

Elena Procopio

List of Publications by Year in descending order

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Version: 2024-02-01

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	Morquio B disease: From pathophysiology towards diagnosis. <i>Molecular Genetics and Metabolism</i> , 2021, 132, 180-188.	1.1	7
2	Neonatal heart failure and noncompaction/dilated cardiomyopathy from mucopolysaccharidosis. First description in literature. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 26, 100714.	1.1	2
3	SARS-CoV-2 infection in patients with primary mitochondrial diseases: Features and outcomes in Italy. <i>Mitochondrion</i> , 2021, 58, 243-245.	3.4	3
4	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. <i>Journal of Clinical Medicine</i> , 2021, 10, 2063.	2.4	8
5	Clinical, imaging, biochemical and molecular features in Leigh syndrome: a study from the Italian network of mitochondrial diseases. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 413.	2.7	16
6	Type I sialidosis, a normosomatic lysosomal disease, in the differential diagnosis of late-onset ataxia and myoclonus: An overview. <i>Molecular Genetics and Metabolism</i> , 2020, 129, 47-58.	1.1	26
7	SARS-CoV-2 infection in a patient with propionic acidemia. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 306.	2.7	14
8	Impact of cardiovascular involvement on the clinical course of paediatric mitochondrial disorders. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 196.	2.7	8
9	Early infantile epileptic-dyskinetic encephalopathy due to biallelic PIGP mutations. <i>Neurology: Genetics</i> , 2020, 6, e387.	1.9	26
10	Pre-diagnosing and managing patients with GM1 gangliosidosis and related disorders by the evaluation of GM1 ganglioside content. <i>Scientific Reports</i> , 2019, 9, 17684.	3.3	11
11	Autophagic vacuolar myopathy caused by a CLN3 mutation. A case report. <i>Neuromuscular Disorders</i> , 2019, 29, 67-69.	0.6	1
12	Clinical profile and outcome of cardiac involvement in MELAS syndrome. <i>International Journal of Cardiology</i> , 2019, 276, 14-19.	1.7	21
13	Neuroimaging Changes in Menkes Disease, Part 1. <i>American Journal of Neuroradiology</i> , 2017, 38, 1850-1857.	2.4	42
14	Neuroimaging Changes in Menkes Disease, Part 2. <i>American Journal of Neuroradiology</i> , 2017, 38, 1858-1865.	2.4	20
15	Leigh-like neuroimaging features associated with new biallelic mutations in OPA1. <i>European Journal of Paediatric Neurology</i> , 2017, 21, 671-677.	1.6	25
16	The treatment of juvenile/adult GM1-gangliosidosis with Miglustat may reverse disease progression. <i>Metabolic Brain Disease</i> , 2017, 32, 1529-1536.	2.9	34
17	Clinical features and outcome of 6 new patients carrying de novo <i>KCNB1</i> gene mutations. <i>Neurology: Genetics</i> , 2017, 3, e206.	1.9	53
18	A rare case of sterol-C4-methyl oxidase deficiency in a young Italian male: Biochemical and molecular characterization. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 329-335.	1.1	9

#	ARTICLE	IF	CITATIONS
19	Defective mitochondrial rRNA methyltransferase MRM2 causes MELAS-like clinical syndrome. Human Molecular Genetics, 2017, 26, 4257-4266.	2.9	63
20	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
21	Teaching Neuro <i>Images</i> : Spinal cord gray matter involvement in complex I deficiency mitochondriopathy. Neurology, 2016, 87, e106-7.	1.1	3
22	Infantile-Onset Pompe Disease: The Care Beyond the Cure. Journal of Neuromuscular Diseases, 2015, 2, S58-S59.	2.6	0
23	Infantile-Onset Pompe Disease: The Care Beyond the Cure. Journal of Neuromuscular Diseases, 2015, 2, S58-S59.	2.6	0
24	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
25	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