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	Etiology and Management of Pediatric Intestinal Failure: Focus on the Non-Digestive Causes. Nutrients, 2021, 13, 786.	4.1	8
2	Clinical and radiological correlates of activities of daily living in cerebellar atrophy caused by PMM2 mutations (PMM2-CDG). Cerebellum, 2021, 20, 596-605.	2.5	8
3	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
4	Clinical and biochemical footprints of inherited metabolic diseases. VI. Metabolic dermatoses. Molecular Genetics and Metabolism, 2021, 134, 87-95.	1.1	10
5	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics, 2020, 21, 87-96.	1.4	14
6	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
7	CUGC for hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome. European Journal of Human Genetics, 2020, 28, 982-987.	2.8	3
8	CUGC for lysinuric protein intolerance (LPI). European Journal of Human Genetics, 2020, 28, 1129-1134.	2.8	4
9	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
10	Corticospinal tract damage in HHH syndrome: a metabolic cause of hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2019, 14, 208.	2.7	12
11	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
12	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
13	Acute hyperammonemia in children under deferasirox treatment: cutting the Gordian knot. Clinical Toxicology, 2019, 57, 375-377.	1.9	7
14	Cerebrospinal Fluid-Directed rAAV9-rsATP7A Plus Subcutaneous Copper Histidinate Advance Survival and Outcomes in a Menkes Disease Mouse Model. Molecular Therapy - Methods and Clinical Development, 2018, 10, 165-178.	4.1	17
15	Skin and Hair Disorders. , 2017, , 341-370.		1
16	Teaching Neuro Images : Galactitol peak and fatal cerebral edema in classic galactosemia. Neurology, 2016, 86, e32-3.	1.1	4
17	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
18	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

#	Article	IF	Citations
19	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
20	The hyperornithinemiaâ€“hyperammonemia-homocitrullinuria syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 29.	2.7	65
21	Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130.	2.7	482
22	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
23	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
24	MEDNIK syndrome: a novel defect of copper metabolism treatable by zinc acetate therapy. Brain, 2013, 136, 872-881.	7.6	130
25	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
26	Impaired phagocytosis in macrophages from patients affected by lysinuric protein intolerance. Molecular Genetics and Metabolism, 2012, 105, 585-589.	1.1	35
27	Suggested guidelines for the diagnosis and management of urea cycle disorders. Orphanet Journal of Rare Diseases, 2012, 7, 32.	2.7	596
28	Creatine metabolism in urea cycle defects. Journal of Inherited Metabolic Disease, 2012, 35, 647-653.	3.6	22
29	Cobalamin C defect: natural history, pathophysiology, and treatment. Journal of Inherited Metabolic Disease, 2011, 34, 127-135.	3.6	242
30	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. Journal of Inherited Metabolic Disease, 2011, 34, 923-927.	3.6	50
31	Cobalamin C defect presenting as severe neonatal hyperammonemia. European Journal of Pediatrics, 2011, 170, 887-890.	2.7	10