Helen Michelakakis

List of Publications by Year
in descending order

43 papers	1,120 citations	430874 18 h-index	414414 32 g-index
44 all docs	44 docs citations	44 times ranked	1659 citing authors

#	Article	IF	Citations
1	Incidence and natural history of mucopolysaccharidosis type III in France and comparison with United Kingdom and Greece. American Journal of Medical Genetics, Part A, 2011, 155, 58-68.	1.2	133
2	Plasma tumor necrosis factor-a (TNF-a) levels in Gaucher disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1996, 1317, 219-222.	3.8	79
3	Update of the pompe disease mutation database with 60 novel GAA sequence variants and additional studies on the functional effect of 34 previously reported variants. Human Mutation, 2012, 33, 1161-1165.	2.5	67
4	Mutation analyses in 17 patients with deficiency in acid Î²-galactosidase: three novel point mutations and high correlation of mutation W273L with Morquio disease type B. Human Genetics, 2001, 109, 159-166.	3.8	62
5	Evidence of an association between the scavenger receptor class B member 2 gene and Parkinson's disease. Movement Disorders, 2012, 27, 400-405.	3.9	56
6	Lysosomal alterations in peripheral blood mononuclear cells of Parkinson's disease patients. Movement Disorders, 2015, 30, 1830-1834.	3.9	53
7	Sanfilippo syndrome type C: mutation spectrum in the heparan sulfate acetyl-CoA: Î±-glucosaminide N-acetyltransferase (HGSNAT) gene. Human Mutation, 2009, 30, 918-925.	2.5	51
8	Î²-Glucocerebrosidase gene mutations in two cohorts of Greek patients with sporadic Parkinson's disease. Molecular Genetics and Metabolism, 2011, 104, 149-152.	1.1	47
9	Loss of Î²-Glucocerebrosidase Activity Does Not Affect Alpha-Synuclein Levels or Lysosomal Function in Neuronal Cells. PLoS ONE, 2013, 8, e60674.	2.5	47
10	GM1 gangliosidosis and Morquio B disease: expression analysis of missense mutations affecting the catalytic site of acid ÃŽÂ²-galactosidase. Human Mutation, 2009, 30, 1214-1221.	2.5	44
11	A novel mutation in the flavin-containing monooxygenase 3 gene, FMO3, that causes fish-odour syndrome: activity of the mutant enzyme assessed by proton NMR spectroscopy. Pharmacogenetics and Genomics, 2000, 10, 439-451.	5.7	43
12	Circulating Brainâ€Enriched MicroRNAs for Detection and Discrimination of Idiopathic and Genetic Parkinson's Disease. Movement Disorders, 2020, 35, 457-467.	3.9	43
13	Mutation analysis in 20 patients with Hunter disease. , 1996, 7, 76-78.		38
14	Alpha-synuclein dimerization in erythrocytes of patients with genetic and non-genetic forms of Parkinsonâ€™s Disease. Neuroscience Letters, 2018, 672, 145-149.	2.1	35
15	Increased dimerization of alpha-synuclein in erythrocytes in Gaucher disease and aging. Neuroscience Letters, 2012, 528, 205-209.	2.1	24
16	Investigation of original multivalent iminosugars as pharmacological chaperones for the treatment of Gaucher disease. Carbohydrate Research, 2016, 429, 98-104.	2.3	22
17	Screening for galactosaemia in Greece. Paediatric and Perinatal Epidemiology, 1997, 11, 436-440.	1.7	21
18	Plasmalogen levels in Gaucher disease. Blood Cells, Molecules, and Diseases, 2008, 41, 196-199.	1.4	19

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19	Homozygosity for the double D409H+H255Q allele in type II Gaucher disease. Journal of Inherited Metabolic Disease, 2006, 29, 591-591.	3.6	18
20	Haplotype analysis suggests a single Balkan origin for the Gaucher disease [D409H;H255Q] double mutant allele. Human Mutation, 2008, 29, E58-E67.	2.5	18
21	Gaucher disease: Plasmalogen levels in relation to primary lipid abnormalities and oxidative stress. Blood Cells, Molecules, and Diseases, 2014, 53, 30-33.	1.4	18
22	Phenotypic Characteristics in GBA-Associated Parkinsonâ€™s Disease: A Study in a Greek Population. Journal of Parkinson's Disease, 2018, 8, 101-105.	2.8	18
23	The effect of diet on total antioxidant status, erythrocyte membrane Na+,K+-ATPase and Mg2+-ATPase activities in patients with classical galactosaemia. Clinical Nutrition, 2005, 24, 151-157.	5.0	15
24	Choline and L-carnitine as precursors of trimethylamine. Biochemical Society Transactions, 1997, 25, 96S-96S.	3.4	14
25	Glucocerebrosidase Enhancers for Selected Gaucher Disease Genotypes by Modification of Îâ€1â€Câ€Substituted Iminoâ€<scp>D</scp>â€xylitols (DIXs) by Click Chemistry. ChemMedChem, 2014, 9, 1744-1754.	3.2	13
26	Î±-Synuclein dimerization in erythrocytes of Gaucher disease patients: correlation with lipid abnormalities and oxidative stress. Neuroscience Letters, 2016, 613, 1-5.	2.1	13
27	A novel mutation deep within intron 7 of the GBA gene causes Gaucher disease. Molecular Genetics & Genomic Medicine, 2020, 8, e1090.	1.2	13
28	Plasma lysosomal enzyme activities in congenital disorders of glycosylation, galactosemia and fructosemia. Clinica Chimica Acta, 2009, 401, 81-83.	1.1	12
29	The Spectrum of Niemann-Pick Type C Disease in Greece. JIMD Reports, 2017, 36, 41-48.	1.5	12
30	Stereodivergent synthesis of right- and left-handed iminoxylitol heterodimers and monomers. Study of their impact on Î²-glucocerebrosidase activity. Organic and Biomolecular Chemistry, 2017, 15, 3681-3705.	2.8	9
31	Novel NPC1 mutations with different segregation in two related Greek patients with Niemann-Pick type C disease: molecular study in the extended pedigree and clinical correlations. BMC Medical Genetics, 2017, 18, 51.	2.1	9
32	Selective chaperone effect of aminocyclitol derivatives on G202R and other mutant glucocerebrosidases causing Gaucher disease. International Journal of Biochemistry and Cell Biology, 2014, 54, 245-254.	2.8	8
33	Segmental and total uniparental isodisomy (UPiD) as a disease mechanism in autosomal recessive lysosomal disorders: evidence from SNP arrays. European Journal of Human Genetics, 2019, 27, 919-927.	2.8	8
34	Gaucher disease: Biochemical and molecular findings in 141 patients diagnosed in Greece. Molecular Genetics and Metabolism Reports, 2020, 24, 100614.	1.1	7
35	Erythrocyte membrane acetylcholinesterase, Na⁺, K⁺â€ATPase and Mg²⁺â€ATPase activities in patients with classical galactosaemia. Acta Paediatrica, International Journal of Paediatrics, 2005, 94, 1223-1226.	1.5	6
36	The effect of cytochalasin B on the release of lysosomal enzymes and intra-lysosomally-stored polyvinylpyrrolidone in the isolated perfused rat liver. Biochemical Pharmacology, 1984, 33, 2047-2053.	4.4	4

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37	Perinatal lethal form of Gaucher disease. Clinical and molecular characterization of a Greek case. Blood Cells, Molecules, and Diseases, 2010, 44, 82-83.	1.4	4
38	Transferrin isoelectric focusing and plasma lysosomal enzyme activities in the diagnosis and followâ€up of hereditary fructose intolerance. Clinica Chimica Acta, 2012, 413, 1714-1715.	1.1	4
39	The Effect of Cytochalasin B on the Release of Lysosomal Enzymes from Isolated Rat Hepatocytes. Biochemical Society Transactions, 1979, 7, 1283-1285.	3.4	3
40	Neuroimaging findings in Hunter disease. Journal of the Neurological Sciences, 2014, 342, 200-201.	0.6	3
41	Prevalence of antibodies to ganglioside and Hep 2 in Gaucher, Niemann â€“ Pick type C and Sanfilippo diseases. Molecular Genetics and Metabolism Reports, 2019, 20, 100477.	1.1	3
42	The uptake and release of lysosomal enzymes by isolated perfused rat liver. Biochemical Society Transactions, 1980, 8, 569-570.	3.4	2
43	The release of intralysosomally-stored 125I-triton WR-1339 and lysosomal enzymes from the isolated perfused rat liver in the presence and absence of cytochalasin B. Biochemical Pharmacology, 1986, 35, 933-938.	4.4	2