

Diego Martinelli

List of Publications by Year in descending order

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31
papers

2,792
citations

471509

17
h-index

454955

30
g-index

31
all docs

31
docs citations

31
times ranked

2953
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	Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130.	2.7	482
3	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
4	Cobalamin C defect: natural history, pathophysiology, and treatment. Journal of Inherited Metabolic Disease, 2011, 34, 127-135.	3.6	242
5	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
6	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
7	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
8	MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. Brain, 2013, 136, 872-881.	7.6	130
9	Guidelines for the diagnosis and management of methylmalonic acidemia and propionic acidemia: First revision. Journal of Inherited Metabolic Disease, 2021, 44, 566-592.	3.6	118
10	The hyperornithinemia "hyperammonemia-homocitrullinuria syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 29.	2.7	65
11	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. Journal of Inherited Metabolic Disease, 2011, 34, 923-927.	3.6	50
12	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
13	Understanding pyrroline-5-carboxylate synthetase deficiency: clinical, molecular, functional, and expression studies, structure-based analysis, and novel therapy with arginine. Journal of Inherited Metabolic Disease, 2012, 35, 761-776.	3.6	44
14	Impaired phagocytosis in macrophages from patients affected by lysinuric protein intolerance. Molecular Genetics and Metabolism, 2012, 105, 585-589.	1.1	35
15	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
16	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
17	Creatine metabolism in urea cycle defects. Journal of Inherited Metabolic Disease, 2012, 35, 647-653.	3.6	22
18	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

#	ARTICLE	IF	CITATIONS
19	Cerebrospinal Fluid-Directed rAAV9-rsATP7A Plus Subcutaneous Copper Histidinate Advance Survival and Outcomes in a Menkes Disease Mouse Model. <i>Molecular Therapy - Methods and Clinical Development</i> , 2018, 10, 165-178.	4.1	17
20	P5CS expression study in a new family with <i>ALDH18A1</i> -associated hereditary spastic paraplegia SPG9. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1533-1540.	3.7	14
21	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. <i>Neurogenetics</i> , 2020, 21, 87-96.	1.4	14
22	Corticospinal tract damage in HHH syndrome: a metabolic cause of hereditary spastic paraplegia. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 208.	2.7	12
23	Cobalamin C defect presenting as severe neonatal hyperammonemia. <i>European Journal of Pediatrics</i> , 2011, 170, 887-890.	2.7	10
24	Clinical and biochemical footprints of inherited metabolic diseases. VI. Metabolic dermatoses. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 87-95.	1.1	10
25	Etiology and Management of Pediatric Intestinal Failure: Focus on the Non-Digestive Causes. <i>Nutrients</i> , 2021, 13, 786.	4.1	8
26	Clinical and radiological correlates of activities of daily living in cerebellar atrophy caused by PMM2 mutations (PMM2-CDG). <i>Cerebellum</i> , 2021, 20, 596-605.	2.5	8
27	Acute hyperammonemia in children under deferasirox treatment: cutting the Gordian knot. <i>Clinical Toxicology</i> , 2019, 57, 375-377.	1.9	7
28	Teaching Neuro <i>Images</i> : Galactitol peak and fatal cerebral edema in classic galactosemia. <i>Neurology</i> , 2016, 86, e32-3.	1.1	4
29	CUGC for lysinuric protein intolerance (LPI). <i>European Journal of Human Genetics</i> , 2020, 28, 1129-1134.	2.8	4
30	CUGC for hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome. <i>European Journal of Human Genetics</i> , 2020, 28, 982-987.	2.8	3
31	Skin and Hair Disorders. , 2017, , 341-370.		1