Barbara Tappino

List of Publications by Year
in descending order

15	425	759233 12	996975 15
papers	citations	h-index	g-index

15	15	15	859
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Assessing Health-Related Quality of Life in Children with Coeliac Disease: The Italian Version of CDDUX. Nutrients, 2021, 13, 485.	4.1	6
2	Predicting the probability of Gaucher disease in subjects with splenomegaly and thrombocytopenia. Scientific Reports, 2021, 11, 2594.	3.3	12
3	The lysosomal storage disorders mucolipidosis type II, type III alpha/beta, and type III gamma: Update onGNPTABandGNPTGmutations. Human Mutation, 2019, 40, 842-864.	2.5	36
4	Concentration-dependent metabolic effects of metformin in healthy and Fanconi anemia lymphoblast cells. Journal of Cellular Physiology, 2018, 233, 1736-1751.	4.1	25
5	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
6	A novel homozygous MCOLN1 double mutant allele leading to TRP channel domain ablation underlies Mucolipidosis IV in an Italian Child. Metabolic Brain Disease, 2015, 30, 681-686.	2.9	8
7	Restoration of the Normal Splicing Pattern of the PLP1 Gene by Means of an Antisense Oligonucleotide Directed against an Exonic Mutation. PLoS ONE, 2013, 8, e73633.	2.5	12
8	Origin and spread of a common deletion causing mucolipidosis type II: insights from patterns of haplotypic diversity. Clinical Genetics, 2011, 80, 273-280.	2.0	15
9	Enigmatic In Vivo iduronate-2-sulfatase (IDS) mutant transcript correction to wild-type in Hunter syndrome. Human Mutation, 2010, 31, E1261-E1285.	2.5	17
10	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. Human Mutation, 2010, 31, E1894-E1914.	2.5	93
11	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
12	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase Î±- and Î²-subunit (GNPTAB) gene mutations causing mucolipidosis types IIIÎ±/Î² and IIIÎ±/Î² in 46 patients. Human Mutation, 2009, 30, E956-E973.	2.5	38
13	Molecular analysis of NPC1 and NPC2 gene in 34 Niemannâ€“Pick C Italian Patients: identification and structural modeling of novel mutations. Neurogenetics, 2009, 10, 229-239.	1.4	39
14	An Alu insertion in compound heterozygosity with a microduplication in GNPTAB gene underlies Mucolipidosis II. Molecular Genetics and Metabolism, 2008, 93, 129-133.	1.1	22
15	Molecular analysis and characterization of nine novel CTSK mutations in twelve patients affected by pycnodysostosis. Human Mutation, 2007, 28, 524-524.	2.5	64