

# Filip Majer

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4095209/publications.pdf>

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  | Photodynamic Therapy of Nonmelanoma Skin Cancer with Topical <i>Hypericum perforatum</i> Extract—A Pilot Study. <i>Photochemistry and Photobiology</i> , 2008, 84, 779-785.                                                                                            | 2.5 | 71        |
| 2  | 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        |
| 3  | 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        |
| 4  | 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        |
| 5  | 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        |
| 6  | 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        |
| 7  | Spinal muscular atrophy caused by a novel <i>Alu</i> -mediated deletion of exons 2a-5 in <i>SMN1</i> undetectable with routine genetic testing. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1238.                                                     | 1.2 | 10        |
| 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  | <i>Alu</i> -mediated Xq24 deletion encompassing <i>CUL4B</i> , <i>LAMP2</i> , <i>ATP1B4</i> , <i>TMEM255A</i> , and <i>ZBTB33</i> genes causes Danon disease in a female patient. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 219-223.            | 1.2 | 9         |
| 10 | 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         |
| 11 | 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         |
| 12 | Changes in GM1 ganglioside content and localization in cholestatic rat liver. <i>Glycoconjugate Journal</i> , 2007, 24, 231-241.                                                                                                                                       | 2.7 | 7         |
| 13 | Ornithine carbamoyltransferase deficiency: molecular characterization of 29 families. <i>Clinical Genetics</i> , 2013, 84, 552-559.                                                                                                                                    | 2.0 | 6         |
| 14 | 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         |
| 15 | 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         |
| 16 | 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         |
| 17 | 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         |
| 18 | Therapeutic targets for influenza—perspectives in drug development. <i>Collection of Czechoslovak Chemical Communications</i> , 2010, 75, 81-103.                                                                                                                      | 1.0 | 3         |

| #  | ARTICLE                                                                                                                                                                                                | IF  | CITATIONS |
|----|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----|-----------|
| 19 | 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         |
| 20 | Easy and fast PCR-based protocol allows characterization of breakpoints resulting from <i>Alu</i> -mediated genomic rearrangements. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1830. | 1.2 | 0         |
| 21 | Loading of cell cultures with cholesterol-dextran particles as a new functional test for Niemann-Pick type C disease. <i>Journal of Inherited Metabolic Disease</i> , 2022, , .                        | 3.6 | 0         |