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	Identification and characterization of 15 novel GALC gene mutations causing Krabbe disease. Human Mutation, 2010, 31, E1894-E1914.	2.5	93
2	Molecular analysis and characterization of nine novel CTSK mutations in twelve patients affected by pycnodysostosis. Human Mutation, 2007, 28, 524-524.	2.5	64
3	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
4	Molecular characterization of 22 novel UDP-N-acetylglucosamine-1-phosphate transferase Î±- and Î²-subunit (GNPTAB) gene mutations causing mucolipidosis types IIÎ±/Î² and IIIÎ±/Î² in 46 patients. Human Mutation, 2009, 30, E956-E973.	2.5	38
5	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
6	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
7	Concentration-dependent metabolic effects of metformin in healthy and Fanconi anemia lymphoblast cells. Journal of Cellular Physiology, 2018, 233, 1736-1751.	4.1	25
8	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
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	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
11	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
12	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
13	Predicting the probability of Gaucher disease in subjects with splenomegaly and thrombocytopenia. Scientific Reports, 2021, 11, 2594.	3.3	12
14	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
15	Assessing Health-Related Quality of Life in Children with Coeliac Disease: The Italian Version of CDDUX. Nutrients, 2021, 13, 485.	4.1	6