# 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 | PUS7 deficiency in human patients causes profound neurodevelopmental phenotype by dysregulating protein translation. Molecular Genetics and Metabolism, 2022, 135, 221-229. | 1.1 | 6 |
| 2 | Diagnosis and discovery: Insights from the <scp>NIH</scp> Undiagnosed Diseases Program. Journal of Inherited Metabolic Disease, 2022, 45, 907-918. | 3.6 | 2 |
| 3 | Progressive pulmonary fibrosis in a murine model of Hermansky-Pudlak syndrome. Respiratory Research, 2022, 23, 112. | 3.6 | 1 |
| 4 | 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 |
| 5 | 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 |
| 6 | 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 |
| 7 | yippee like 3Â(ypel3) is a novel gene required for myelinating and perineurial glia development. PLoS Genetics, 2020, 16, e1008841. | 3.5 | 11 |
| 8 | Generation and characterization of four Chediak-Higashi Syndrome (CHS) induced pluripotent stem cell (iPSC) lines. Stem Cell Research, 2020, 47, 101883. | 0.7 | 5 |
| 9 | Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. Molecular Genetics and Metabolism, 2020, 130, 49-57. | 1.1 | 15 |
| 10 | Deficiency in the endocytic adaptor proteins PHETA1/2 impair renal and craniofacial development. DMM Disease Models and Mechanisms, 2020, 13, . | 2.4 | 7 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | 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 |
| 15 | Glycomics in rare diseases: from diagnosis tomechanism. Translational Research, 2019, 206, 5-17. | 5.0 | 8 |
| 16 | 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 |
| 17 | Quantification of lectin fluorescence in GNE myopathy muscle biopsies. Muscle and Nerve, 2018, 58, 286-292. | 2.2 | 13 |
| 18 | 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 |

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|---|---|---|---|
| 19 | 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 |
| 20 | 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 |
| 21 | 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 |
| 22 | 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 |
| 23 | 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 |
| 24 | GNE Myopathy: Etiology, Diagnosis, and Therapeutic Challenges. Neurotherapeutics, 2018, 15, 900-914. | 4.4 | 63 |
| 25 | 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 |
| 26 | 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 |
| 27 | Novel Hermanksky-Pudlak Syndrome Type 6 Missense Variant Associated with Subclinical Oculocutaneous Albinism and Mild Bleeding. Blood, 2018, 132, 1153-1153. | 1.4 | 0 |
| 28 | Combined alpha-delta platelet storage pool deficiency is associated with mutations in GFI1B. Molecular Genetics and Metabolism, 2017, 120, 288-294. | 1.1 | 22 |
| 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 | Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. Human Genetics, 2017, 136, 399-408. | 3.8 | 30 |
| 31 | Exome analysis of Smithâ€“Magenis-like syndrome cohort identifies de novo likely pathogenic variants. Human Genetics, 2017, 136, 409-420. | 3.8 | 22 |
| 32 | 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 |
| 33 | A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137. | 6.2 | 96 |
| 34 | 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 |
| 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 | Late diagnosis and atypical brain imaging of Aicardiâ€“GoutiÃ¨res syndrome: are we failing to diagnose Aicardiâ€“GoutiÃ¨res syndromeâ€2?. Developmental Medicine and Child Neurology, 2017, 59, 1307-1311. | 2.1 | 14 |

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|---|---|---|---|
| 37 | Cover Image, Volume 173A, Number 12, December 2017. American Journal of Medical Genetics, Part A, 2017, 173, i. | 1.2 | 0 |
| 38 | Defective ciliogenesis in INPP5Eâ€related Joubert syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3231-3237. | 1.2 | 22 |
| 39 | Spermine synthase deficiency causes lysosomal dysfunction and oxidative stress in models of Snyder-Robinson syndrome. Nature Communications, 2017, 8, 1257. | 12.8 | 64 |
| 40 | 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 |
| 41 | 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 |
| 42 | Cellular and molecular defects in a patient with Hermansky-Pudlak syndrome type 5. PLoS ONE, 2017, 12, e0173682. | 2.5 | 11 |
| 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 | 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 |
| 45 | 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 |
| 46 | 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 |
| 47 | 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 |
| 48 | Mutation Update forGNEGene Variants Associated with GNE Myopathy. Human Mutation, 2014, 35, 915-926. | 2.5 | 90 |
| 49 | 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 |
| 50 | Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544. | 9.1 | 3,122 |
| 51 | Autophagy in Lysosomal Myopathies. Brain Pathology, 2012, 22, 82-88. | 4.1 | 67 |
| 52 | 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 |
| 53 | Identification of biomarkers for GNE myopathy. FASEB Journal, 2012, 26, 1122.3. | 0.5 | 0 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | Lysosomal myopathies: An excessive build-up in autophagosomes is too much to handle. Neuromuscular Disorders, 2008, 18, 521-529. | 0.6 | 136 |
| 57 | 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 |
| 58 | 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 |
| 59 | Autophagy in a Mouse Model of Distal Myopathy with Rimmed Vacuoles or Hereditary Inclusion Body Myopathy. Autophagy, 2007, 3, 396-398. | 9.1 | 49 |
| 60 | 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 |