Elena Procopio

List of Publications by Year in descending order

25 papers	455 citations	759233 12 h-index	752698 20 g-index
26 all docs	26 docs citations	26 times ranked	940 citing authors

#	Article	IF	Citations
1	Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. Human Molecular Genetics, 2017, 26, 4257-4266.	2.9	63
2	Clinical features and outcome of 6 new patients carrying de novo KCNB1 gene mutations. Neurology: Genetics, 2017, 3, e206.	1.9	53
3	Neuroimaging Changes in Menkes Disease, Part 1. American Journal of Neuroradiology, 2017, 38, 1850-1857.	2.4	42
4	The treatment of juvenile/adult GM1-gangliosidosis with Miglustat may reverse disease progression. Metabolic Brain Disease, 2017, 32, 1529-1536.	2.9	34
5	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
6	Type I sialidosis, a normosomatic lysosomal disease, in the differential diagnosis of late-onset ataxia and myoclonus: An overview. Molecular Genetics and Metabolism, 2020, 129, 47-58.	1.1	26
7	Early infantile epileptic-dyskinetic encephalopathy due to biallelicPIGPmutations. Neurology: Genetics, 2020, 6, e387.	1.9	26
8	Leigh-like neuroimaging features associated with new biallelic mutations in OPA1. European Journal of Paediatric Neurology, 2017, 21, 671-677.	1.6	25
9	Allergic bronchopulmonary aspergillosis in Italian cystic fibrosis patients: prevalence and percentage of positive tests in the employed diagnostic criteria. European Journal of Epidemiology, 2000, 16, 837-842.	5.7	22
10	Clinical profile and outcome of cardiac involvement in MELAS syndrome. International Journal of Cardiology, 2019, 276, 14-19.	1.7	21
11	Neuroimaging Changes in Menkes Disease, Part 2. American Journal of Neuroradiology, 2017, 38, 1858-1865.	2.4	20
12	Clinical, imaging, biochemical and molecular features in Leigh syndrome: a study from the Italian network of mitochondrial diseases. Orphanet Journal of Rare Diseases, 2021, 16, 413.	2.7	16
13	SARS-CoV-2 infection in a patient with propionic acidemia. Orphanet Journal of Rare Diseases, 2020, 15, 306.	2.7	14
14	Pre-diagnosing and managing patients with GM1 gangliosidosis and related disorders by the evaluation of GM1 ganglioside content. Scientific Reports, 2019, 9, 17684.	3.3	11
15	A rare case of sterol-C4-methyl oxidase deficiency in a young Italian male: Biochemical and molecular characterization. Molecular Genetics and Metabolism, 2017, 121, 329-335.	1.1	9
16	Impact of cardiovascular involvement on the clinical course of paediatric mitochondrial disorders. Orphanet Journal of Rare Diseases, 2020, 15, 196.	2.7	8
17	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. Journal of Clinical Medicine, 2021, 10, 2063.	2.4	8
18	Morquio B disease: From pathophysiology towards diagnosis. Molecular Genetics and Metabolism, 2021, 132, 180-188.	1.1	7

#	Article	IF	Citations
19	Autoantibodies against bactericidal/permeability-increasing protein in cystic fibrosis patients: Comment on the article by Hoffman and Specks. Arthritis and Rheumatism, 1999, 42, 1305-1306.	6.7	3
20	Teaching Neuro Images : Spinal cord gray matter involvement in complex I deficiency mitochondriopathy. Neurology, 2016, 87, e106-7.	1.1	3
21	SARS-CoV-2 infection in patients with primary mitochondrial diseases: Features and outcomes in Italy. Mitochondrion, 2021, 58, 243-245.	3.4	3
22	Neonatal heart failure and noncompaction/dilated cardiomyopathy from mucopolysaccharidosis. First description in literature. Molecular Genetics and Metabolism Reports, 2021, 26, 100714.	1.1	2
23	Autophagic vacuolar myopathy caused by a CLN3 mutation. A case report. Neuromuscular Disorders, 2019, 29, 67-69.	0.6	1
24	Infantile-Onset Pompe Disease: The Care Beyond the Cure. Journal of Neuromuscular Diseases, 2015, 2, S58-S59.	2.6	0
25	Infantile-Onset Pompe Disease: The Care Beyond the Cure. Journal of Neuromuscular Diseases, 2015, 2, S58-S59.	2.6	0