methylenetetrahydrofolate reductase (MTHFR) deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 115-124.	3.6	52
80	TMEM70 deficiency: longâ€term outcome of 48 patients. Journal of Inherited Metabolic Disease, 2015, 38, 417-426.	3.6	51
81	LYRM7mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. Brain, 2016, 139, 782-794.	7.6	51
82	A novel AIFM1 mutation expands the phenotype to an infantile motor neuron disease. European Journal of Human Genetics, 2016, 24, 463-466.	2.8	51
83	Characteristic Acylcarnitine Profiles in Inherited Defects of Peroxisome Biogenesis: A Novel Tool for Screening Diagnosis Using Tandem Mass Spectrometry. Pediatric Research, 2003, 53, 1013-1018.	2.3	50
84	Tyrosine hydroxylase deficiency with severe clinical course: Clinical and biochemical investigations and optimization of therapy. Journal of Pediatrics, 2000, 136, 560-562.	1.8	49
85	Glutathione: A redox signature in monitoring EPI-743 therapy in children with mitochondrial encephalomyopathies. Molecular Genetics and Metabolism, 2013, 109, 208-214.	1.1	49
86	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	6.2	49
87	AP1S1 defect causing MEDNIK syndrome: a new adaptinopathy associated with defective copper metabolism. Annals of the New York Academy of Sciences, 2014, 1314, 55-63.	3.8	48
88	Biochemical and Molecular Genetic Characteristics of the Severe Form of Tyrosine Hydroxylase Deficiency. Clinical Chemistry, 1999, 45, 2073-2078.	3.2	48
89	Wholeâ€exome sequencing detects somatic mutations of IDH1 in metaphyseal chondromatosis with <scp>D</scp>â€2â€hydroxyglutaric aciduria (MCâ€HGA). American Journal of Medical Genetics, Part A, 2011, 155, 2609-2616.	1.2	47
90	Ketogenic diet in early myoclonic encephalopathy due to non ketotic hyperglycinemia. European Journal of Paediatric Neurology, 2012, 16, 509-513.	1.6	47

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91	Phase I/II Trial of Liverâ€“derived Mesenchymal Stem Cells in Pediatric Liverâ€“based Metabolic Disorders: A Prospective, Open Label, Multicenter, Partially Randomized, Safety Study of One Cycle of Heterologous Human Adult Liverâ€“derived Progenitor Cells (HepaStem) in Urea Cycle Disorders and Crigler-Najjar Syndrome Patients. Transplantation, 2019, 103, 1903-1915.	1.0	47
92	Glutathione metabolism in cobalamin deficiency type C (cblC). Journal of Inherited Metabolic Disease, 2014, 37, 125-129.	3.6	46
93	Short-term survival of hyperammonemic neonates treated with dialysis. Pediatric Nephrology, 2015, 30, 839-847.	1.7	46
94	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
95	Dystonia and deafness due to SUCLA2 defect; Clinical course and biochemical markers in 16 children. Mitochondrion, 2009, 9, 438-442.	3.4	45
96	Understanding pyrrolineâ€5â€carboxylate synthetase deficiency: clinical, molecular, functional, and expression studies, structureâ€based analysis, and novel therapy with arginine. Journal of Inherited Metabolic Disease, 2012, 35, 761-776.	3.6	44
97	Cardiomyopathy and multicore myopathy with accumulation of intermediate filaments. European Journal of Pediatrics, 1990, 149, 856-858.	2.7	43
98	Progressive cavitating leukoencephalopathy associated with respiratory chain complex I deficiency and a novel mutation in NDUFS1. Neurogenetics, 2011, 12, 9-17.	1.4	43
99	N-acetylaspartylglutamate in Canavan disease: an adverse effector?. European Journal of Pediatrics, 1999, 158, 406-409.	2.7	42
100	Wolman disease associated with hemophagocytic lymphohistiocytosis: attempts for an explanation. European Journal of Pediatrics, 2014, 173, 1391-1394.	2.7	41
101	The impact of biomarkers analysis in the diagnosis of Niemann-Pick C disease and acid sphingomyelinase deficiency. Clinica Chimica Acta, 2018, 486, 387-394.	1.1	41
102	Allopurinol challenge test in children. Journal of Inherited Metabolic Disease, 1992, 15, 707-712.	3.6	39
103	Ethylmalonic encephalopathy. Journal of Neurology, 2002, 249, 1446-1450.	3.6	39
104	Riboflavin responsive mitochondrial myopathy is a new phenotype of dihydrolipoamide dehydrogenase deficiency. The chaperon-like effect of vitamin B2. Mitochondrion, 2014, 18, 49-57.	3.4	39
105	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. Journal of Neurology, 2017, 264, 102-111.	3.6	38
106	HHH syndrome (hyperornithinaemia, hyperammonaemia, homocitrullinuria), with fulminant hepatitis-like presentation. Journal of Inherited Metabolic Disease, 2006, 29, 186-189.	3.6	37
107	LC-MS/MS Method for Simultaneous Determination on a Dried Blood Spot of Multiple Analytes Relevant for Treatment Monitoring in Patients with Tyrosinemia Type I. Analytical Chemistry, 2012, 84, 1184-1188.	6.5	37
108	De Barsy Syndrome: A genetically heterogeneous autosomal recessive cutis laxa syndrome related to P5CS and PYCR1 dysfunction. American Journal of Medical Genetics, Part A, 2012, 158A, 927-931.	1.2	37

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109	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	3.6	37
110	COG5-CDG: expanding the clinical spectrum. Orphanet Journal of Rare Diseases, 2012, 7, 94.	2.7	36
111	Impaired phagocytosis in macrophages from patients affected by lysinuric protein intolerance. Molecular Genetics and Metabolism, 2012, 105, 585-589.	1.1	35
112	Argininosuccinic aciduria: Recent pathophysiological insights and therapeutic prospects. Journal of Inherited Metabolic Disease, 2019, 42, 1147-1161.	3.6	35
113	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, 93-106.	3.6	35
114	Molecular prenatal diagnosis of 3-hydroxyâ^'3-methylglutaryl coa lyase deficiency. Prenatal Diagnosis, 1995, 15, 725-729.	2.3	34
115	Differential diagnosis of food protein-induced enterocolitis syndrome. Current Opinion in Allergy and Clinical Immunology, 2014, 14, 246-254.	2.3	34
116	The proteome of cblC defect: in vivo elucidation of altered cellular pathways in humans. Journal of Inherited Metabolic Disease, 2015, 38, 969-979.	3.6	34
117	The treatment of juvenile/adult GM1-gangliosidosis with Miglustat may reverse disease progression. Metabolic Brain Disease, 2017, 32, 1529-1536.	2.9	34
118	Liver and/or kidney transplantation in amino and organic acidâ€related inborn errors of metabolism: An overview on European data. Journal of Inherited Metabolic Disease, 2021, 44, 593-605.	3.6	34
119	Congenital disorders of glycosylation presenting as epileptic encephalopathy with migrating partial seizures in infancy. Developmental Medicine and Child Neurology, 2016, 58, 1085-1091.	2.1	33
120	Phenotypic Heterogeneity in Two Unrelated Danon Patients Associated with the SameLAMPâ€2Gene Mutation. Neuropediatrics, 2005, 36, 309-313.	0.6	32
121	A new Caucasian case of neonatal intrahepatic cholestasis caused by citrin deficiency (NICCD): A clinical, molecular, and functional study. Molecular Genetics and Metabolism, 2011, 104, 501-506.	1.1	32
122	Peroxisomal acylâ€CoAâ€oxidase deficiency: Two new cases. American Journal of Medical Genetics, Part A, 2008, 146A, 1676-1681.	1.2	31
123	Persistent pulmonary arterial hypertension in the newborn (PPHN): A frequent manifestation of TMEM70 defective patients. Molecular Genetics and Metabolism, 2014, 111, 353-359.	1.1	31
124	Hypertrophic cardiomyopathy and mtDNA depletion. Successful treatment with heart transplantation. Neuromuscular Disorders, 2002, 12, 56-59.	0.6	30
125	Hypertrophic cardiomyopathy, cataract, developmental delay, lactic acidosis: A novel subtype of 3-methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2006, 29, 546-550.	3.6	30
126	Opposite Clinical Phenotypes of Glucokinase Disease: Description of a Novel Activating Mutation and Contiguous Inactivating Mutations in Human Glucokinase (GCK) Gene. Molecular Endocrinology, 2009, 23, 1983-1989.	3.7	30

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127	Fetal akinesia in metatropic dysplasia: The combined phenotype of chondrodysplasia and neuropathy?. American Journal of Medical Genetics, Part A, 2011, 155, 2860-2864.	1.2	30
128	Cobalamin C Defect Presenting With Isolated Pulmonary Hypertension. Pediatrics, 2013, 132, e248-e251.	2.1	30
129	Atypical Leigh syndrome associated with the D393N mutation in the mitochondrial ND5 subunit. Neurology, 2003, 61, 1017-1018.	1.1	29
130	Tyrosinemia Type 1: Metastatic Hepatoblastoma With a Favorable Outcome. Pediatrics, 2010, 126, e235-e238.	2.1	29
131	Involvement of respiratory muscles in cytoplasmic body myopathy â€" A pathological studyâ€". Brain and Development, 1990, 12, 798-806.	1.1	27
132	Vigabatrin improves paroxysmal dystonia in succinic semialdehyde dehydrogenase deficiency. Neurology, 2007, 68, 1320-1321.	1.1	27
133	Renal Mitochondrial Cytopathies. International Journal of Nephrology, 2011, 2011, 1-10.	1.3	27
134	Successful pregnancy in a woman with mutâ^' methylmalonic acidaemia. Journal of Inherited Metabolic Disease, 2002, 25, 133-134.	3.6	26
135	Therapeutic Drug Monitoring Is a Feasible Tool to Personalize Drug Administration in Neonates Using New Techniques: An Overview on the Pharmacokinetics and Pharmacodynamics in Neonatal Age. International Journal of Molecular Sciences, 2020, 21, 5898.	4.1	26
136	RFT1 deficiency in three novel CDG patients. Human Mutation, 2009, 30, 1428-1434.	2.5	25
137	TMEM70: a mutational hot spot in nuclear ATP synthase deficiency with a pivotal role in complex V biogenesis. Neurogenetics, 2012, 13, 375-386.	1.4	25
138	Early effect of NTBC on renal tubular dysfunction in hereditary tyrosinemia type 1. Molecular Genetics and Metabolism, 2014, 113, 188-193.	1.1	25
139	The hidden Niemann-Pick type C patient: clinical niches for a rare inherited metabolic disease. Current Medical Research and Opinion, 2017, 33, 877-890.	1.9	25
140	ISCA1 mutation in a patient with infantile-onset leukodystrophy causes defects in mitochondrial [4Feâ€"4S] proteins. Human Molecular Genetics, 2018, 27, 2739-2754.	2.9	25
141	The New Horizon of Splitâ€Liver Transplantation: Ex Situ Liver Splitting During Hypothermic Oxygenated Machine Perfusion. Liver Transplantation, 2020, 26, 1363-1367.	2.4	25
142	Acute pancreatitis in propionic acidaemia. Journal of Inherited Metabolic Disease, 1995, 18, 169-172.	3.6	24
143	RFT1â€CDG: Deafness as a novel feature of congenital disorders of glycosylation. Journal of Inherited Metabolic Disease, 2009, 32, 335-338.	3.6	24
144	Expanding the molecular diversity and phenotypic spectrum of glycerol 3â€phosphate dehydrogenase 1 deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 689-695.	3.6	24

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145	A novel mutation in <scp>NDUFB11</scp> unveils a new clinical phenotype associated with lactic acidosis and sideroblastic anemia. Clinical Genetics, 2017, 91, 441-447.	2.0	24
146	Hereditary Spastic Paraplegia Is a Common Phenotypic Finding in ARG1 Deficiency, P5CS Deficiency and HHH Syndrome: Three Inborn Errors of Metabolism Caused by Alteration of an Interconnected Pathway of Glutamate and Urea Cycle Metabolism. Frontiers in Neurology, 2019, 10, 131.	2.4	24
147	Progressive involvement of cardiac conduction system in paediatric patients with Kearnsâ€“Sayre syndrome: how to predict occurrence of complete heart block and sudden cardiac death?. Europace, 2021, 23, 948-957.	1.7	24
148	Intravenous immune globulin in lysinuric protein intolerance. Journal of Inherited Metabolic Disease, 1998, 21, 95-102.	3.6	23
149	Pyroglutamic aciduria and nephropathic cystinosis. Journal of Inherited Metabolic Disease, 1999, 22, 224-226.	3.6	23
150	Allogeneic bone marrow transplantation for infantile globoid-cell leukodystrophy (Krabbe's disease). Pediatric Transplantation, 2002, 6, 427-431.	1.0	23
151	Acute thiamine deficiency and refeeding syndrome: Similar findings but different pathogenesis. Nutrition, 2014, 30, 948-952.	2.4	23
152	The Vici syndrome protein EPG5 regulates intracellular nucleic acid trafficking linking autophagy to innate and adaptive immunity. Autophagy, 2018, 14, 22-37.	9.1	23
153	Delineating the neurological phenotype in children with defects in the <scp>ECHS1</scp> or <scp>HIBCH</scp> gene. Journal of Inherited Metabolic Disease, 2021, 44, 401-414.	3.6	23
154	Genetic disorders of cellular trafficking. Trends in Genetics, 2022, 38, 724-751.	6.7	23
155	Assessment of Visual Function in Children with Methylmalonic Aciduria and Homocystinuria. Neuropediatrics, 2005, 36, 181-185.	0.6	22
156	Lyonization Effects of the t(X;16) Translocation on the Phenotypic Expression in a Rare Female With Menkes Disease. Pediatric Research, 2009, 65, 347-351.	2.3	22
157	Creatine metabolism in urea cycle defects. Journal of Inherited Metabolic Disease, 2012, 35, 647-653.	3.6	22
158	<scp>MALDIâ€MS</scp> profiling of serum Oâ€glycosylation and Nâ€glycosylation in <scp>COG5â€CDG</scp>. Journal of Mass Spectrometry, 2017, 52, 372-377.	1.6	22
159	Increased NO production in lysinuric protein intolerance. Journal of Inherited Metabolic Disease, 2005, 28, 123-129.	3.6	21
160	Genotype-phenotype correlation in PEX5-deficient peroxisome biogenesis defective cell lines. Human Mutation, 2009, 30, 93-98.	2.5	21
161	Congenital hyperinsulinism: Clinical and molecular analysis of a large Italian cohort. Gene, 2013, 521, 160-165.	2.2	21
162	Urine acylcarnitine analysis by ESIâ€“MS/MS: A new tool for the diagnosis of peroxisomal biogenesis disorders. Clinica Chimica Acta, 2008, 398, 86-89.	1.1	20

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163	Cystathionine Î²â€synthase deficiency in the <scp>Eâ€HOD registryâ€part</scp> I: pyridoxine responsiveness as a determinant of biochemical and clinical phenotype at diagnosis. Journal of Inherited Metabolic Disease, 2021, 44, 677-692.	3.6	20
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165	Ketogenic diet in a patient with congenital hyperinsulinism: a novel approach to prevent brain damage. Orphanet Journal of Rare Diseases, 2015, 10, 120.	2.7	19
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