May Christine V Malicdan

List of Publications by Year
in descending order

60	4,979	304743 22	149698 56
papers	citations	h-index	g-index

64	64	64	12520
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	9.1	3,122
2	Prophylactic treatment with sialic acid metabolites precludes the development of the myopathic phenotype in the DMRV-hIBM mouse model. Nature Medicine, 2009, 15, 690-695.	30.7	167
3	A Gne knockout mouse expressing human GNE D176V mutation develops features similar to distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy. Human Molecular Genetics, 2007, 16, 2669-2682.	2.9	136
4	Lysosomal myopathies: An excessive build-up in autophagosomes is too much to handle. Neuromuscular Disorders, 2008, 18, 521-529.	0.6	136
5	A Gne knockout mouse expressing human V572L mutation develops features similar to distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy. Human Molecular Genetics, 2007, 16, 115-128.	2.9	111
6	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
7	Mutation Update forGNEGene Variants Associated with GNE Myopathy. Human Mutation, 2014, 35, 915-926.	2.5	90
8	Autophagy in Lysosomal Myopathies. Brain Pathology, 2012, 22, 82-88.	4.1	67
9	Spermine synthase deficiency causes lysosomal dysfunction and oxidative stress in models of Snyder-Robinson syndrome. Nature Communications, 2017, 8, 1257.	12.8	64
10	GNE Myopathy: Etiology, Diagnosis, and Therapeutic Challenges. Neurotherapeutics, 2018, 15, 900-914.	4.4	63
11	Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. American Journal of Human Genetics, 2015, 97, 99-110.	6.2	61
12	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
13	Autophagy in a Mouse Model of Distal Myopathy with Rimmed Vacuoles or Hereditary Inclusion Body Myopathy. Autophagy, 2007, 3, 396-398.	9.1	49
14	Mutations in human homologue of chickentalpid3gene (KIAA0586) cause a hybrid ciliopathy with overlapping features of Jeune and Joubert syndromes. Journal of Medical Genetics, 2015, 52, 830-839.	3.2	47
15	A novel inborn error of the coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 Câ€methyltransferase deficiency. Human Mutation, 2018, 39, 69-79.	2.5	43
16	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
17	Safety, pharmacokinetics and sialic acid production after oral administration of N -acetylmannosamine (ManNAc) to subjects with GNE myopathy. Molecular Genetics and Metabolism, 2017, 122, 126-134.	1.1	41
18	Peracetylated N-Acetylmannosamine, a Synthetic Sugar Molecule, Efficiently Rescues Muscle Phenotype and Biochemical Defects in Mouse Model of Sialic Acid-deficient Myopathy. Journal of Biological Chemistry, 2012, 287, 2689-2705.	3.4	40

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19	Congenital protein losing enteropathy: an inborn error of lipid metabolism due to DGAT1 mutations. European Journal of Human Genetics, 2016, 24, 1268-1273.	2.8	37
20	Muscle weakness correlates with muscle atrophy and precedes the development of inclusion body or rimmed vacuoles in the mouse model of DMRV/hIBM. Physiological Genomics, 2008, 35, 106-115.	2.3	36
21	Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. Human Genetics, 2017, 136, 399-408.	3.8	30
22	Mitochondrial epileptic encephalopathy, 3â€methylglutaconic aciduria and variable complex V deficiency associated with <scp>TIMM50</scp> mutations. Clinical Genetics, 2017, 91, 690-696.	2.0	28
23	Clinical and molecular phenotyping of a child with Hermansky-Pudlak syndrome-7, an uncommon genetic type of HPS. Molecular Genetics and Metabolism, 2017, 120, 378-383.	1.1	25
24	CELSR2, encoding a planar cell polarity protein, is a putative gene in Joubert syndrome with cortical heterotopia, microophthalmia, and growth hormone deficiency. American Journal of Medical Genetics, Part A, 2017, 173, 661-666.	1.2	25
25	Combined alpha-delta platelet storage pool deficiency is associated with mutations in GFI1B. Molecular Genetics and Metabolism, 2017, 120, 288-294.	1.1	22
26	Exome analysis of Smithâ€“Magenis-like syndrome cohort identifies de novo likely pathogenic variants. Human Genetics, 2017, 136, 409-420.	3.8	22
27	Defective ciliogenesis in INPP5Eâ€related Joubert syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3231-3237.	1.2	22
28	Characteristics of Liver Disease in 100 Individuals With Joubert Syndrome Prospectively Evaluated at a Single Center. Journal of Pediatric Gastroenterology and Nutrition, 2018, 66, 428-435.	1.8	21
29	Muscle Weakness and Fibrosis Due to Cell Autonomous and Non-cell Autonomous Events in Collagen VI Deficient Congenital Muscular Dystrophy. EBioMedicine, 2017, 15, 193-202.	6.1	19
30	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	6.2	18
31	Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. American Journal of Human Genetics, 2018, 103, 794-807.	6.2	18
32	Allele-specific Gene Silencing of Mutant mRNA Restores Cellular Function in Ullrich Congenital Muscular Dystrophy Fibroblasts. Molecular Therapy - Nucleic Acids, 2014, 3, e171.	5.1	17
33	Bleomycin Induces Drug Efflux in Lungs. A Pitfall for Pharmacological Studies of Pulmonary Fibrosis. American Journal of Respiratory Cell and Molecular Biology, 2020, 62, 178-190.	2.9	16
34	A preclinical trial of sialic acid metabolites on distal myopathy with rimmed vacuoles/ hereditary inclusion body myopathy, a sugar-deficient myopathy: a review. Therapeutic Advances in Neurological Disorders, 2010, 3, 127-135.	3.5	15
35	Identification of an Alu elementâ€mediated deletion in the promoter region of <scp>GNE</scp> in siblings with <scp>GNE</scp> myopathy. Molecular Genetics & Genomic Medicine, 2017, 5, 410-417.	1.2	15
36	Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. Molecular Genetics and Metabolism, 2020, 130, 49-57.	1.1	15

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37	Late diagnosis and atypical brain imaging of Aicardiâ€“GoutiÃ¨res syndrome: are we failing to diagnose Aicardiâ€“GoutiÃ¨res syndromeâ€?. Developmental Medicine and Child Neurology, 2017, 59, 1307-1311.	2.1	14
38	Quantification of lectin fluorescence in GNE myopathy muscle biopsies. Muscle and Nerve, 2018, 58, 286-292.	2.2	13
39	Loss of function mutations in VARS encoding cytoplasmic valyl-tRNA synthetase cause microcephaly, seizures, and progressive cerebral atrophy. Human Genetics, 2018, 137, 293-303.	3.8	12
40	Cellular and molecular defects in a patient with Hermansky-Pudlak syndrome type 5. PLoS ONE, 2017, 12, e0173682.	2.5	11
41	yippee like 3Â(ypel3) is a novel gene required for myelinating and perineurial glia development. PLoS Genetics, 2020, 16, e1008841.	3.5	11
42	Novel mutations in CLN6 cause late-infantile neuronal ceroid lipofuscinosis without visual impairment in two unrelated patients. Molecular Genetics and Metabolism, 2019, 126, 188-195.	1.1	10
43	Identification of a novel mutation in HPS6 in a patient with hemophilia B and oculocutaneous albinism. Molecular Genetics and Metabolism, 2016, 119, 284-287.	1.1	9
44	Early infantile-onset epileptic encephalopathy 28 due to a homozygous microdeletion involving the WWOX gene in a region of uniparental disomy. Human Mutation, 2019, 40, 42-47.	2.5	8
45	Glycomics in rare diseases: from diagnosis tomechanism. Translational Research, 2019, 206, 5-17.	5.0	8
46	Severe bleeding with subclinical oculocutaneous albinism in a patient with a novel HPS6 missense variant. American Journal of Medical Genetics, Part A, 2018, 176, 2819-2823.	1.2	7
47	Cortical atrophy and hypofibrinogenemia due to FGG and TBCD mutations in a single family: a case report. BMC Medical Genetics, 2018, 19, 80.	2.1	7
48	Deficiency in the endocytic adaptor proteins PHETA1/2 impair renal and craniofacial development. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	7
49	PUS7 deficiency in human patients causes profound neurodevelopmental phenotype by dysregulating protein translation. Molecular Genetics and Metabolism, 2022, 135, 221-229.	1.1	6
50	Generation and characterization of four Chediak-Higashi Syndrome (CHS) induced pluripotent stem cell (iPSC) lines. Stem Cell Research, 2020, 47, 101883.	0.7	5
51	Diagnosis of Chediak Higashi disease in a 67â€year old woman. American Journal of Medical Genetics, Part A, 2020, 182, 3007-3013.	1.2	4
52	Novel truncating mutation in TENM3 in siblings with motor developmental delay, ocular coloboma, oval cornea, without microphthalmia. American Journal of Medical Genetics, Part A, 2018, 176, 2930-2933.	1.2	3
53	Biallelic variants in two complex I genes cause abnormal splicing defects in probands with mild Leigh syndrome. Molecular Genetics and Metabolism, 2020, 131, 98-106.	1.1	2
54	Diagnosis and discovery: Insights from the <scp>NIH</scp> Undiagnosed Diseases Program. Journal of Inherited Metabolic Disease, 2022, 45, 907-918.	3.6	2

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55	Clinical, biochemical and genetic characteristics of MOGS-CDG: a rare congenital disorder of glycosylation. Journal of Medical Genetics, 2022, 59, 1104-1115.	3.2	2
56	Progressive pulmonary fibrosis in a murine model of Hermansky-Pudlak syndrome. Respiratory Research, 2022, 23, 112.	3.6	1
57	Cover Image, Volume 173A, Number 12, December 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
58	Metabolic changes in sialic acid synthesis pathway in GNEâ€myopathy model mice with longâ€term sialic acid treatment. FASEB Journal, 2012, 26, 551.5.	0.5	0
59	Identification of biomarkers for GNE myopathy. FASEB Journal, 2012, 26, 1122.3.	0.5	0
60	Novel Hermanksky-Pudlak Syndrome Type 6 Missense Variant Associated with Subclinical Oculocutaneous Albinism and Mild Bleeding. Blood, 2018, 132, 1153-1153.	1.4	0