

Filip Majer

List of Publications by Year in descending order

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Version: 2024-02-01

21
papers

274
citations

1040056

9
h-index

888059

17
g-index

23
all docs

23
docs citations

23
times ranked

483
citing authors

#	ARTICLE	IF	CITATIONS
1	Loading of cell cultures with cholesterolâ€dextran particles as a new functional test for Niemannâ€Pick type C disease. <i>Journal of Inherited Metabolic Disease</i> , 2022, , .	3.6	0
2	Pigmentary retinopathy can indicate the presence of pathogenic LAMP2 variants even in somatic mosaic carriers with no additional signs of Danon disease. <i>Acta Ophthalmologica</i> , 2021, 99, 61-68.	1.1	5
3	Easy and fast PCRâ€based protocol allows characterization of breakpoints resulting from <i>Alu</i> -mediated genomic rearrangements. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1830.	1.2	0
4	<i>Alu</i> -mediated Xq24 deletion encompassing CUL4B , LAMP2 , ATP1B4 , TMEM255A , and ZBTB33 genes causes Danon disease in a female patient. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 219-223.	1.2	9
5	Danon disease is an underdiagnosed cause of advanced heart failure in young female patients: a LAMP2 flow cytometric study. <i>ESC Heart Failure</i> , 2020, 7, 2534-2543.	3.1	8
6	Spinal muscular atrophy caused by a novel <i>Alu</i> -mediated deletion of exons 2aâ€5 in <i>SMN1</i> undetectable with routine genetic testing. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1238.	1.2	10
7	Transcript, protein, metabolite and cellular studies in skin fibroblasts demonstrate variable pathogenic impacts of NPC1 mutations. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 85.	2.7	5
8	<i>LAMP2</i> exon copy number variations in Danon disease heterozygote female probands: Infrequent or underdetected?. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2430-2434.	1.2	9
9	Detailed molecular characterization of a novel IDS exonic mutation associated with multiple pseudoexon activation. <i>Journal of Molecular Medicine</i> , 2017, 95, 299-309.	3.9	5
10	LAMP2 flow cytometry in peripheral white blood cells is an established method that facilitates identification of heterozygous Danon disease female patients and mosaic mutation carriers. <i>Journal of Cardiology</i> , 2015, 66, 88-89.	1.9	6
11	Mosaic tissue distribution of the tandem duplication of <i>LAMP2</i> exons 4 and 5 demonstrates the limits of Danon disease cellular and molecular diagnostics. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 117-124.	3.6	17
12	Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. <i>Clinical Genetics</i> , 2013, 84, 552-559.	2.0	6
13	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. <i>Gene</i> , 2012, 498, 183-195.	2.2	27
14	Bioinformatic and biochemical studies point to AAGR-1 as the ortholog of human acid Î±-glucosidase in <i>Caenorhabditis elegans</i> . <i>Molecular and Cellular Biochemistry</i> , 2010, 341, 51-63.	3.1	2
15	Therapeutic targets for influenza â€ perspectives in drug development. <i>Collection of Czechoslovak Chemical Communications</i> , 2010, 75, 81-103.	1.0	3
16	Two aspartic proteinases secreted by the pathogenic yeast <i>Candida parapsilosis</i> differ in expression pattern and catalytic properties. <i>Biological Chemistry</i> , 2009, 390, 259-68.	2.5	26
17	Photodynamic Therapy of Nonmelanoma Skin Cancer with Topical <i>Hypericum perforatum</i> Extractâ€A Pilot Study. <i>Photochemistry and Photobiology</i> , 2008, 84, 779-785.	2.5	71
18	Estrogen-induced cholestasis results in a dramatic increase of b-series gangliosides in the rat liver. <i>Biomedical Chromatography</i> , 2007, 21, 446-450.	1.7	11

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19	Changes in GM1 ganglioside content and localization in cholestatic rat liver. <i>Glycoconjugate Journal</i> , 2007, 24, 231-241.	2.7	7
20	Identification of bilirubin reduction products formed by <i>Clostridium perfringens</i> isolated from human neonatal fecal flora. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2006, 833, 149-157.	2.3	39
21	Structure-based specificity mapping of secreted aspartic proteases of <i>Candida parapsilosis</i> , <i>Candida albicans</i> , and <i>Candida tropicalis</i> using peptidomimetic inhibitors and homology modeling. <i>Biological Chemistry</i> , 2006, 387, 1247-1254.	2.5	8