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	Ocular manifestations in patients with inborn errors of intracellular cobalamin metabolism: a systematic review. Human Genetics, 2022, 141, 1239-1251.	3.8	6
2	Diagnosis, treatment, and follow-up of a case of Wolman disease with hemophagocytic lymphohistiocytosis. Molecular Genetics and Metabolism Reports, 2022, 30, 100833.	1.1	2
3	Cardiomyopathies in Children and Systemic Disorders When Is It Useful to Look beyond the Heart?. Journal of Cardiovascular Development and Disease, 2022, 9, 47.	1.6	5
4	Hypoglycaemia Metabolic Gene Panel Testing. Frontiers in Endocrinology, 2022, 13, 826167.	3.5	4
5	Genetic disorders of cellular trafficking. Trends in Genetics, 2022, 38, 724-751.	6.7	23
6	The diagnostic challenge of mild citrulline elevation at newborn screening. Molecular Genetics and Metabolism, 2022, 135, 327-332.	1.1	7
7	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	3.6	5
8	Therapeutic Drug Monitoring of Amphotericin-B in Plasma and Peritoneal Fluid of Pediatric Patients after Liver Transplantation: A Case Series. Antibiotics, 2022, 11, 640.	3.7	3
9	PET/CT in congenital hyperinsulinism: transforming patient's lives by molecular hybrid imaging.. American Journal of Nuclear Medicine and Molecular Imaging, 2022, 12, 44-53.	1.0	0
10	Glycogen storage diseases with liver involvement: a literature review of GSD type 0, IV, VI, IX and XI. Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	12
11	High Incidence of Partial Biotinidase Deficiency in the First 3 Years of a Regional Newborn Screening Program in Italy. International Journal of Environmental Research and Public Health, 2022, 19, 8141.	2.6	4
12	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
13	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
14	Combined proteomic and lipidomic studies in Pompe disease allow a better disease mechanism understanding. Journal of Inherited Metabolic Disease, 2021, 44, 705-717.	3.6	8
15	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
16	A new UHPLC-MS/MS method for the screening of urinary oligosaccharides expands the detection of storage disorders. Orphanet Journal of Rare Diseases, 2021, 16, 24.	2.7	7
17	U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet Journal of Rare Diseases, 2021, 16, 95.	2.7	15
18	Partial Biotinidase Deficiency Revealed Imbalances in Acylcarnitines Profile at Tandem Mass Spectrometry Newborn Screening. International Journal of Environmental Research and Public Health, 2021, 18, 1659.	2.6	1

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19	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
20	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	2.5	12
21	Ethylmalonic encephalopathy and liver transplantation: long-term outcome of the first treated patient. Orphanet Journal of Rare Diseases, 2021, 16, 229.	2.7	7
22	Long-term safety and outcomes in hereditary tyrosinaemia type 1 with nitisinone treatment: a 15-year non-interventional, multicentre study. Lancet Diabetes and Endocrinology,the, 2021, 9, 427-435.	11.4	19
23	ASL expression in ALDH1A1+ neurons in the substantia nigra metabolically contributes to neurodegenerative phenotype. Human Genetics, 2021, 140, 1471-1485.	3.8	10
24	Dysbiosis, Host Metabolism, and Non-communicable Diseases: Trialogue in the Inborn Errors of Metabolism. Frontiers in Physiology, 2021, 12, 716520.	2.8	15
25	Renal Manifestations of Metabolic Disorders in Children. , 2021, , 1-20.		0
26	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
27	A new strategy implementing mass spectrometry in the diagnosis of congenital disorders of N-glycosylation (CDG). Clinical Chemistry and Laboratory Medicine, 2021, 59, 165-171.	2.3	4
28	Ketogenic diet as elective treatment in patients with drug-unresponsive hyperinsulinemic hypoglycemia caused by glucokinase mutations. Orphanet Journal of Rare Diseases, 2021, 16, 424.	2.7	5
29	Myocardial and Arrhythmic Spectrum of Neuromuscular Disorders in Children. Biomolecules, 2021, 11, 1578.	4.0	5
30	Benefits and Toxicity of Disulfiram in Preclinical Models of Nephropathic Cystinosis. Cells, 2021, 10, 3294.	4.1	5
31	Comprehensive-targeted lipidomic analysis in Niemann-Pick C disease. Molecular Genetics and Metabolism, 2021, 134, 337-343.	1.1	6
32	Pharmacokinetic Evaluation of Eltrombopag in ITP Pediatric Patients. Frontiers in Pharmacology, 2021, 12, 772873.	3.5	2
33	Early neurodevelopmental characterization in children with cobalamin C/defect. Journal of Inherited Metabolic Disease, 2020, 43, 367-374.	3.6	9
34	AISF update on the diagnosis and management of adult-onset lysosomal storage diseases with hepatic involvement. Digestive and Liver Disease, 2020, 52, 359-367.	0.9	9
35	Delayed appearance of 3â€methylglutaconic aciduria in neonates with early onset metabolic cardiomyopathies: A potential pitfall for the diagnosis. American Journal of Medical Genetics, Part A, 2020, 182, 64-70.	1.2	4
36	Nocturnal enteral nutrition is therapeutic for growth failure in Fanconiâ€Bickel syndrome. Journal of Inherited Metabolic Disease, 2020, 43, 540-548.	3.6	16

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37	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
38	Clinical relevance of endpoints in clinical trials for acid sphingomyelinase deficiency enzyme replacement therapy. Molecular Genetics and Metabolism, 2020, 131, 116-123.	1.1	18
39	Plasma methylcitric acid and its correlations with other disease biomarkers: The impact in the follow up of patients with propionic and methylmalonic acidemia. Journal of Inherited Metabolic Disease, 2020, 43, 1173-1185.	3.6	19
40	Orofacial features and pediatric dentistry in the long-term management of Infantile Pompe Disease children. Orphanet Journal of Rare Diseases, 2020, 15, 329.	2.7	3
41	Children with special health care needs attending emergency department in Italy: analysis of 3479 cases. Italian Journal of Pediatrics, 2020, 46, 173.	2.6	2
42	Safety of vaccines administration in hereditary fructose intolerance. Orphanet Journal of Rare Diseases, 2020, 15, 274.	2.7	8
43	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
44	The Ketogenic Diet Increases In Vivo Glutathione Levels in Patients with Epilepsy. Metabolites, 2020, 10, 504.	2.9	15
45	The contribution of plasma oxysterols in the challenging diagnostic work-up of infantile cholestasis. Clinica Chimica Acta, 2020, 507, 181-186.	1.1	5
46	A new HPLCâ€“DAD method for contemporary quantification of 10 antibiotics for therapeutic drug monitoring of critically ill pediatric patients. Biomedical Chromatography, 2020, 34, e4880.	1.7	18
47	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
48	CUGC for hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome. European Journal of Human Genetics, 2020, 28, 982-987.	2.8	3
49	CUGC for lysinuric protein intolerance (LPI). European Journal of Human Genetics, 2020, 28, 1129-1134.	2.8	4
50	A False-Positive Case of Methylmalonic Aciduria by Tandem Mass Spectrometry Newborn Screening Dependent on Maternal Malnutrition in Pregnancy. International Journal of Environmental Research and Public Health, 2020, 17, 3601.	2.6	15
51	A Case of Suspected Hyperphenylalaninemia at Newborn Screening by Tandem Mass Spectrometry during Total Parenteral Nutrition. Metabolites, 2020, 10, 44.	2.9	9
52	P5CS expression study in a new family with ALDH18A1â€associated hereditary spastic paraplegia SPG9. Annals of Clinical and Translational Neurology, 2019, 6, 1533-1540.	3.7	14
53	Chronic liver involvement in urea cycle disorders. Journal of Inherited Metabolic Disease, 2019, 42, 1118-1127.	3.6	17
54	Corticospinal tract damage in HHH syndrome: a metabolic cause of hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2019, 14, 208.	2.7	12

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55	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
56	Maple Syrup Urine Disease and Domino Liver Transplantation: When and How?. Liver Transplantation, 2019, 25, 827-828.	2.4	8
57	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
58	Uniparental isodisomy of chromosome 1 results in glycogen storage disease type III with profound growth retardation. Molecular Genetics & Genomic Medicine, 2019, 7, e634.	1.2	7
59	Analysis of LPI-causing mutations on y+LAT1 function and localization. Orphanet Journal of Rare Diseases, 2019, 14, 63.	2.7	6
60	Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. PLoS ONE, 2019, 14, e0214250.	2.5	59
61	Argininosuccinic aciduria: Recent pathophysiological insights and therapeutic prospects. Journal of Inherited Metabolic Disease, 2019, 42, 1147-1161.	3.6	35
62	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	3.6	37
63	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
64	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
65	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
66	Recommendations for clinical monitoring of patients with acid sphingomyelinase deficiency (ASMD). Molecular Genetics and Metabolism, 2019, 126, 98-105.	1.1	56
67	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.	3.6	4
68	The urinary organic acids profile in single large-scale mitochondrial DNA deletion disorders. Clinica Chimica Acta, 2018, 481, 156-160.	1.1	12
69	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
70	ISCA1 mutation in a patient with infantile-onset leukodystrophy causes defects in mitochondrial [4Feâ€“4S] proteins. Human Molecular Genetics, 2018, 27, 3650-3650.	2.9	6
71	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: data from the E-HOD registry. Journal of Inherited Metabolic Disease, 2018, , .	3.6	2
72	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

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73	Persistent Hypoglycemia in Children: Targeted Gene Panel Improves the Diagnosis of Hypoglycemia Due to Inborn Errors of Metabolism. Journal of Pediatrics, 2018, 202, 272-278.e4.	1.8	11
74	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
75	Axonal peripheral neuropathy in propionic acidemia. Neurology, 2018, 91, 565-567.	1.1	9
76	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
77	Novel mutation in mitochondrial Elongation Factor EF-Tu associated to dysplastic leukoencephalopathy and defective mitochondrial DNA translation. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 961-967.	3.8	12
78	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
79	Consensus recommendation for a diagnostic guideline for acid sphingomyelinase deficiency. Genetics in Medicine, 2017, 19, 967-974.	2.4	77
80	<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
81	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
82	3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. Clinica Chimica Acta, 2017, 471, 95-100.	1.1	14
83	The treatment of juvenile/adult GM1-gangliosidosis with Miglustat may reverse disease progression. Metabolic Brain Disease, 2017, 32, 1529-1536.	2.9	34
84	Consensus recommendation on a diagnostic guideline for acid sphingomyelinase deficiency. Molecular Genetics and Metabolism, 2017, 120, S138.	1.1	1
85	NTBC and Correction of Renal Dysfunction. Advances in Experimental Medicine and Biology, 2017, 959, 93-100.	1.6	7
86	Recommendations for the detection and diagnosis of Niemann-Pick disease type C. Neurology: Clinical Practice, 2017, 7, 499-511.	1.6	119
87	Hyperinsulinemic hypoglycemia: clinical, molecular and therapeutical novelties. Journal of Inherited Metabolic Disease, 2017, 40, 531-542.	3.6	18
88	Novel mutations in IBA57 are associated with leukodystrophy and variable clinical phenotypes. Journal of Neurology, 2017, 264, 102-111.	3.6	38
89	Skin and Hair Disorders. , 2017, , 341-370.		1
90	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

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91	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
92	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
93	When silence is noise: infantileâ€onset Barth syndrome caused by a synonymous substitution affecting <scp>TAZ</scp> gene transcription. Clinical Genetics, 2016, 90, 461-465.	2.0	18
94	A new multiplex method for the diagnosis of peroxisomal disorders allowing simultaneous determination of plasma very-long-chain fatty acids, phytanic, pristanic, docosahexaenoic and bile acids by high-performance liquid chromatography-atmospheric pressure chemical ionization-tandem mass spectrometry. Clinica Chimica Acta, 2016, 458, 159-164.	1.1	6
95	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
96	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. American Journal of Human Genetics, 2016, 99, 974-983.	6.2	49
97	Disorders of Ornithine and Proline Metabolism. , 2016, , 321-331.		4
98	MRI and 1H-MRS in adenosine kinase deficiency. Neuroradiology, 2016, 58, 697-703.	2.2	9
99	Vici syndrome: a review. Orphanet Journal of Rare Diseases, 2016, 11, 21.	2.7	55
100	Thiamine Deficiency in a Developed Country. Journal of Parenteral and Enteral Nutrition, 2016, 40, 886-889.	2.6	11
101	Teaching Neuro Images : Galactitol peak and fatal cerebral edema in classic galactosemia. Neurology, 2016, 86, e32-3.	1.1	4
102	Deferasirox-induced serious adverse reaction in a pediatric patient: pharmacokinetic and pharmacogenetic analysis. European Journal of Clinical Pharmacology, 2016, 72, 247-248.	1.9	17
103	LYRM7mutations cause a multifocal cavitating leukoencephalopathy with distinct MRI appearance. Brain, 2016, 139, 782-794.	7.6	51
104	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	3.6	55
105	Liver transplant in ethylmalonic encephalopathy: a new treatment for an otherwise fatal disease. Brain, 2016, 139, 1045-1051.	7.6	65
106	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
107	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
108	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

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109	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
110	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
111	Branched-chain Organic Acidurias/Acidaemias. , 2016, , 277-294.		8
112	Renal Manifestations of Metabolic Disorders in Children. , 2016, , 1569-1607.		0
113	Emergency Treatments. , 2016, , 109-117.		0
114	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
115	Short-term survival of hyperammonemic neonates treated with dialysis. Pediatric Nephrology, 2015, 30, 839-847.	1.7	46
116	Progression of Renal Damage in Glycogen Storage Disease Type I Is Associated to Hyperlipidemia: A Multicenter Prospective Italian Study. Journal of Pediatrics, 2015, 166, 1079-1082.	1.8	15
117	TMEM70 deficiency: longâ€term outcome of 48 patients. Journal of Inherited Metabolic Disease, 2015, 38, 417-426.	3.6	51
118	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
119	Vitamin E Improves Clinical Outcome of Patients Affected by Glycogen Storage Disease Type Ib. JIMD Reports, 2015, 25, 39-45.	1.5	13
120	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
121	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
122	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
123	The hyperornithinemiaâ€“hyperammonemia-homocitrullinuria syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 29.	2.7	65
124	Determination of plasma pipecolic acid by an easy and rapid liquid chromatographyâ€“tandem mass spectrometry method. Clinica Chimica Acta, 2015, 440, 108-112.	1.1	5
125	Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130.	2.7	482
126	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

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127	Clinical utility gene card for: Vici Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	2.8	13
128	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
129	Focal congenital hyperinsulinism managed by medical treatment: a diagnostic algorithm based on molecular genetic screening. Clinical Endocrinology, 2014, 81, 679-688.	2.4	16
130	Immune Tolerance Induced Using Plasma Exchange and Rituximab in an Infantile Pompe Disease Patient. Journal of Child Neurology, 2014, 29, 850-854.	1.4	11
131	Impaired Bone Metabolism in Glycogen Storage Disease Type 1 Is Associated with Poor Metabolic Control in Type 1a and with Granulocyte Colony-Stimulating Factor Therapy in Type 1b. Hormone Research in Paediatrics, 2014, 81, 55-62.	1.8	17
132	Acute thiamine deficiency and refeeding syndrome: Similar findings but different pathogenesis. Nutrition, 2014, 30, 948-952.	2.4	23
133	Gender-related effects on urine l-cystine metastability. Amino Acids, 2014, 46, 415-427.	2.7	6
134	Glutathione metabolism in cobalamin deficiency type C (cblC). Journal of Inherited Metabolic Disease, 2014, 37, 125-129.	3.6	46
135	Early effect of NTBC on renal tubular dysfunction in hereditary tyrosinemia type 1. Molecular Genetics and Metabolism, 2014, 113, 188-193.	1.1	25
136	Fruit-Induced FPIES Masquerading as Hereditary Fructose Intolerance. Pediatrics, 2014, 134, e602-e605.	2.1	11
137	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
138	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
139	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
140	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
141	Measurement of succinyl-carnitine and methylmalonyl-carnitine on dried blood spot by liquid chromatography-tandem mass spectrometry. Clinica Chimica Acta, 2014, 429, 30-33.	1.1	15
142	Wolman disease associated with hemophagocytic lymphohistiocytosis: attempts for an explanation. European Journal of Pediatrics, 2014, 173, 1391-1394.	2.7	41
143	Hyperargininemia: 7-Month Follow-Up Under Sodium Benzoate Therapy in an Italian Child Presenting Progressive Spastic Paraparesis, Cognitive Decline, and Novel Mutation in ARG1 Gene. Pediatric Neurology, 2014, 51, 430-433.	2.1	9
144	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

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145	Differential diagnosis of food protein-induced enterocolitis syndrome. Current Opinion in Allergy and Clinical Immunology, 2014, 14, 246-254.	2.3	34
146	Emergency Diagnostic Procedures and Emergency Treatment. , 2014, , 709-717.		4
147	Renal Manifestations of Metabolic Disorders in Children. , 2014, , 1-43.		0
148	Recommendations for the management of tyrosinaemia type 1. Orphanet Journal of Rare Diseases, 2013, 8, 8.	2.7	182
149	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
150	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
151	Optimizing the dose of hydroxocobalamin in cobalamin C (cblC) defect. Molecular Genetics and Metabolism, 2013, 109, 329-330.	1.1	10
152	Efficacy of Miglustat in Niemannâ€“Pick C disease: A single centre experience. Molecular Genetics and Metabolism, 2013, 110, 329-335.	1.1	17
153	MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. Brain, 2013, 136, 872-881.	7.6	130
154	Congenital hyperinsulinism: Clinical and molecular analysis of a large Italian cohort. Gene, 2013, 521, 160-165.	2.2	21
155	Co-inheritance of two ABCC8 mutations causing an unresponsive congenital hyperinsulinism: Clinical and functional characterization of two novel ABCC8 mutations. Gene, 2013, 516, 122-125.	2.2	11
156	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	21.4	231
157	Cobalamin C Defect Presenting With Isolated Pulmonary Hypertension. Pediatrics, 2013, 132, e248-e251.	2.1	30
158	Emergency Treatments. , 2012, , 103-111.		5
159	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
160	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
161	Glutathione Status in MMACHC Patients. Free Radical Biology and Medicine, 2012, 53, S69-S70.	2.9	0
162	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

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163	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
164	Impaired phagocytosis in macrophages from patients affected by lysinuric protein intolerance. Molecular Genetics and Metabolism, 2012, 105, 585-589.	1.1	35
165	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
166	Pediatric reference intervals for muscle coenzyme Q10. Biomarkers, 2012, 17, 764-766.	1.9	2
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