

Jakub Sikora

List of Publications by Year in descending order

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Version: 2024-02-01

45
papers

1,386
citations

331670

21
h-index

345221

36
g-index

47
all docs

47
docs citations

47
times ranked

2184
citing authors

#	ARTICLE	IF	CITATIONS
1	Dominant Renin Gene Mutations Associated with Early-Onset Hyperuricemia, Anemia, and Chronic Kidney Failure. <i>American Journal of Human Genetics</i> , 2009, 85, 204-213.	6.2	146
2	Alterations of uromodulin biology: a common denominator of the genetically heterogeneous FJHN/MCKD syndrome. <i>Kidney International</i> , 2006, 70, 1155-1169.	5.2	111
3	A Murine Niemann-Pick C1 I1061T Knock-In Model Recapitulates the Pathological Features of the Most Prevalent Human Disease Allele. <i>Journal of Neuroscience</i> , 2015, 35, 8091-8106.	3.6	97
4	Acid sphingomyelinase deficiency. Phenotype variability with prevalence of intermediate phenotype in a series of twenty-five Czech and Slovak patients. A multi-approach study. <i>Journal of Inherited Metabolic Disease</i> , 2005, 28, 203-227.	3.6	85
5	Efficacy and ototoxicity of different cyclodextrins in Niemann-Pick C disease. <i>Annals of Clinical and Translational Neurology</i> , 2016, 3, 366-380.	3.7	78
6	Mutations in TMEM76* Cause Mucopolysaccharidosis IIIC (Sanfilippo C Syndrome). <i>American Journal of Human Genetics</i> , 2006, 79, 807-819.	6.2	77
7	Mutations of ATIC and ADSL affect purinosome assembly in cultured skin fibroblasts from patients with AICA-ribosiduria and ADSL deficiency. <i>Human Molecular Genetics</i> , 2012, 21, 1534-1543.	2.9	62
8	A novel transgenic mouse model of CBS-deficient homocystinuria does not incur hepatic steatosis or fibrosis and exhibits a hypercoagulative phenotype that is ameliorated by betaine treatment. <i>Molecular Genetics and Metabolism</i> , 2010, 101, 153-162.	1.1	60
9	Cystathionine beta-synthase null homocystinuric mice fail to exhibit altered hemostasis or lowering of plasma homocysteine in response to betaine treatment. <i>Molecular Genetics and Metabolism</i> , 2010, 101, 163-171.	1.1	57
10	Quantitation of plasmatic lysosphingomyelin and lysosphingomyelin-509 for differential screening of Niemann-Pick A/B and C diseases. <i>Analytical Biochemistry</i> , 2017, 525, 73-77.	2.4	49
11	Lysosomal Membrane Permeability Stimulates Protein Aggregate Formation in Neurons of a Lysosomal Disease. <i>Journal of Neuroscience</i> , 2013, 33, 10815-10827.	3.6	47
12	Clinical and molecular characterization of a family with a dominant renin gene mutation and response to treatment with fludrocortisone. <i>Clinical Nephrology</i> , 2010, 74, 411-422.	0.7	44
13	Acid Ceramidase Deficiency in Mice Results in a Broad Range of Central Nervous System Abnormalities. <i>American Journal of Pathology</i> , 2017, 187, 864-883.	3.8	41
14	Seven Novel Acid Sphingomyelinase Gene Mutations in Niemann-Pick Type A and B Patients. <i>Annals of Human Genetics</i> , 2003, 67, 63-70.	0.8	39
15	Neurolysosomal pathology in human prosaposin deficiency suggests essential neurotrophic function of prosaposin. <i>Acta Neuropathologica</i> , 2007, 113, 163-175.	7.7	30
16	Replacement of Î±-galactosidase A in Fabry disease: effect on fibroblast cultures compared with biopsied tissues of treated patients. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2008, 452, 651-665.	2.8	29
17	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
18	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. <i>Kidney International</i> , 2020, 98, 1589-1604.	5.2	27

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19	Lysosomal compromise and brain dysfunction: examining the role of neuroaxonal dystrophy. <i>Biochemical Society Transactions</i> , 2010, 38, 1436-1441.	3.4	26
20	Chronic lung injury and impaired pulmonary function in a mouse model of acid ceramidase deficiency. <i>American Journal of Physiology - Lung Cellular and Molecular Physiology</i> , 2018, 314, L406-L420.	2.9	26
21	Subclinical course of adult visceral Niemann-Pick type C1 disease. A rare or underdiagnosed disorder?. <i>Journal of Inherited Metabolic Disease</i> , 2006, 29, 591-591.	3.6	25
22	X-linked Christianson syndrome: heterozygous female <i>Slc9a6</i> knockout mice develop mosaic neuropathological changes and related behavioral abnormalities. <i>DMM Disease Models and Mechanisms</i> , 2015, 9, 13-23.	2.4	19
23	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
24	Neural cells generated from human induced pluripotent stem cells as a model of CNS involvement in mucopolysaccharidosis type II. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 221-229.	3.6	16
25	Acid Ceramidase Deficiency in Mice Leads to Severe Ocular Pathology and Visual Impairment. <i>American Journal of Pathology</i> , 2019, 189, 320-338.	3.8	16
26	Characterization of <i>gana-1</i> , a <i>Caenorhabditis elegans</i> gene encoding a single ortholog of vertebrate alpha-galactosidase and alpha-N-acetylgalactosaminidase. <i>BMC Cell Biology</i> , 2005, 6, 5.	3.0	13
27	N-butyldeoxynojirimycin delays motor deficits, cerebellar microgliosis, and Purkinje cell loss in a mouse model of mucopolidosis type IV. <i>Neurobiology of Disease</i> , 2017, 105, 257-270.	4.4	13
28	Hepatic pathology and altered gene transcription in a murine model of acid ceramidase deficiency. <i>Laboratory Investigation</i> , 2019, 99, 1572-1592.	3.7	12
29	Autosomal-dominant adult neuronal ceroid lipofuscinosis caused by duplication in <i>DNAJC5</i> initially missed by Sanger and whole-exome sequencing. <i>European Journal of Human Genetics</i> , 2020, 28, 783-789.	2.8	10
30	A mutation in the <i>SAA1</i> promoter causes hereditary amyloid A amyloidosis. <i>Kidney International</i> , 2022, 101, 349-359.	5.2	10
31	<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
32	Alu-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
33	Danon disease is an underdiagnosed cause of advanced heart failure in young female patients: a <i>LAMP2</i> flow cytometric study. <i>ESC Heart Failure</i> , 2020, 7, 2534-2543.	3.1	8
34	<i>NOTCH2NLC</i> CCG Repeats Are Not Expanded and Skin Biopsy Was Negative in an Infantile Patient With Neuronal Intranuclear Inclusion Disease. <i>Journal of Neuropathology and Experimental Neurology</i> , 2020, 79, 1065-1071.	1.7	8
35	Late diagnosis of mucopolysaccharidosis type IVB and successful aortic valve replacement in a 60-year-old female patient. <i>Cardiovascular Pathology</i> , 2018, 35, 52-56.	1.6	7
36	<i>LAMP2</i> 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

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37	Specific storage of glycoconjugates with terminal β -galactosyl moieties in the exocrine pancreas of Fabry disease patients with blood group B. <i>Glycobiology</i> , 2018, 28, 382-391.	2.5	5
38	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
39	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
40	Combined valve replacement and aortocoronary bypass in an adult mucopolysaccharidosis type VII patient. <i>Cardiovascular Pathology</i> , 2021, 50, 107297.	1.6	5
41	Pitfalls of X-chromosome inactivation testing in females with Fabry disease. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1979-1989.	1.2	5
42	Bioinformatic and biochemical studies point to AAGR-1 as the ortholog of human acid β -glucosidase in <i>Caenorhabditis elegans</i> . <i>Molecular and Cellular Biochemistry</i> , 2010, 341, 51-63.	3.1	2
43	Genetic heterogeneity of neuronal intranuclear inclusion disease: What about the infantile variant?. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 994-1001.	3.7	2
44	Retrospective sequence analysis of the human PRNP gene from the formaldehyde-fixed paraffin-embedded tissues: Report of two cases of creutzfeldt-jakob disease. <i>Folia Microbiologica</i> , 2006, 51, 619-625.	2.3	1
45	Easy and fast PCR-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