Miriam Rigoldi

List of Publications by Year
in descending order

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33	931	394421	454955
papers	citations	19	30
		h-index	g-index

34	34	34	1675
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Morquio B disease: From pathophysiology towards diagnosis. Molecular Genetics and Metabolism, 2021, 132, 180-188.	1.1	7
2	Dietary lipids in glycogen storage disease type III: A systematic literature study, case studies, and future recommendations. Journal of Inherited Metabolic Disease, 2020, 43, 770-777.	3.6	23
3	Chronic liver involvement in urea cycle disorders. Journal of Inherited Metabolic Disease, 2019, 42, 1118-1127.	3.6	17
4	Novel mutations in two unrelated Italian patients with SSADH deficiency. Metabolic Brain Disease, 2019, 34, 1515-1518.	2.9	4
5	Resting energy expenditure in argininosuccinic aciduria and in other urea cycle disorders. Journal of Inherited Metabolic Disease, 2019, 42, 1105-1117.	3.6	5
6	Refining the Phenotype of Recurrent Rearrangements of Chromosome 16. International Journal of Molecular Sciences, 2019, 20, 1095.	4.1	34
7	Hepatocellular carcinoma in Gaucher disease: an international case series. Journal of Inherited Metabolic Disease, 2018, 41, 819-827.	3.6	37
8	A new case report of severe mucopolysaccharidosis type VII: diagnosis, treatment with haematopoietic cell transplantation and prenatal diagnosis in a second pregnancy. Italian Journal of Pediatrics, 2018, 44, 128.	2.6	12
9	Clinical hints to diagnosis of attenuated forms of Mucopolysaccharidoses. Italian Journal of Pediatrics, 2018, 44, 132.	2.6	25
10	Familiar unbalanced complex rearrangements involving 13 p-arm: description of two cases. Molecular Cytogenetics, 2018, 11, 52.	0.9	1
11	Mis-splicing of the GALNS gene resulting from deep intronic mutations as a cause of Morquio a disease. BMC Medical Genetics, 2018, 19, 183.	2.1	14
12	Improving the diagnosis of cobalamin and related defects by genomic analysis, plus functional and structural assessment of novel variants. Orphanet Journal of Rare Diseases, 2018, 13, 125.	2.7	3
13	Home infusion program with enzyme replacement therapy for Fabry disease: The experience of a large Italian collaborative group. Molecular Genetics and Metabolism Reports, 2017, 12, 85-91.	1.1	20
14	Vagal Nerve Stimulation in the Treatment of Drug-Resistant Epileptic Encephalopathies in Inborn Errors of Metabolism. Child Neurology Open, 2015, 2, 2329048X1561243.	1.1	2
15	Enzymatic replacement therapy for Hunter disease: Up to 9years experience with 17 patients. Molecular Genetics and Metabolism Reports, 2015, 3, 65-74.	1.1	63
16	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
17	Optimizing the Molecular Diagnosis of GALNS: Novel Methods to Define and Characterize Morquio-A Syndrome-Associated Mutations. Human Mutation, 2015, 36, 357-368.	2.5	26
18	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

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19	Genotype-phenotype correlation in Pompe disease, a step forward. Orphanet Journal of Rare Diseases, 2014, 9, 102.	2.7	54
20	Sudden unexpected fatal encephalopathy in adults with OTC gene mutations-Clues for early diagnosis and timely treatment. Orphanet Journal of Rare Diseases, 2014, 9, 105.	2.7	28
21	Intrafamilial phenotypic variability in four families with Andersonâ€Fabry disease. Clinical Genetics, 2014, 86, 258-263.	2.0	45
22	Homozygous MTTP and APOB mutations may lead to hepatic steatosis and fibrosis despite metabolic differences in congenital hypocholesterolemia. Journal of Hepatology, 2014, 61, 891-902.	3.7	116
23	Fertility and pregnancy in women affected by glycogen storage disease type I, results of a multicenter Italian study. Journal of Inherited Metabolic Disease, 2013, 36, 83-89.	3.6	29
24	Quality of Life in Adult Patients with Glycogen Storage Disease Type I: Results of a Multicenter Italian Study. JIMD Reports, 2013, 14, 47-53.	1.5	16
25	Integration of PCR-Sequencing Analysis with Multiplex Ligation-Dependent Probe Amplification for Diagnosis of Hereditary Fructose Intolerance. JIMD Reports, 2012, 6, 31-37.	1.5	11
26	IDUA mutational profiling of a cohort of 102 European patients with mucopolysaccharidosis type I: identification and characterization of 35 novel Î±-L-iduronidase (IDUA) alleles. Human Mutation, 2011, 32, E2189-E2210.	2.5	66
27	Longâ€term observational, nonâ€randomized study of enzyme replacement therapy in lateâ€onset glycogenosis type II. Journal of Inherited Metabolic Disease, 2010, 33, 727-735.	3.6	79
28	Intravenous enzyme replacement therapy: hospital vs home. British Journal of Nursing, 2010, 19, 892-898.	0.7	7
29	First manifestation of citrullinemia type I as differential diagnosis to postpartum psychosis in the puerperal period. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2010, 149, 228-229.	1.1	28
30	Identification and molecular characterization of six novel mutations in the UDP-N-acetylglucosamine-1-phosphotransferase gamma subunit (GNPTG) gene in patients with mucolipidosis III gamma. Human Mutation, 2009, 30, 978-984.	2.5	26
31	Vitamin E supplementation improves neutropenia and reduces the frequency of infections in patients with glycogen storage disease type 1b. European Journal of Pediatrics, 2009, 168, 1069-1074.	2.7	21
32	Enzyme replacement therapy with agalsidase alfa in a cohort of Italian patients with Andersonâ€“Fabry disease: testing the effects with the Mainz Severity Score Index. Clinical Genetics, 2008, 74, 260-266.	2.0	31
33	APOE influences vasospasm and cognition of noncomatose patients with subarachnoid hemorrhage. Neurology, 2005, 64, 1238-1244.	1.1	49