Lenka MrÃ¡zovÃ¡

List of Publications by Year
in descending order

11 papers	294 citations	1162889 8 h-index	1281743 11 g-index
12 all docs	12 docs citations	12 times ranked	576 citing authors

#	Article	IF	Citations
1	Mutations in TMEM76* Cause Mucopolysaccharidosis IIIC (Sanfilippo C Syndrome). American Journal of Human Genetics, 2006, 79, 807-819.	2.6	77
2	Sanfilippo syndrome type C: mutation spectrum in the heparan sulfate acetyl-CoA: Î±-glucosaminide N-acetyltransferase (HGSNAT) gene. Human Mutation, 2009, 30, 918-925.	1.1	51
3	Analysis of the Î²-Glucocerebrosidase Gene in Czech and Slovak Gaucher Patients: Mutation Profile and Description of Six Novel Mutant Alleles. Blood Cells, Molecules, and Diseases, 1999, 25, 287-298.	0.6	37
4	Contiguous X-chromosome Deletion Syndrome Encompassing the BTK, TIMM8A, TAF7L, and DRP2 Genes. Journal of Clinical Immunology, 2007, 27, 640-646.	2.0	35
5	3-Hydroxy-3-methylglutaryl-coenzyme A lyase deficiency: Clinical presentation and outcome in a series of 37 patients. Molecular Genetics and Metabolism, 2017, 121, 206-215.	0.5	32
6	Rare variants in known and novel candidate genes predisposing to statin-associated myopathy. Pharmacogenomics, 2016, 17, 1405-1414.	0.6	17
7	Biochemical and molecular analyses in three patients with 3-hydroxy-3-methylglutaric aciduria. Journal of Inherited Metabolic Disease, 2003, 26, 433-441.	1.7	13
8	Two novel mutations in mitochondrial acetoacetyl-CoA thiolase deficiency. Journal of Inherited Metabolic Disease, 2005, 28, 235-236.	1.7	13
9	A mutation in the SAA1 promoter causes hereditary amyloid A amyloidosis. Kidney International, 2022, 101, 349-359.	2.6	10
10	Glucocerebrosidase gene has an alternative upstream promoter, which has features and expression characteristic of housekeeping genes. Blood Cells, Molecules, and Diseases, 2011, 46, 239-245.	0.6	6
11	HGSNAT has a TATA-less promoter with multiple starts of transcription. Gene, 2016, 592, 36-42.	1.0	2