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	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
2	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
3	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
4	Clinical and Genetic Study of X-Linked Juvenile Retinoschisis in the Czech Population. Genes, 2021, 12, 1816.	2.4	4
5	X-linked adrenoleukodystrophy: phenotype-genotype correlation in hemizygous males and heterozygous females with ABCD1 mutations. Neuroendocrinology Letters, 2021, 42, 359-367.	0.2	0
6	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
7	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
8	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
9	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
10	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
11	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
12	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
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	Familiar Amyloid Polyneuropathy â€“â€Š a Case Report. Ceska A Slovenska Neurologie A Neurochirurgie, 2015, 78/111, 710-714.	0.1	1
15	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
16	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
17	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
18	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

#	Article	IF	Citations
19	Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. Clinical Genetics, 2013, 84, 552-559.	2.0	6
20	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
21	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
22	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
23	Disruption of OTC promoter-enhancer interaction in a patient with symptoms of ornithine carbamoyltransferase deficiency. Human Mutation, 2010, 31, E1294-E1303.	2.5	14
24	Danon disease - a disorder of autophagy as a cause of hypertrophic cardiomyopathy. Cor Et Vasa, 2010, 52, 706-712.	0.1	1
25	Mutations in CLN7/MFSD8 are a common cause of variant late-infantile neuronal ceroid lipofuscinosis. Brain, 2009, 132, 810-819.	7.6	116
26	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
27	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