Hana Vlaskova

List of Publications by Year
in descending order

27 papers	540 citations	687363 13 h-index	642732 23 g-index
30 all docs	30 docs citations	30 times ranked	1554 citing authors

#	Article	IF	Citations
1	Mutations in CLN7/MFSD8 are a common cause of variant late-infantile neuronal ceroid lipofuscinosis. Brain, 2009, 132, 810-819.	7.6	116
2	Insights into Severe 5,10-Methylenetetrahydrofolate Reductase Deficiency: Molecular Genetic and Enzymatic Characterization of 76 Patients. Human Mutation, 2015, 36, 611-621.	2.5	63
3	Observational, retrospective study of a large cohort of patients with Niemann-Pick disease type C in the Czech Republic: a surprisingly stable diagnostic rate spanning almost 40 years. Orphanet Journal of Rare Diseases, 2014, 9, 140.	2.7	56
4	Prevalence of Fabry disease in male patients with unexplained left ventricular hypertrophy in primary cardiology practice: prospective Fabry cardiomyopathy screening study (FACSS). Journal of Inherited Metabolic Disease, 2014, 37, 455-460.	3.6	47
5	Atypical CLN2 with later onset and prolonged course: a neuropathologic study showing different sensitivity of neuronal subpopulations to TPP1 deficiency. Acta Neuropathologica, 2008, 116, 119-124.	7.7	32
6	Mucopolysaccharidosis type I in 21 Czech and Slovak patients: Mutation analysis suggests a functional importance of Câ€terminus of the IDUA protein. American Journal of Medical Genetics, Part A, 2009, 149A, 965-974.	1.2	27
7	Danon disease: A focus on processing of the novel LAMP2 mutation and comments on the beneficial use of peripheral white blood cells in the diagnosis of LAMP2 deficiency. Gene, 2012, 498, 183-195.	2.2	27
8	Genotypeâ€“phenotype correlation in 44 Czech, Slovak, Croatian and Serbian patients with mucopolysaccharidosis type <scp>II</scp>. Clinical Genetics, 2017, 91, 787-796.	2.0	24
9	Treatment of cataplexy in Niemannâ€“Pick disease type C with the use of miglustat. European Journal of Paediatric Neurology, 2011, 15, 84-87.	1.6	21
10	Amyotrophy, cerebellar impairment and psychiatric disease are the main symptoms in a cohort of 14 Czech patients with the late-onset form of Tayâ€“Sachs disease. Journal of Neurology, 2019, 266, 1953-1959.	3.6	19
11	Mosaic tissue distribution of the tandem duplication of LAMP2 exons 4 and 5 demonstrates the limits of Danon disease cellular and molecular diagnostics. Journal of Inherited Metabolic Disease, 2014, 37, 117-124.	3.6	17
12	Disruption of OTC promoter-enhancer interaction in a patient with symptoms of ornithine carbamoyltransferase deficiency. Human Mutation, 2010, 31, E1294-E1303.	2.5	14
13	Hyperuricemia and gout due to deficiency of hypoxanthineâ€“guanine phosphoribosyltransferase in female carriers: New insight to differential diagnosis. Clinica Chimica Acta, 2015, 440, 214-217.	1.1	14
14	LAMP2 exonâ€copy number variations in Danon disease heterozygote female probands: Infrequent or underdetected?. American Journal of Medical Genetics, Part A, 2018, 176, 2430-2434.	1.2	9
15	Alu â€mediated Xq24 deletion encompassing CUL4B , LAMP2 , ATP1B4 , TMEM255A , and ZBTB33 genes causes Danon disease in a female patient. American Journal of Medical Genetics, Part A, 2020, 182, 219-223.	1.2	9
16	Late diagnosis of mucopolysaccharidosis type IVB and successful aortic valve replacement in a 60-year-old female patient. Cardiovascular Pathology, 2018, 35, 52-56.	1.6	7
17	Clinical spectrum in CADASIL family with a new mutation. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2013, 157, 379-382.	0.6	7
18	Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. Clinical Genetics, 2013, 84, 552-559.	2.0	6

#	Article	IF	Citations
19	Transcript, protein, metabolite and cellular studies in skin fibroblasts demonstrate variable pathogenic impacts of NPC1 mutations. Orphanet Journal of Rare Diseases, 2020, 15, 85.	2.7	5
20	Pigmentary retinopathy can indicate the presence of pathogenic LAMP2 variants even in somatic mosaic carriers with no additional signs of Danon disease. Acta Ophthalmologica, 2021, 99, 61-68.	1.1	5
21	Impact of Newborn Screening and Early Dietary Management on Clinical Outcome of Patients with Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency and Medium Chain Acyl-CoA Dehydrogenase Deficiencyâ€"A Retrospective Nationwide Study. Nutrients, 2021, 13, 2925.	4.1	4
22	Clinical and Genetic Study of X-Linked Juvenile Retinoschisis in the Czech Population. Genes, 2021, 12, 1816.	2.4	4
23	Systemic AL amyloidosis with unusual cutaneous presentation unmasked by carotenoderma. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2014, 21, 57-61.	3.0	3
24	Danon disease - a disorder of autophagy as a cause of hypertrophic cardiomyopathy. Cor Et Vasa, 2010, 52, 706-712.	0.1	1
25	Familiar Amyloid Polyneuropathy â€"â€Š a Case Report. Ceska A Slovenska Neurologie A Neurochirurgie, 2015, 78/111, 710-714.	0.1	1
26	Autopsy confirmed CADASIL with the c.1490Câ€‰>â€‰T (p.S497L) variant of uncertain causal significance. Neurological Sciences, 2022, 43, 2115-2118.	1.9	0
27	X-linked adrenoleukodystrophy: phenotype-genotype correlation in hemizygous males and heterozygous females with ABCD1 mutations. Neuroendocrinology Letters, 2021, 42, 359-367.	0.2	0