Jakub Sikora

List of Publications by Year
in descending order

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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	A mutation in the SAA1 promoter causes hereditary amyloid A amyloidosis. Kidney International, 2022, 101, 349-359.	5.2	10
2	Pitfalls of Xâ€chromosome inactivation testing in females with Fabry disease. American Journal of Medical Genetics, Part A, 2022, 188, 1979-1989.	1.2	5
3	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
4	Combined valve replacement and aortocoronary bypass in an adult mucopolysaccharidosis type VII patient. Cardiovascular Pathology, 2021, 50, 107297.	1.6	5
5	Genetic heterogeneity of neuronal intranuclear inclusion disease: What about the infantile variant?. Annals of Clinical and Translational Neurology, 2021, 8, 994-1001.	3.7	2
6	Easy and fast PCRâ€based protocol allows characterization of breakpoints resulting from Alu/Aluâ€mediated genomic rearrangements. Molecular Genetics & Genomic Medicine, 2021, 9, e1830.	1.2	0
7	Autosomal-dominant adult neuronal ceroid lipofuscinosis caused by duplication in DNAJC5 initially missed by Sanger and whole-exome sequencing. European Journal of Human Genetics, 2020, 28, 783-789.	2.8	10
8	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
9	Danon disease is an underdiagnosed cause of advanced heart failure in young female patients: a LAMP2 flow cytometric study. ESC Heart Failure, 2020, 7, 2534-2543.	3.1	8
10	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. Kidney International, 2020, 98, 1589-1604.	5.2	27
11	NOTCH2NLC CGG Repeats Are Not Expanded and Skin Biopsy Was Negative in an Infantile Patient With Neuronal Intranuclear Inclusion Disease. Journal of Neuropathology and Experimental Neurology, 2020, 79, 1065-1071.	1.7	8
12	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
13	Hepatic pathology and altered gene transcription in a murine model of acid ceramidase deficiency. Laboratory Investigation, 2019, 99, 1572-1592.	3.7	12
14	Acid Ceramidase Deficiency in Mice Leads to Severe Ocular Pathology and Visual Impairment. American Journal of Pathology, 2019, 189, 320-338.	3.8	16
15	Specific storage of glycoconjugates with terminal Î±-galactosyl moieties in the exocrine pancreas of Fabry disease patients with blood group B. Glycobiology, 2018, 28, 382-391.	2.5	5
16	Neural cells generated from human induced pluripotent stem cells as a model of CNS involvement in mucopolysaccharidosis type II. Journal of Inherited Metabolic Disease, 2018, 41, 221-229.	3.6	16
17	Chronic lung injury and impaired pulmonary function in a mouse model of acid ceramidase deficiency. American Journal of Physiology - Lung Cellular and Molecular Physiology, 2018, 314, L406-L420.	2.9	26
18	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

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19	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
20	Quantitation of plasmatic lysosphingomyelin and lysosphingomyelin-509 for differential screening of Niemann-Pick A/B and C diseases. Analytical Biochemistry, 2017, 525, 73-77.	2.4	49
21	Acid Ceramidase Deficiency in Mice Results in a Broad Range of Central Nervous System Abnormalities. American Journal of Pathology, 2017, 187, 864-883.	3.8	41
22	N-butyldeoxynojirimycin delays motor deficits, cerebellar microgliosis, and Purkinje cell loss in a mouse model of mucolipidosis type IV. Neurobiology of Disease, 2017, 105, 257-270.	4.4	13
23	Efficacy and ototoxicity of different cyclodextrins in Niemannâ€“Pick C disease. Annals of Clinical and Translational Neurology, 2016, 3, 366-380.	3.7	78
24	X-linked Christianson syndrome: heterozygous female Slc9a6 knockout mice develop mosaic neuropathological changes and related behavioral abnormalities. DMM Disease Models and Mechanisms, 2015, 9, 13-23.	2.4	19
25	A Murine Niemann-Pick C1 I1061T Knock-In Model Recapitulates the Pathological Features of the Most Prevalent Human Disease Allele. Journal of Neuroscience, 2015, 35, 8091-8106.	3.6	97
26	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. Journal of Cardiology, 2015, 66, 88-89.	1.9	6
27	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
28	Lysosomal Membrane Permeability Stimulates Protein Aggregate Formation in Neurons of a Lysosomal Disease. Journal of Neuroscience, 2013, 33, 10815-10827.	3.6	47
29	Mutations of ATIC and ADSL affect purinosome assembly in cultured skin fibroblasts from patients with AICA-ribosiduria and ADSL deficiency. Human Molecular Genetics, 2012, 21, 1534-1543.	2.9	62
30	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
31	Lysosomal compromise and brain dysfunction: examining the role of neuroaxonal dystrophy. Biochemical Society Transactions, 2010, 38, 1436-1441.	3.4	26
32	Bioinformatic and biochemical studies point to AAGR-1 as the ortholog of human acid Î±-glucosidase in CaenorhabditisÂelegans. Molecular and Cellular Biochemistry, 2010, 341, 51-63.	3.1	2
33	Cystathionine beta-synthase null homocystinuric mice fail to exhibit altered hemostasis or lowering of plasma homocysteine in response to betaine treatment. Molecular Genetics and Metabolism, 2010, 101, 163-171.	1.1	57
34	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. Molecular Genetics and Metabolism, 2010, 101, 153-162.	1.1	60
35	Clinical and molecular characterization of a family with a dominant renin gene mutation and response to treatment with fludrocortisone. Clinical Nephrology, 2010, 74, 411-422.	0.7	44
36	Dominant Renin Gene Mutations Associated with Early-Onset Hyperuricemia, Anemia, and Chronic Kidney Failure. American Journal of Human Genetics, 2009, 85, 204-213.	6.2	146

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37	Replacement of Î±-galactosidase A in Fabry disease: effect on fibroblast cultures compared with biopsied tissues of treated patients. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2008, 452, 651-665.	2.8	29
38	Neurolysosomal pathology in human prosaposin deficiency suggests essential neurotrophic function of prosaposin. Acta Neuropathologica, 2007, 113, 163-175.	7.7	30
39	Mutations in TMEM76* Cause Mucopolysaccharidosis IIIC (Sanfilippo C Syndrome). American Journal of Human Genetics, 2006, 79, 807-819.	6.2	77
40	Alterations of uromodulin biology: a common denominator of the genetically heterogeneous FJHN/MCKD syndrome. Kidney International, 2006, 70, 1155-1169.	5.2	111
41	Subclinical course of adult visceral Niemannâ€“Pick type C1 disease. A rare or underdiagnosed disorder?. Journal of Inherited Metabolic Disease, 2006, 29, 591-591.	3.6	25
42	Retrospective sequence analysis of the humanPRNP gene from the formaldehyde-fixed paraffin-embedded tissues: Report of two cases of creutzfeldt-jakob disease. Folia Microbiologica, 2006, 51, 619-625.	2.3	1
43	Acid sphingomyelinase deficiency. Phenotype variability with prevalence of intermediate phenotype in a series of twenty-five Czech and Slovak patients. A multi-approach study. Journal of Inherited Metabolic Disease, 2005, 28, 203-227.	3.6	85
44	Characterization of gana-1, a Caenorhabditis elegans gene encoding a single ortholog of vertebrate alpha-galactosidase and alpha-N-acetylgalactosaminidase. BMC Cell Biology, 2005, 6, 5.	3.0	13
45	Seven Novel Acid Sphingomyelinase Gene Mutations in Niemann-Pick Type A and B Patients. Annals of Human Genetics, 2003, 67, 63-70.	0.8	39