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	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
2	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
3	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
4	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
5	Genotype-phenotype correlation in Pompe disease, a step forward. Orphanet Journal of Rare Diseases, 2014, 9, 102.	2.7	54
6	APOE influences vasospasm and cognition of noncomatose patients with subarachnoid hemorrhage. Neurology, 2005, 64, 1238-1244.	1.1	49
7	Intrafamilial phenotypic variability in four families with Andersonâ€Fabry disease. Clinical Genetics, 2014, 86, 258-263.	2.0	45
8	Hepatocellular carcinoma in Gaucher disease: an international case series. Journal of Inherited Metabolic Disease, 2018, 41, 819-827.	3.6	37
9	Refining the Phenotype of Recurrent Rearrangements of Chromosome 16. International Journal of Molecular Sciences, 2019, 20, 1095.	4.1	34
10	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
11	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
12	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
13	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
14	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
15	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
16	Clinical hints to diagnosis of attenuated forms of Mucopolysaccharidoses. Italian Journal of Pediatrics, 2018, 44, 132.	2.6	25
17	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
18	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

#	Article	IF	Citations
19	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
20	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
21	Chronic liver involvement in urea cycle disorders. Journal of Inherited Metabolic Disease, 2019, 42, 1118-1127.	3.6	17
22	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
23	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
24	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
25	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
26	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
27	Intravenous enzyme replacement therapy: hospital vs home. British Journal of Nursing, 2010, 19, 892-898.	0.7	7
28	Morquio B disease: From pathophysiology towards diagnosis. Molecular Genetics and Metabolism, 2021, 132, 180-188.	1.1	7
29	Resting energy expenditure in argininosuccinic aciduria and in other urea cycle disorders. Journal of Inherited Metabolic Disease, 2019, 42, 1105-1117.	3.6	5
30	Novel mutations in two unrelated Italian patients with SSADH deficiency. Metabolic Brain Disease, 2019, 34, 1515-1518.	2.9	4
31	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
32	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
33	Familiar unbalanced complex rearrangements involving 13 p-arm: description of two cases. Molecular Cytogenetics, 2018, 11, 52.	0.9	1