Gustavohenrique Maegawa

List of Publications by Year
in descending order

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	<scp>MRI</scp> surveillance of boys with Xâ€linked adrenoleukodystrophy identified by newborn screening: Metaâ€analysis and consensus guidelines. Journal of Inherited Metabolic Disease, 2021, 44, 728-739.	3.6	39
2	Clinical, biochemical, and genotypeâ€phenotype correlations of 118 patients with Niemannâ€Pick disease Types A/B. Human Mutation, 2021, 42, 614-625.	2.5	18
3	Antigen-encapsulating host extracellular vesicles derived from Salmonella-infected cells stimulate pathogen-specific Th1-type responses in vivo. PLoS Pathogens, 2021, 17, e1009465.	4.7	26
4	Potential Disease-Modifying Effects of Lithium Carbonate in Niemann-Pick Disease, Type C1. Frontiers in Pharmacology, 2021, 12, 667361.	3.5	1
5	Microphthalmia and linear skin defects syndrome: Precise diagnosis guides prognosis. Pediatric Dermatology, 2020, 37, 217-218.	0.9	1
6	Ambroxol improves skeletal and hematological manifestations on a child with Gaucher disease. Journal of Human Genetics, 2020, 65, 345-349.	2.3	11
7	CNS-Targeting Therapies for Lysosomal Storage Diseases: Current Advances and Challenges. Frontiers in Molecular Biosciences, 2020, 7, 559804.	3.5	38
8	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. Molecular Genetics and Metabolism, 2020, 130, 164-169.	1.1	25
9	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
10	Pegunigalsidase alfa, a novel PEGylated enzyme replacement therapy for Fabry disease, provides sustained plasma concentrations and favorable pharmacodynamics: A 1â€year Phase 1/2 clinical trial. Journal of Inherited Metabolic Disease, 2019, 42, 534-544.	3.6	86
11	Lysosomal Leukodystrophies Lysosomal Storage Diseases Associated With White Matter Abnormalities. Journal of Child Neurology, 2019, 34, 339-358.	1.4	14
12	KCTD7 deficiency defines a distinct neurodegenerative disorder with a conserved autophagyâ€lysosome defect. Annals of Neurology, 2018, 84, 766-780.	5.3	42
13	Serendipitous effects of Î²-cyclodextrin on murine model of Krabbe disease. Molecular Genetics and Metabolism Reports, 2018, 15, 98-99.	1.1	4
14	Premature Identical Twin Neonates With Sleep Apnea. Clinical Pediatrics, 2017, 56, 1075-1078.	0.8	0
15	Cellâ€based highâ€throughput screening identifies galactocerebrosidase enhancers as potential smallâ€molecule therapies for <scp>K</scp>rabbe's disease. Journal of Neuroscience Research, 2016, 94, 1231-1245.	2.9	2
16	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
17	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
18	Reduction of Plasma Globotriaosylsphingosine Levels After Switching from Agalsidase Alfa to Agalsidase Beta as Enzyme Replacement Therapy for Fabry Disease. JIMD Reports, 2015, 25, 95-106.	1.5	22

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19	Early axonal loss accompanied by impaired endocytosis, abnormal axonal transport, and decreased microtubule stability occur in the model of Krabbe's disease. Neurobiology of Disease, 2014, 66, 92-103.	4.4	55
20	Mucopolysaccharidoses type I and II: New neuroimaging findings in the cerebellum. European Journal of Paediatric Neurology, 2014, 18, 211-217.	1.6	26
21	Characterization and application of a disease-cell model for a neurodegenerative lysosomal disease. Molecular Genetics and Metabolism, 2014, 111, 172-183.	1.1	29
22	Clinical Neurogenetics. Neurologic Clinics, 2013, 31, 1051-1071.	1.8	30
23	A high-throughput screening assay using Krabbe disease patient cells. Analytical Biochemistry, 2013, 434, 15-25.	2.4	26
24	Novel proton MR spectroscopy findings in adenylosuccinate lyase deficiency. Journal of Magnetic Resonance Imaging, 2013, 37, 974-980.	3.4	7
25	Developing therapeutic approaches for metachromatic leukodystrophy. Drug Design, Development and Therapy, 2013, 7, 729.	4.3	42
26	GM2 gangliosidosis: the prototype of lysosomal storage disorders. Developmental Medicine and Child Neurology, 2012, 54, 104-105.	2.1	4
27	Novel Patient Cell-Based HTS Assay for Identification of Small Molecules for a Lysosomal Storage Disease. PLoS ONE, 2011, 6, e29504.	2.5	11
28	Intracerebral Periventricular Pseudocysts in a Fetus with Mitochondrial Depletion Syndrome: An Association or Coincidence. Fetal Diagnosis and Therapy, 2009, 25, 177-182.	1.4	18
29	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
30	Pharmacokinetics, safety and tolerability of miglustat in the treatment of pediatric patients with GM2 gangliosidosis. Molecular Genetics and Metabolism, 2009, 97, 284-291.	1.1	28
31	Substrate reduction therapy in juvenile GM2 gangliosidosis. Molecular Genetics and Metabolism, 2009, 98, 215-224.	1.1	59
32	Identification and Characterization of Ambroxol as an Enzyme Enhancement Agent for Gaucher Disease. Journal of Biological Chemistry, 2009, 284, 23502-23516.	3.4	260
33	Interstitial deletion of 1p22.2p31.1 and medium-chain acyl-CoA dehydrogenase deficiency in a patient with global developmental delay. American Journal of Medical Genetics, Part A, 2008, 146A, 1581-1586.	1.2	11
34	Pyrimethamine as a Potential Pharmacological Chaperone for Late-onset Forms of GM2 Gangliosidosis. Journal of Biological Chemistry, 2007, 282, 9150-9161.	3.4	152
35	Duodenal and biliary atresia associated with facial, thyroid and auditory apparatus abnormalities: a new mandibulofacial dysostosis syndrome?. Clinical Dysmorphology, 2006, 15, 191-196.	0.3	7
36	The Natural History of Juvenile or Subacute GM2 Gangliosidosis: 21 New Cases and Literature Review of 134 Previously Reported. Pediatrics, 2006, 118, e1550-e1562.	2.1	165

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37	Focal dermal hypoplasia associated with split sternum ??? Goltz syndrome. Clinical Dysmorphology, 2005, 14, 37-39.	0.3	2
38	Novel parkin mutations detected in patients with early-onset Parkinson's disease. Movement Disorders, 2005, 20, 424-431.	3.9	60
39	Impaired P50 sensory gating in Machado-Joseph disease. Clinical Neurophysiology, 2004, 115, 2231-2235.	1.5	10
40	Comparative analysis of PCR-deletion detection and immunohistochemistry in Brazilian Duchenne and Becker muscular dystrophy patients. American Journal of Medical Genetics Part A, 2001, 103, 115-120.	2.4	15