

# Gustavohenrique Maegawa

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/925125/publications.pdf>

Version: 2024-02-01

40  
papers

6,239  
citations

304743

22  
h-index

302126

39  
g-index

40  
all docs

40  
docs citations

40  
times ranked

15580  
citing authors

#	ARTICLE	IF	CITATIONS
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). <i>Autophagy</i> , 2016, 12, 1-222.	9.1	4,701
2	Identification and Characterization of Ambroxol as an Enzyme Enhancement Agent for Gaucher Disease. <i>Journal of Biological Chemistry</i> , 2009, 284, 23502-23516.	3.4	260
3	The Natural History of Juvenile or Subacute GM2 Gangliosidosis: 21 New Cases and Literature Review of 134 Previously Reported. <i>Pediatrics</i> , 2006, 118, e1550-e1562.	2.1	165
4	Pyrimethamine as a Potential Pharmacological Chaperone for Late-onset Forms of GM2 Gangliosidosis. <i>Journal of Biological Chemistry</i> , 2007, 282, 9150-9161.	3.4	152
5	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. <i>American Journal of Human Genetics</i> , 2016, 98, 347-357.	6.2	98
6	Pegunigalsidase alfa, a novel PEGylated enzyme replacement therapy for Fabry disease, provides sustained plasma concentrations and favorable pharmacodynamics: A 1-year Phase 1/2 clinical trial. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 534-544.	3.6	86
7	Novel parkin mutations detected in patients with early-onset Parkinson's disease. <i>Movement Disorders</i> , 2005, 20, 424-431.	3.9	60
8	Substrate reduction therapy in juvenile GM2 gangliosidosis. <i>Molecular Genetics and Metabolism</i> , 2009, 98, 215-224.	1.1	59
9	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. <i>American Journal of Human Genetics</i> , 2019, 104, 1210-1222.	6.2	56
10	Early axonal loss accompanied by impaired endocytosis, abnormal axonal transport, and decreased microtubule stability occur in the model of Krabbe's disease. <i>Neurobiology of Disease</i> , 2014, 66, 92-103.	4.4	55
11	Developing therapeutic approaches for metachromatic leukodystrophy. <i>Drug Design, Development and Therapy</i> , 2013, 7, 729.	4.3	42
12	<i>KCTD7</i> deficiency defines a distinct neurodegenerative disorder with a conserved autophagy-lysosome defect. <i>Annals of Neurology</i> , 2018, 84, 766-780.	5.3	42
13	MR surveillance of boys with X-linked adrenoleukodystrophy identified by newborn screening: Meta-analysis and consensus guidelines. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 728-739.	3.6	39
14	Expanding <i>CEP290</i> mutational spectrum in ciliopathies. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2173-2180.	1.2	38
15	CNS-Targeting Therapies for Lysosomal Storage Diseases: Current Advances and Challenges. <i>Frontiers in Molecular Biosciences</i> , 2020, 7, 559804.	3.5	38
16	Clinical Neurogenetics. <i>Neurologic Clinics</i> , 2013, 31, 1051-1071.	1.8	30
17	Characterization and application of a disease-cell model for a neurodegenerative lysosomal disease. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 172-183.	1.1	29
18	Pharmacokinetics, safety and tolerability of miglustat in the treatment of pediatric patients with GM2 gangliosidosis. <i>Molecular Genetics and Metabolism</i> , 2009, 97, 284-291.	1.1	28

#	ARTICLE	IF	CITATIONS
19	A high-throughput screening assay using Krabbe disease patient cells. <i>Analytical Biochemistry</i> , 2013, 434, 15-25.	2.4	26
20	Mucopolysaccharidoses type I and II: New neuroimaging findings in the cerebellum. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 211-217.	1.6	26
21	Antigen-encapsulating host extracellular vesicles derived from Salmonella-infected cells stimulate pathogen-specific Th1-type responses in vivo. <i>PLoS Pathogens</i> , 2021, 17, e1009465.	4.7	26
22	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 164-169.	1.1	25
23	Reduction of Plasma Globotriaosylsphingosine Levels After Switching from Agalsidase Alfa to Agalsidase Beta as Enzyme Replacement Therapy for Fabry Disease. <i>JIMD Reports</i> , 2015, 25, 95-106.	1.5	22
24	Intracerebral Periventricular Pseudocysts in a Fetus with Mitochondrial Depletion Syndrome: An Association or Coincidence. <i>Fetal Diagnosis and Therapy</i> , 2009, 25, 177-182.	1.4	18
25	Clinical, biochemical, and genotype-phenotype correlations of 118 patients with Niemann-Pick disease Types A/B. <i>Human Mutation</i> , 2021, 42, 614-625.	2.5	18
26	Comparative analysis of PCR-deletion detection and immunohistochemistry in Brazilian Duchenne and Becker muscular dystrophy patients. <i>American Journal of Medical Genetics Part A</i> , 2001, 103, 115-120.	2.4	15
27	Lysosomal Leukodystrophies Lysosomal Storage Diseases Associated With White Matter Abnormalities. <i>Journal of Child Neurology</i> , 2019, 34, 339-358.	1.4	14
28	Interstitial deletion of 1p22.2p31.1 and medium-chain acyl-CoA dehydrogenase deficiency in a patient with global developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1581-1586.	1.2	11
29	Novel Patient Cell-Based HTS Assay for Identification of Small Molecules for a Lysosomal Storage Disease. <i>PLoS ONE</i> , 2011, 6, e29504.	2.5	11
30	Ambroxol improves skeletal and hematological manifestations on a child with Gaucher disease. <i>Journal of Human Genetics</i> , 2020, 65, 345-349.	2.3	11
31	Impaired P50 sensory gating in Machado-Joseph disease. <i>Clinical Neurophysiology</i> , 2004, 115, 2231-2235.	1.5	10
32	Duodenal and biliary atresia associated with facial, thyroid and auditory apparatus abnormalities: a new mandibulofacial dysostosis syndrome?. <i>Clinical Dysmorphology</i> , 2006, 15, 191-196.	0.3	7
33	Novel proton MR spectroscopy findings in adenylosuccinate lyase deficiency. <i>Journal of Magnetic Resonance Imaging</i> , 2013, 37, 974-980.	3.4	7
34	GM2 gangliosidosis: the prototype of lysosomal storage disorders. <i>Developmental Medicine and Child Neurology</i> , 2012, 54, 104-105.	2.1	4
35	Serendipitous effects of Î²-cyclodextrin on murine model of Krabbe disease. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 15, 98-99.	1.1	4
36	Focal dermal hypoplasia associated with split sternum ??? Goltz syndrome. <i>Clinical Dysmorphology</i> , 2005, 14, 37-39.	0.3	2

#	ARTICLE	IF	CITATIONS
37	Cell-based high-throughput screening identifies galactocerebrosidase enhancers as potential small-molecule therapies for Krabbe's disease. <i>Journal of Neuroscience Research</i> , 2016, 94, 1231-1245.	2.9	2
38	Microphthalmia and linear skin defects syndrome: Precise diagnosis guides prognosis. <i>Pediatric Dermatology</i> , 2020, 37, 217-218.	0.9	1
39	Potential Disease-Modifying Effects of Lithium Carbonate in Niemann-Pick Disease, Type C1. <i>Frontiers in Pharmacology</i> , 2021, 12, 667361.	3.5	1
40	Premature Identical Twin Neonates With Sleep Apnea. <i>Clinical Pediatrics</i> , 2017, 56, 1075-1078.	0.8	0