May Christine V. Malicdan, MD, PhD

Office of the Clinical Director and the NIH
Undiagnosed Diseases Program
malicdanm@mail.nih.gov

EDUCATION

PhD in Neuroscience
Kyoto University, Kyoto, Japan (2007 - 2010)

Residency in Neurology and Psychiatry
University of Santo Tomas Hospital, Manila, Philippines (2002 - 2006)

Doctor of Medicine
Saint Louis University, Baguio City, Philippines (1994 - 1999)

Bachelor of Science, Nursing
Saint Louis University, Baguio City, Philippines (1990 - 1994)

TEACHING EXPERIENCE

HONORS AND DISTINCTIONS

2019 – NIH Director's Award, as part of the UDN Leadership Committee

2018 – Mentorship award, NHGRI Intramural award

2016 – Doctor of the Year award – Hermansky-Pudlak Syndrome annual meeting

2015 - Elsevier Award for Best Oral presentation, 15th World Muscle Society, Kumamoto, Japan

2009 - Elsevier Award for Best Oral presentation, 14th World Muscle Society, Geneva, Switzerland

2006 - Elsevier Award for Best Poster presentation, 11th World Muscle Society, Brugge, Belgium

2004 - Excellence in Neurology Training Award, UST, Manila, Philippines

PROFESSIONAL MEMBERSHIP

2006 - Present World Muscle Society (WMS)

2006 - Present Asian Oceanian Myology Conference (AOMC)

2007 - Present American Society of Cell Biology (ASCB)

2011 - Present Society for Glycobiology

2012 - Present American Society of Human Genetics (ASHG)

2017 - Present American Society of Gene & Cell Therapy (ASGCT)

PROFESSIONAL ACTIVITIES

Director, NIH UDP Translational Laboratory 2019 - Present

Deputy Director of the NIH UDP Translational Laboratory 2018 - 2019

Staff Scientist 2015 - Present
NIH Undiagnosed Diseases Program
National Institutes of Health, Bethesda, MD
Adjunct Professor, Anatomy and Physiology Jul 2016 - Present
Howard Community College
Howard, MD, USA

Postdoctoral Visiting Fellow
Office of the Clinical Director and the NIH Undiagnosed Diseases Program, Gahl Lab
Bethesda, MA, USA Mar 2011 - Dec 2015

Postdoctoral Visiting Fellow
Department of Neuromuscular Research, NIN, NCNP, Nishino Lab
Tokyo, Japan USA Jan 2010 - Mar 2011

Neuromuscular Research Fellow
Department of Neuromuscular Research, NIN, NCNP, Nishino Lab
Tokyo, Japan USA Dec 2006 - Jan 2010

Vice-Chair 2021 - present
Animal Care and Use Committee, NHGRI

Co-Chair 2021 - present
Undiagnosed Diseases Network International Functional Study Working Group

Co-Chair 2015 - present
Undiagnosed Diseases Network Model Organisms Working Group

Steering Committee member 2015 - Present
Undiagnosed Disease Network

ACUC Member 2017 – present
NHGRI Animal and Care Use Committee

NHGRI Advisory Committee 2016 - present
Embryonic Stem Cell and Transgenic Mouse Core

Steering Committee member 2014 - present
Clinical Glycoscience Group

Ad Hoc Reviewer – Manuscripts 2008 - present
New England Journal of Medicine, Nature Communications, Cell Reports, Journal of Experimental Medicine, Neuromuscular Disorders, Biomed Central Medical Genetics, Biomed Central Journal series, Acta Neuropathologica, Acta Neuropathologica Communications, PLoS ONE, Rubriq, Brain
Ad Hoc Reviewer – Neuromuscular Clinical cases, NIH Undiagnosed Disease Program 2014 - 2016
Associate Editor – Manuscripts, Biomed Central Musculoskeletal Disorders 2014 - present
Grant Reviewer
AMED, Japan 2018 - present
Telethon 2016 - present

PUBLICATIONS

1. Shimada S, Ng BG, White AL, Nickander KK, Turgeon C, Liedtke KL, Lam CT, Font-Montgomery E, Lourenco CM, He M, Peck DS, Umana LA, Uhles CL, Haynes D, Wheeler PG, Bamshad MJ, Nickerson DA, Cushing T, Gates R, Gomez-Ospina N, Byers HM; UW Center for Mendelian Genomics, Scalco FB, Martinez NN, Sachdev R,

Smith L, Poduri A, Malone S, Harris RV, Scheffer IE, Rosenzweig SD, Adams DR, Gahl WA, Malicdan MCV, Raymond KM, Freeze HH, Wolfe LA. Clinical, biochemical and genetic characteristics of MOGS-CDG: a rare congenital disorder of glycosylation. **J Med Genet**. 2022 Jul 5:jmedgenet-2021-108177. doi: 10.1136/jmedgenet-2021-108177. PMID: 35790351.

2. Imani J, Bodine SPM, Lamattina AM, Ma DD, Shrestha S, Maynard DM, Bishop K, Nwokeji A, Malicdan MCV, Testa LC, Sood R, Stump B, Rosas IO, Perrella MA, Handin R, Young LR, Gochuico BR, El-Chemaly S. Dysregulated myosin in Hermansky-Pudlak syndrome lung fibroblasts is associated with increased cell motility. **Respir Res**. 2022 Jun 23;23(1):167. doi: 10.1186/s12931-022-02083-w. PMID: 35739508.
3. Abudi-Sinreich S, Bodine SP, Yokoyama T, Tolman NJ, Tyrlik M, Testa LC, Han CG, Dorward HM, Wincovitch SM, Anikster Y, Gahl WA, Cinar R, Gochuico BR, Malicdan MCV. Progressive pulmonary fibrosis in a murine model of Hermansky-Pudlak syndrome. **Respir Res**. 2022 May 4;23(1):112. doi: 10.1186/s12931-022-02002-z. PMID: 35509004.
4. Montano C, Cassini T, Ziegler SG, Boehm M, Nicoli ER, Mindell JA, Soldatos AG, Manoli I, Wolfe L, Macnamara EF, Malicdan MCV, Adams DR, Tifft CJ, Toro C, Gahl WA. Diagnosis and discovery: Insights from the NIH Undiagnosed Diseases Program. **J Inherit Metab Dis**. 2022 Apr 30. doi: 10.1002/jimd.12506. PMID 35490291.
5. Forsyth R, Parisi MA, Altintas B, Malicdan MC, Vilboux T, Knoll J, Brooks BP, Zein WM, Gahl WA, Toro C, Gunay-Aygun M. Systematic analysis of physical examination characteristics of 94 individuals with Joubert syndrome: Keys to suspecting the diagnosis. **Am J Med Genet C Semin Med Genet**. 2022 Mar;190(1):121-130. doi: 10.1002/ajmg.c.31966. PMID 35312150.
6. Han ST, Kim AC, Garcia K, Schimmenti LA, Macnamara E, Network UD, Gahl WA, Malicdan MC, Tifft CJ. PUS7 deficiency in human patients causes profound neurodevelopmental phenotype by dysregulating protein translation. **Mol Genet Metab**. 2022 Mar;135(3):221-229. doi: 10.1016/j.ymgme.2022.01.103. Epub 2022 Feb 1. PMID: 35144859.
7. Mota A, Waxman HK, Hong R, Lagani GD, Niu SY, Bertherat FL, Wolfe L, Malicdan CM, Markello TC, Adams DR, Gahl WA, Cheng CS, Beffert U, Ho A. FOXR1 regulates stress response pathways and is necessary for proper brain development. **PLoS Genet**. 2021 Nov 1;17(11):e1009854. doi: 10.1371/journal.pgen.1009854. eCollection 2021 Nov. PMID: 34723967.
8. Cinar R, Park JK, Zawatsky CN, Coffey NJ, Bodine SP, Abdalla J, Yokoyama T, Jourdan T, Jay L, Zuo MXG, O'Brien KJ, Huang J, Mackie K, Alimardanov A, Iyer MR, Gahl WA, Kunos G, Gochuico BR, Malicdan MCV. CB1 R and iNOS are distinct players promoting pulmonary fibrosis in Hermansky-Pudlak syndrome. **Clin Transl Med**. 2021 Jul;11(7):e471. doi: 10.1002/ctm2.471. PMID: 34323400
9. Boulin T, Itani O, El Mouridi S, Leclercq-Blondel A, Gendrel M, Macnamara E, Soldatos A, Murphy JL, Gorman MP, Lindsey A, Shimada S, Turner D, Silverman GA, Baldridge D; Undiagnosed Diseases Network, Malicdan MC, Schedl T, Pak SC. Functional analysis of a de novo variant in the neurodevelopment and generalized epilepsy disease gene NBEA. **Mol Genet Metab**. 2021 Sep-Oct;134(1-2):195-202. doi: 10.1016/j.ymgme.2021.07.013. Epub 2021 Aug 9. PMID: 34412939.
10. Carrillo N, Malicdan MC, Leoyklang P, Shrader JA, Joe G, Slota C, Perreault J, Heiss JD, Class B, Liu CY, Bradley K, Jodarski C, Ciccone C, Driscoll C, Parks R, Van Wart S, Bayman L, Coffey CS, Quintana M, Berry SM, Huizing M, Gahl WA. **Genet Med**. 2021 Nov;23(11):2067-2075. doi: 10.1038/s41436-021-01259-x. Epub 2021 Jul 13. PMID: 34257421
11. Testa LC, Jule Y, Lundh L, Bertotti K, Merideth MA, O'Brien KJ, Nathan SD, Venuto DC, El-Chemaly S, Malicdan MCV, Gochuico BR. Automated Digital Quantification of Pulmonary Fibrosis in Human Histopathology Specimens. **Front Med (Lausanne)**. 2021 Jun 15;8:607720. doi: 10.3389/fmed.2021.607720. eCollection 2021. PMID: 34211981.
12. Schoch K, Esteves C, Bican A, Spillmann R, Cope H, McConkie-Rosell A, Walley N, Fernandez L, Kohler JN, Bonner D, Reuter C, Stong N, Mulvihill JJ, Novacic D, Wolfe L, Abdelbaki A, Toro C, Tifft C, Malicdan M, Gahl W, Liu P, Newman J, Goldstein DB, Hom J, Sampson J, Wheeler MT, Undiagnosed Diseases N, Cogan J, Barnstein JA, Adams DA, McCray AT, Shashi V. Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. **Genetics in medicine : official journal of the American College of Medical Genetics** 2021;23(2):259-71.
13. O'Brien KJ, Parisi X, Shelman NR, Merideth MA, Introne WJ, Heller T, Gahl WA, Malicdan MCV, Gochuico BR. Inflammatory bowel disease in Hermansky-Pudlak syndrome: a retrospective single-centre cohort study. **J Intern Med** 2021.

14. Burke EA, Sturgeon M, Zastrow DB, Fernandez L, Prybol C, Marwaha S, Frothingham EP, Ward PA, Eng CM, Fresard L, Montgomery SB, Enns GM, Fisher PG, Wolfe LA, Harding B, Carrington B, Bishop K, Sood R, Huang Y, Elkahloun A, Toro C, Bassuk AG, Wheeler MT, Markello TC, Gahl WA, Malicdan MCV. Compound heterozygous KCTD7 variants in progressive myoclonus epilepsy. **J Neurogenet** 2021:1-10.
15. Huizing M, Hackbarth ME, Adams DR, Wasserstein M, Patterson MC, Walkley SU, Gahl WA, Consortium F, Adams DR, Dobrenis K, Foglio J, Gahl WA, Gasnier B, Hackbarth M, Huizing M, Lek M, Malicdan MCV, Paavola LE, Patterson MC, Reimer R, Walkley SU, Wasserstein M, Wang RY, Zoncu R. Free sialic acid storage disorder: Progress and promise. **Neurosci Lett** 2021;755:135896.
16. Nistala H, Dronzek J, Gonzaga-Jauregui C, Chim SM, Rajamani S, Nuwayhid S, Delgado D, Burke E, Karaca E, Franklin MC, Sarangapani P, Podgorski M, Tang Y, Dominguez MG, Withers M, Deckelbaum RA, Scheonherr CJ, Gahl WA, Malicdan MC, Zambrowicz B, Gale NW, Gibbs RA, Chung WK, Lupski JR, Economides AN. NMIHBA results from hypomorphic PRUNE1 variants that lack short-chain exopolyphosphatase activity. **Hum Mol Genet** 2021;29(21):3516-31
17. Beck DB, Ferrada MA, Sikora KA, Ombrello AK, Collins JC, Pei W, Balanda N, Ross DL, Ospina Cardona D, Wu Z, Patel B, Manthiram K, Groarke EM, Gutierrez-Rodrigues F, Hoffmann P, Rosenzweig S, Nakabo S, Dillon LW, Hourigan CS, Tsai WL, Gupta S, Carmona-Rivera C, Asmar AJ, Xu L, Oda H, Goodspeed W, Barron KS, Nehrebecky M, Jones A, Laird RS, Deuitch N, Rowczenio D, Rominger E, Wells KV, Lee C-CR, Wang W, Trick M, Mullikin J, Wigerblad G, Brooks S, Dell'Orso S, Deng Z, Chae JJ, Dulau-Florea A, Malicdan MCV, Novacic D, Colbert RA, Kaplan MJ, Gadina M, Savic S, Lachmann HJ, Abu-Asab M, Solomon BD, Retterer K, Gahl WA, Burgess SM, Aksentijevich I, Young NS, Calvo KR, Werner A, Kastner DL, Grayson PC. Somatic Mutations in UBA1 and Severe Adult-Onset Autoinflammatory Disease. **N Engl J Med** 2020;383(27):2628-38.
18. Johnstone T, Wang J, Ross D, Balanda N, Huang Y, Godfrey R, Groden C, Barton BR, Gahl W, Toro C, Malicdan MCV. Biallelic variants in two complex I genes cause abnormal splicing defects in probands with mild Leigh syndrome. **Mol Genet Metab** 2020;131(1-2):98-106.
19. Sharma P, Nicoli E-R, Serra-Vinardell J, Morimoto M, Toro C, Malicdan MCV, Introne WJ. Chediak-Higashi syndrome: a review of the past, present, and future. **Drug Discov Today Dis Models** 2020;31:31-36.
20. Yarnell DS, Roney JC, Teixeira C, Freitas MI, Cipriano A, Leuschner P, Krzewski K, Stephen J, Dorward H, Gahl WA, Gochuico BR, Toro C, Malicdan MC, Introne WJ. Diagnosis of Chediak Higashi disease in a 67-year old woman. **Am J Med Genet A** 2020;182(12):3007-13.
21. Tambe MA, Ng BG, Shimada S, Wolfe LA, Adams DR, Undiagnosed Diseases N, Gahl WA, Bamshad MJ, Nickerson DA, Malicdan MCV, Freeze HH. Mutations in GET4 disrupt the transmembrane domain recognition complex pathway. **J Inherit Metab Dis** 2020.
22. Serra-Vinardell J, Sandler MB, Pak E, Zheng W, Dutra A, Introne W, Gahl WA, Malicdan MC. Generation and characterization of four Chediak-Higashi Syndrome (CHS) induced pluripotent stem cell (iPSC) lines. **Stem Cell Res** 2020;47:101883.
23. Park JK, Coffey NJ, Bodine SP, Zawatsky CN, Jay L, Gahl WA, Kunos G, Gochuico BR, Malicdan MCV, Cinar R. Bleomycin Induces Drug Efflux in Lungs. A Pitfall for Pharmacological Studies of Pulmonary Fibrosis. Am **J Respir Cell Mol Biol** 2020;62(2):178-90. PMCID: PMC6993545.
24. Huizing M, Malicdan MCV, Wang JA, Pri-Chen H, Hess RA, Fischer R, O'Brien KJ, Merideth MA, Gahl WA, Gochuico BR. Hermansky-Pudlak syndrome: Mutation update. **Hum Mutat** 2020;41(3):543-80.
25. Gochuico BR, Ziegler SG, Ten NS, Balanda NJ, Mason CE, Zumbo P, Evans CA, Van Waes C, Gahl WA, Malicdan MCV. A comprehensive, multidisciplinary, precision medicine approach to discover effective therapy for an undiagnosed, progressive, fibroinflammatory disease. **Transl Res** 2020;215:31-40. PMCID: PMC6939610.
26. Davids M, Menezes M, Guo Y, McLean SD, Hakonarson H, Collins F, Worgan L, Billington CJ, Jr., Maric I, Littlejohn RO, Onyekweli T, Members Of The U, Adams DR, Tifft CJ, Gahl WA, Wolfe LA, Christodoulou J, Malicdan MCV. Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency. **Mol Genet Metab** 2020;130(1):49-57. PMCID: PMC7303973.
27. Blanco-Sanchez B, Clement A, Stednitz SJ, Kyle J, Peirce JL, McFadden M, Wegner J, Phillips JB, Macnamara E, Huang Y, Adams DR, Toro C, Gahl WA, Malicdan MCV, Tifft CJ, Zink EM, Bloodsworth KJ, Stratton KG, Undiagnosed Diseases N, Koeller DM, Metz TO, Washbourne P, Westerfield M. yippee like 3 (ypel3) is a novel gene required for myelinating and perineurial glia development. **PLoS Genet** 2020;16(6):e1008841. PMCID: PMC7319359.
28. Ates KM, Wang T, Moreland T, Veeranan-Karmegam R, Ma M, Jeter C, Anand P, Wenzel W, Kim HG, Wolfe LA, Stephen J, Adams DR, Markello T, Tifft CJ, Settlage R, Gahl WA, Gonsalvez GB, Malicdan MC, Flanagan-Steet H, Pan YA. Deficiency in the endocytic adaptor proteins PHETA1/2 impairs renal and craniofacial development. **Dis Model Mech** 2020;13(5). PMCID: PMC7272357.
29. Carrillo N, Malicdan MC, Huizing M. GNE Myopathy. In: Adam MP, Ardinger HH, Pagon RA, et al., eds. **GeneReviews**((R)). Seattle (WA), 1993.

30. Tian E, Wang S, Zhang L, Zhang Y, Malicdan MC, Mao Y, Christoffersen C, Tabak LA, Schjoldager KT, Ten Hagen KG. Galnt11 regulates kidney function by glycosylating the endocytosis receptor megalin to modulate ligand binding. **Proc Natl Acad Sci U S A** 2019;116(50):25196-202. PMCID: PMC6911204.
31. Sharma P, Reichert M, Lu Y, Markello TC, Adams DR, Steinbach PJ, Fuqua BK, Parisi X, Kaler SG, Vulpe CD, Anderson GJ, Gahl WA, Malicdan MCV. Biallelic HEPHL1 variants impair ferroxidase activity and cause an abnormal hair phenotype. **PLoS Genet 2019**;15(5):e1008143. PMCID: PMC6534290.
32. Rodriguez-Gil JL, Watkins-Chow DE, Baxter LL, Yokoyama T, Zerfas PM, Starost MF, Gahl WA, Malicdan MCV, Porter FD, Platt FM, Pavan WJ. NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology. **J Clin Med 2019**;9(1). PMCID: PMC7019814.
33. Power B, Ferreira CR, Chen D, Zein WM, O'Brien KJ, Introne WJ, Stephen J, Gahl WA, Huizing M, Malicdan MCV, Adams DR, Gochuico BR. Hermansky-Pudlak syndrome and oculocutaneous albinism in Chinese children with pigmentation defects and easy bruising. **Orphanet J Rare Dis** 2019;14(1):52. PMCID: PMC6385472.
34. Pode-Shakked B, Heimer G, Vilboux T, Marek-Yagel D, Ben-Zeev B, Davids M, Ferreira CR, Philosoph AM, Veber A, Pode-Shakked N, Kenet G, Soudack M, Hoffmann C, Vernitsky H, Safaniev M, Lodzki M, Lahad A, Shouval DS, Levinkopf D, Weiss B, Barg AA, Daka A, Amariglio N, Malicdan MCV, Gahl WA, Anikster Y. Cerebral and portal vein thrombosis, macrocephaly and atypical absence seizures in Glycosylphosphatidyl inositol deficiency due to a PIGM promoter mutation. **Mol Genet Metab** 2019;128(1-2):151-61.
35. Nicoli ER, Weston MR, Hackbarth M, Becerril A, Larson A, Zein WM, Baker PR, 2nd, Burke JD, Dorward H, Davids M, Huang Y, Adams DR, Zerfas PM, Chen D, Markello TC, Toro C, Wood T, Elliott G, Vu M, Undiagnosed Diseases N, Zheng W, Garrett LJ, Tifft CJ, Gahl WA, Day-Salvatore DL, Mindell JA, Malicdan MCV. Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. **Am J Hum Genet** 2019;104(6):1127-38. PMCID: PMC6562152.
36. Macnamara EF, Koehler AE, D'Souza P, Estwick T, Lee P, Vezina G, Undiagnosed Diseases N, Fauni H, Braddock SR, Torti E, Holt JM, Sharma P, Malicdan MCV, Tifft CJ. Kilquist syndrome: A novel syndromic hearing loss disorder caused by homozygous deletion of SLC12A2. **Hum Mutat** 2019;40(5):532-38. PMCID: PMC6693334.
37. Kuo ME, Theil AF, Kievit A, Malicdan MC, Introne WJ, Christian T, Verheijen FW, Smith DEC, Mendes MI, Hussaarts-Odijk L, van der Meijden E, van Slegtenhorst M, Wilke M, Vermeulen W, Raams A, Groden C, Shimada S, Meyer-Schuman R, Hou YM, Gahl WA, Antonellis A, Salomons GS, Mancini GMS. Cysteinyl-tRNA Synthetase Mutations Cause a Multi-System, Recessive Disease That Includes Microcephaly, Developmental Delay, and Brittle Hair and Nails. **Am J Hum Genet** 2019;104(3):520-29. PMCID: PMC6407526.
38. Kanca O, Andrews JC, Lee PT, Patel C, Braddock SR, Slavotinek AM, Cohen JS, Gubbels CS, Aldinger KA, Williams J, Indaram M, Fatemi A, Yu TW, Agrawal PB, Vezina G, Simons C, Crawford J, Lau CC, Undiagnosed Diseases N, Chung WK, Markello TC, Dobyns WB, Adams DR, Gahl WA, Wangler MF, Yamamoto S, Bellen HJ, Malicdan MCV. De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. Am J **Hum Genet** 2019;105(2):413-24. PMCID: PMC6699142.
39. Kanca O, Andrews JC, Lee PT, Patel C, Braddock SR, Slavotinek AM, Cohen JS, Gubbels CS, Aldinger KA, Williams J, Indaram M, Fatemi A, Yu TW, Agrawal PB, Vezina G, Simons C, Crawford J, Lau CC, Undiagnosed Diseases N, Chung WK, Markello TC, Dobyns WB, Adams DR, Gahl WA, Wangler MF, Yamamoto S, Bellen HJ, Malicdan MCV. De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. **Am J Hum Genet** 2019;105(3):672-74. PMCID: PMC6732524.
40. Huizing M, Yardeni T, Fuentes F, Malicdan MCV, Leoyklang P, Volkov A, Dekel B, Brede E, Blake J, Powell A, Chatrathi H, Anikster Y, Carrillo N, Gahl WA, Kopp JB. Rationale and Design for a Phase 1 Study of N-Acetylmannosamine for Primary Glomerular Diseases. **Kidney Int Rep** 2019;4(10):1454-62. PMCID: PMC6829193.
41. Gu F, Wu A, Gordon MG, Vlahos L, Macnamara S, Burke E, Malicdan MC, Adams DR, Tifft CJ, Toro C, Gahl WA, Markello TC. A suite of automated sequence analyses reduces the number of candidate deleterious variants and reveals a difference between probands and unaffected siblings. **Genet Med** 2019;21(8):1772-80. PMCID: PMC6669106.
42. Davids M, Markello T, Wolfe LA, Chepa-Lotrea X, Tifft CJ, Gahl WA, Malicdan MCV. Early infantile-onset epileptic encephalopathy 28 due to a homozygous microdeletion involving the WWOX gene in a region of uniparental disomy. **Hum Mutat** 2019;40(1):42-47. PMCID: PMC6296882.
43. Davids M, Kane MS, Wolfe LA, Toro C, Tifft CJ, Adams D, Li X, Raihan MA, He M, Gahl WA, Boerkoel CF, Malicdan MCV. Glycomics in rare diseases: from diagnosis tomechanism. **Transl Res** 2019;206:5-17.

44. Chin JJ, Behnam B, Davids M, Sharma P, Zein WM, Wang C, Chepa-Lotrea X, Gallantine WB, Toro C, Adams DR, Tifft CJ, Gahl WA, Malicdan MCV. Novel mutations in CLN6 cause late-infantile neuronal ceroid lipofuscinosis without visual impairment in two unrelated patients. **Mol Genet Metab** 2019;126(2):188-95.
45. Toro C, Hori RT, Malicdan MCV, Tifft CJ, Goldstein A, Gahl WA, Adams DR, Fauni HB, Wolfe LA, Xiao J, Khan MM, Tian J, Hope KA, Reiter LT, Tremblay MG, Moss T, Franks AL, Balak C, Group CRR, LeDoux MS. A recurrent de novo missense mutation in UBTF causes developmental neuroregression. **Hum Mol Genet**. 2018;27(4):691-705. PubMed PMID: Medline:29300972. English.
46. Toro C, Hori RT, Malicdan MCV, Tifft CJ, Goldstein A, Gahl WA, Adams DR, Fauni HB, Wolfe LA, Xiao J, Khan MM, Tian J, Hope KA, Reiter LT, Tremblay MG, Moss T, Franks AL, Balak C, Group CRR, LeDoux MS. A recurrent de novo missense mutation in UBTF causes developmental neuroregression. **Hum Mol Genet**. 2018;27(7):1310. PubMed PMID: Medline:29447355. English.
47. Strongin A, Heller T, Doherty D, Glass IA, Parisi MA, Bryant J, Choyke P, Turkbey B, Daryanani K, Yildirimli D, Vemulapalli M, Mullikin JC, Malicdan MC, Vilboux T, Gahl WA, Gunay-Aygun M, Program NCS. Characteristics of Liver Disease in 100 Individuals With Joubert Syndrome Prospectively Evaluated at a Single Center. **Journal of pediatric gastroenterology and nutrition**. 2018;66(3):428-35. PubMed PMID: Medline:29112083. English.
48. Stephen J, Nampoothiri S, Vinayan KP, Yesodharan D, Remesh P, Gahl WA, Malicdan MCV. Cortical atrophy and hypofibrinogenemia due to FGG and TBCD mutations in a single family: a case report. **BMC medical genetics**. 2018;19(1):80. PubMed PMID: Medline:29769041. English.
49. Stephen J, Nampoothiri S, Kuppa S, Yesodharan D, Radhakrishnan N, Gahl WA, Malicdan MCV. Novel truncating mutation in TENM3 in siblings with motor developmental delay, ocular coloboma, oval cornea, without microphthalmia. **Am J Med Genet A**. 2018. PubMed PMID: Medline:30513139. English.
50. Stephen J, Nampoothiri S, Banerjee A, Tolman NJ, Penninger JM, Elling U, Agu CA, Burke JD, Devadathan K, Kannan R, Huang Y, Steinbach PJ, Martinis SA, Gahl WA, Malicdan MCV. Loss of function mutations in VARS encoding cytoplasmic valyl-tRNA synthetase cause microcephaly, seizures, and progressive cerebral atrophy. **Human genetics**. 2018;137(4):293-303. PubMed PMID: Medline:29691655. English.
51. Stephen J, Maddirevula S, Nampoothiri S, Burke JD, Herzog M, Shukla A, Steindl K, Eskin A, Patil SJ, Joset P, Lee H, Garrett LJ, Yokoyama T, Balanda N, Bodine SP, Tolman NJ, Zerfas PM, Zheng A, Ramantani G, Girisha KM, Rivas C, Suresh PV, Elkahloun A, Alsaif HS, Wakil SM, Mahmoud L, Ali R, Prochazkova M, Undiagnosed Diseases Network m, Kulkarni AB, Ben-Omran T, Colak D, Morris HD, Rauch A, Martinez-Agosto JA, Nelson SF, Alkuraya FS, Gahl WA, Malicdan MCV. Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism**. Am J Hum Genet.** 2018;103(6):948-67. PubMed PMID: Medline:30526868. English.
52. Shanbhag NM, Geschwind MD, DiGiovanna JJ, Groden C, Godfrey R, Yousefzadeh MJ, Wade EA, Niedernhofer LJ, Malicdan MCV, Kraemer KH, Gahl WA, Toro C. Neurodegeneration as the presenting symptom in 2 adults with xeroderma pigmentosum complementation group F. **Neurology Genetics**. 2018;4(3):e240. PubMed PMID: Medline:29892709. English.
53. Morimoto M, Waller-Evans H, Ammous Z, Song X, Strauss KA, Pehlivan D, Gonzaga-Jauregui C, Puffenberger EG, Holst CR, Karaca E, Brigatti KW, Maguire E, Coban-Akdemir ZH, Amagata A, Lau CC, Chepa-Lotrea X, Macnamara E, Tos T, Isikay S, Nehrebecky M, Overton JD, Klein M, Markello TC, Posey JE, Adams DR, Lloyd-Evans E, Lupski JR, Gahl WA, Malicdan MCV. Bi-allelic CCDC47 Variants Cause a Disorder Characterized by Woolly Hair, Liver Dysfunction, Dysmorphic Features, and Global Developmental Delay. **Am J Hum Genet**. 2018;103(5):794-807. PubMed PMID: Medline:30401460. English.
54. May-Simera HL, Wan Q, Jha BS, Hartford J, Khristov V, Dejene R, Chang J, Patnaik S, Lu Q, Banerjee P, Silver J, Insinna-Kettenhofen C, Patel D, Lotfi M, Malicdan M, Hotaling N, Maminishkis A, Sridharan R, Brooks B, Miyagishima K, Gunay-Aygun M, Pal R, Westlake C, Miller S, Sharma R, Bharti K. Primary Cilium-Mediated Retinal Pigment Epithelium Maturation Is Disrupted in Ciliopathy Patient Cells. **Cell reports**. 2018;22(1):189-205. PubMed PMID: Medline:29298421. English.
55. Malicdan MCV, Vilboux T, Ben-Zeev B, Guo J, Eliyahu A, Pode-Shakked B, Dori A, Kakani S, Chandrasekharappa SC, Ferreira CR, Shelestovich N, Marek-Yagel D, Pri-Chen H, Blatt I, Niederhuber JE, He L, Toro C, Taylor RW, Deeken J, Yardeni T, Wallace DC, Gahl WA, Anikster Y. A novel inborn error of the

coenzyme Q10 biosynthesis pathway: cerebellar ataxia and static encephalomyopathy due to COQ5 C-methyltransferase deficiency. **Hum Mutat**. 2018;39(1):69-79. PubMed PMID: Medline:29044765. English.

56. Leoyklang P, Class B, Noguchi S, Gahl WA, Carrillo N, Nishino I, Huizing M, Malicdan MC. Quantification of lectin fluorescence in GNE myopathy muscle biopsies. **Muscle Nerve**. 2018;58(2):286-92. PubMed PMID: Medline:29603301. English.
57. Han CG, O'Brien KJ, Coon LM, Majerus JA, Huryn LA, Haroutunian SG, Moka N, Introne WJ, Macnamara E, Gahl WA, Malicdan MCV, Chen D, Krishnan K, Gochuico BR. Severe bleeding with subclinical oculocutaneous albinism in a patient with a novel HPS6 missense variant. **Am J Med Genet A**. 2018:e40514. PubMed PMID: Medline:30369044. English.
58. Ferreira CR, Xia Z-J, Clement A, Parry DA, Davids M, Taylan F, Sharma P, Turgeon CT, Blanco-Sanchez B, Ng BG, Logan CV, Wolfe LA, Solomon BD, Cho MT, Douglas G, Carvalho DR, Bratke H, Haug MG, Phillips JB, Wegner J, Tiemeyer M, Aoki K, Undiagnosed Diseases N, Scottish Genome P, Nordgren A, Hammarsjo A, Duker AL, Rohena L, Hove HB, Ek J, Adams D, Tifft CJ, Onyekweli T, Weixel T, Macnamara E, Radtke K, Powis Z, Earl D, Gabriel M, Russi AHS, Brick L, Kozenko M, Tham E, Raymond KM, Phillips JA, 3rd, Tiller GE, Wilson WG, Hamid R, Malicdan MCV, Nishimura G, Grigelioniene G, Jackson A, Westerfield M, Bober MB, Gahl WA, Freeze HH. A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. **Am J Hum Genet. 2018**;103(4):553-67. PubMed PMID: Medline:30290151. English.
59. Davids M, Kane MS, Wolfe LA, Toro C, Tifft CJ, Adams D, Li X, Raihan MA, He M, Gahl WA, Boerkoel CF, Malicdan MCV. Glycomics in rare diseases: from diagnosis to mechanism. **Translational research** : the journal of laboratory and clinical medicine. 2018. PubMed PMID: Medline:30423312. English.
60. Chin JJ, Behnam B, Davids M, Sharma P, Zein WM, Wang C, Chepa-Lotrea X, Gallantine WB, Toro C, Adams DR, Tifft CJ, Gahl WA, Malicdan MCV. Novel mutations in CLN6 cause late-infantile neuronal ceroid lipofuscinosis without visual impairment in two unrelated patients. **Mol Genet Metab**. 2018. PubMed PMID: Medline:30528883. English.
61. Carrillo N, Malicdan MC, Huizing M. GNE Myopathy: Etiology, Diagnosis, and Therapeutic Challenges. **Neurotherapeutic**s : the journal of the American Society for Experimental NeuroTherapeutics. 2018;15(4):900-14. PubMed PMID: Medline:30338442. English.
62. Burke EA, Frucht SJ, Thompson K, Wolfe LA, Yokoyama T, Bertoni M, Huang Y, Sincan M, Adams DR, Taylor RW, Gahl WA, Toro C, Malicdan MCV. Biallelic mutations in mitochondrial tryptophanyl-tRNA synthetase cause Levodopa-responsive infantile-onset Parkinsonism. Clin Genet. 2018;93(3):712-8. PubMed PMID: Medline:29120065. English.
63. Brooks BP, Zein WM, Thompson AH, Mokhtarzadeh M, Doherty DA, Parisi M, Glass IA, Malicdan MC, Vilboux T, Vemulapalli M, Mullikin JC, Gahl WA, Gunay-Aygun M. Joubert Syndrome: Ophthalmological Findings in Correlation with Genotype and Hepatorenal Disease in 99 Patients Prospectively Evaluated at a Single Center. **Ophthalmology**. 2018;125(12):1937-52. PubMed PMID: Medline:30055837. English.
64. Zhang Y, Huang H, Zhao G, Yokoyama T, Vega H, Huang Y, Sood R, Bishop K, Maduro V, Accardi J, Toro C, Boerkoel CF, Lyons K, Gahl WA, Duan X, Malicdan MCV, Lin S. ATP6V1H Deficiency Impairs Bone Development through Activation of MMP9 and MMP13. **PLoS genetics**. 2017;13(2):e1006481. PubMed PMID: Medline:28158191. English.
65. Xu X, Wang AQ, Latham LL, Celeste F, Ciccone C, Malicdan MC, Goldspiel B, Terse P, Cradock J, Yang N, Yorke S, McKew JC, Gahl WA, Huizing M, Carrillo N. Safety, pharmacokinetics and sialic acid production after oral administration of N-acetylmannosamine (ManNAc) to subjects with GNE myopathy. **Mol Genet Metab**. 2017;122(1-2):126-34. PubMed PMID: Medline:28641925. English.
66. Vilboux T, Malicdan MCV, Roney JC, Cullinane AR, Stephen J, Yildirimli D, Bryant J, Fischer R, Vemulapalli M, Mullikin JC, Program NCS, Steinbach PJ, Gahl WA, Gunay-Aygun M. CELSR2, encoding a planar cell polarity protein, is a putative gene in Joubert syndrome with cortical heterotopia, microophthalmia, and growth hormone deficiency. **Am J Med Genet A**. 2017;173(3):661-6. PubMed PMID: Medline:28052552. English.
67. Vilboux T, Doherty DA, Glass IA, Parisi MA, Phelps IG, Cullinane AR, Zein W, Brooks BP, Heller T, Soldatos A, Oden NL, Yildirimli D, Vemulapalli M, Mullikin JC, Nisc Comparative Sequencing P, Malicdan MCV, Gahl WA, Gunay-Aygun M. Molecular genetic findings and clinical correlations in 100 patients with Joubert syndrome and

related disorders prospectively evaluated at a single center. **Genetics in medicine** : official journal of the American College of Medical Genetics. 2017;19(8):875-82. PubMed PMID: Medline:28125082. English.

68. Svingen L, Goheen M, Godfrey R, Wahl C, Baker EH, Gahl WA, Malicdan MCV, Toro C. Late diagnosis and atypical brain imaging of Aicardi-Goutieres syndrome: are we failing to diagnose Aicardi-Goutieres syndrome-2? **Developmental medicine and child neurology**. 2017;59(12):1307-11. PubMed PMID: Medline:28762473. English.
69. Stephen J, Yokoyama T, Tolman NJ, O'Brien KJ, Nicoli E-R, Brooks BP, Huryn L, Titus SA, Adams DR, Chen D, Gahl WA, Gochuico BR, Malicdan MCV. Cellular and molecular defects in a patient with Hermansky-Pudlak syndrome type 5. **PLoS One**. 2017;12(3):e0173682. PubMed PMID: Medline:28296950. English.
70. Stephen J, Vilboux T, Mian L, Kuptanon C, Sinclair CM, Yildirimli D, Maynard DM, Bryant J, Fischer R, Vemulapalli M, Mullikin JC, Program NCS, Huizing M, Gahl WA, Malicdan MCV, Gunay-Aygun M. Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. **Human genetics**. 2017;136(4):399-408. PubMed PMID: Medline:28220259. English.
71. Shahrour MA, Staretz-Chacham O, Dayan D, Stephen J, Weech A, Damseh N, Pri Chen H, Edvardson S, Mazaheri S, Saada A, Sequencing NI, Hershkovitz E, Shaag A, Huizing M, Abu-Libdeh B, Gahl WA, Azem A, Anikster Y, Vilboux T, Elpeleg O, Malicdan MC. Mitochondrial epileptic encephalopathy, 3-methylglutaconic aciduria and variable complex V deficiency associated with TIMM50 mutations. **Clin Genet**. 2017;91(5):690-6. PubMed PMID: Medline:27573165. English.
72. Oprescu SN, Chepa-Lotrea X, Takase R, Golas G, Markello TC, Adams DR, Toro C, Gropman AL, Hou Y-M, Malicdan MCV, Gahl WA, Tifft CJ, Antonellis A. Compound heterozygosity for loss-of-function GARS variants results in a multisystem developmental syndrome that includes severe growth retardation. **Hum Mutat**. 2017;38(10):1412-20. PubMed PMID: Medline:28675565. English.
73. Noguchi S, Ogawa M, Malicdan MC, Nonaka I, Nishino I. 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. PubMed PMID: Medline:28043812. English.
74. Li C, Brazill JM, Liu S, Bello C, Zhu Y, Morimoto M, Cascio L, Pauly R, Diaz-Perez Z, Malicdan MCV, Wang H, Boccuto L, Schwartz CE, Gahl WA, Boerkoel CF, Zhai RG. Spermine synthase deficiency causes lysosomal dysfunction and oxidative stress in models of Snyder-Robinson syndrome. **Nature communications**. 2017;8(1):1257. PubMed PMID: Medline:29097652. English.
75. Kane MS, Davids M, Bond MR, Adams CJ, Grout ME, Phelps IG, O'Day DR, Dempsey JC, Li X, Golas G, Vezina G, Gunay-Aygun M, Hanover JA, Doherty D, He M, Malicdan MCV, Gahl WA, Boerkoel CF. Abnormal glycosylation in Joubert syndrome type 10. **Cilia**. 2017;6:2. PubMed PMID: Medline:28344780. English.
76. Kambouris M, Thevenon J, Soldatos A, Cox A, Stephen J, Ben-Omran T, Al-Sarraj Y, Boulos H, Bone W, Mullikin JC, Program NCS, Masurel-Paulet A, St-Onge J, Dufford Y, Chantegret C, Thauvin-Robinet C, Al-Alami J, Faivre L, Riviere JB, Gahl WA, Bassuk AG, Malicdan MCV, El-Shanti H. Biallelic SCN10A mutations in neuromuscular disease and epileptic encephalopathy. **Ann Clin Transl Neurol**. 2017;4(1):26-35. PubMed PMID: Medline:28078312. English.
77. Introne WJ, Westbroek W, Groden CA, Bhambhani V, Golas GA, Baker EH, Lehky TJ, Snow J, Ziegler SG, Malicdan MCV, Adams DR, Dorward HM, Hess RA, Huizing M, Gahl WA, Toro C. Neurologic involvement in patients with atypical Chediak-Higashi disease. **Neurology**. 2017;88(7):e57-e65. PubMed PMID: Medline:28193763. English.
78. Hardee I, Soldatos A, Davids M, Vilboux T, Toro C, David KL, Ferreira CR, Nehrebecky M, Snow J, Thurm A, Heller T, Macnamara EF, Gunay-Aygun M, Zein WM, Gahl WA, Malicdan MCV. Cover Image, Volume 173A, Number 12, December 2017. **Am J Med Genet A**. 2017;173(12):i. PubMed PMID: Medline:29136352. English.
79. Hardee I, Soldatos A, Davids M, Vilboux T, Toro C, David KL, Ferreira CR, Nehrebecky M, Snow J, Thurm A, Heller T, Macnamara EF, Gunay-Aygun M, Zein WM, Gahl WA, Malicdan MCV. Defective ciliogenesis in INPP5E-related Joubert syndrome. **Am J Med Genet** A. 2017;173(12):3231-7. PubMed PMID: Medline:29052317. English.
80. Garland J, Stephen J, Class B, Gruber A, Ciccone C, Poliak A, Hayes CP, Singhal V, Slota C, Perreault J, Gavrilova R, Shrader JA, Chittiboina P, Joe G, Heiss J, Gahl WA, Huizing M, Carrillo N, Malicdan MCV.

Identification of an Alu element-mediated deletion in the promoter region of GNE in siblings with GNE myopathy. **Molecular genetics & genomic medicine**. 2017;5(4):410-7. PubMed PMID: Medline:28717665. English.

81. Gall T, Valkanas E, Bello C, Markello T, Adams C, Bone WP, Brandt AJ, Brazill JM, Carmichael L, Davids M, Davis J, Diaz-Perez Z, Draper D, Elson J, Flynn ED, Godfrey R, Groden C, Hsieh C-K, Fischer R, Golas GA, Guzman J, Huang Y, Kane MS, Lee E, Li C, Links AE, Maduro V, Malicdan MCV, Malik FS, Nehrebecky M, Park J, Pemberton P, Schaffer K, Simeonov D, Sincan M, Smedley D, Valivullah Z, Wahl C, Washington N, Wolfe LA, Xu K, Zhu Y, Gahl WA, Tifft CJ, Toro C, Adams DR, He M, Robinson PN, Haendel MA, Zhai RG, Boerkoel CF. Defining Disease, Diagnosis, and Translational Medicine within a Homeostatic Perturbation Paradigm: The National Institutes of Health Undiagnosed Diseases Program Experience. **Frontiers in medicine**. 2017;4:62. PubMed PMID: Medline:28603714. English.
82. Fleming LR, Doherty DA, Parisi MA, Glass IA, Bryant J, Fischer R, Turkbey B, Choyke P, Daryanani K, Vemulapalli M, Mullikin JC, Malicdan MC, Vilboux T, Sayer JA, Gahl WA, Gunay-Aygun M. Prospective Evaluation of Kidney Disease in Joubert Syndrome. Clin **J Am Soc Nephrol**. 2017;12(12):1962-73. PubMed PMID: Medline:29146704. English.
83. Ferreira CR, Chen D, Abraham SM, Adams DR, Simon KL, Malicdan MC, Markello TC, Gunay-Aygun M, Gahl WA. Combined alpha-delta platelet storage pool deficiency is associated with mutations in GFI1B. **Mol Genet Metab**. 2017;120(3):288-94. PubMed PMID: Medline:28041820. English.
84. Cho A, Malicdan MCV, Miyakawa M, Nonaka I, Nishino I, Noguchi S. Sialic acid deficiency is associated with oxidative stress leading to muscle atrophy and weakness in GNE myopathy. **Hum Mol Genet**. 2017;26(16):3081-93. PubMed PMID: Medline:28505249. English.
85. Chao H-T, Davids M, Burke E, Pappas JG, Rosenfeld JA, McCarty AJ, Davis T, Wolfe L, Toro C, Tifft C, Xia F, Stong N, Johnson TK, Warr CG, Undiagnosed Diseases N, Yamamoto S, Adams DR, Markello TC, Gahl WA, Bellen HJ, Wangler MF, Malicdan MCV. A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. **Am J Hum Genet**. 2017;100(1):128-37. PubMed PMID: Medline:28017372. English.
86. Toro C, Nicoli ER, Malicdan MC, Adams DR, Introne WJ. Chediak-Higashi Syndrome. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, et al., editors. **GeneReviews**((R)). Seattle (WA)1993.
87. RA, Wallace SE, Bean LJH, Stephens K, et al., editors. **GeneReviews**((R)). Seattle (WA)1993.
88. Huizing M, Malicdan MCV, Gochuico BR, Gahl WA. Hermansky-Pudlak Syndrome. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, et al., editors. **GeneReviews**((R)). Seattle (WA)1993.
89. Bryan MM, Tolman NJ, Simon KL, Huizing M, Hufnagel RB, Brooks BP, Speransky V, Mullikin JC, Gahl WA, Malicdan MCV, Gochuico BR. Clinical and molecular phenotyping of a child with Hermansky-Pudlak syndrome-7, an uncommon genetic type of HPS. **Mol Genet Metab**. 2017;120(4):378-83. PubMed PMID: Medline:28259707. English.
90. Berger SI, Ciccone C, Simon KL, Malicdan MC, Vilboux T, Billington C, Fischer R, Introne WJ, Gropman A, Blancato JK, Mullikin JC, Program NCS, Gahl WA, Huizing M, Smith ACM. Exome analysis of Smith-Magenis-like syndrome cohort identifies de novo likely pathogenic variants. **Human genetics**. 2017;136(4):409-20. PubMed PMID: Medline:28213671. English.
91. Barel O, Malicdan MCV (Co-first author), Ben-Zeev B, Kandel J, Pri-Chen H, Stephen J, Castro IG, Metz J, Atawa O, Moshkovitz S, Ganelin E, Barshack I, Polak-Charcon S, Nass D, Marek-Yagel D, Amariglio N, Shalva N, Vilboux T, Ferreira C, Pode-Shakked B, Heimer G, Hoffmann C, Yardeni T, Nissenkorn A, Avivi C, Eyal E, Kol N, Glick Saar E, Wallace DC, Gahl WA, Rechavi G, Schrader M, Eckmann DM, Anikster Y. Deleterious variants in TRAK1 disrupt mitochondrial movement and cause fatal encephalopathy. **Brai**n. 2017;140(3):568-81. PubMed PMID: Medline:28364549. English.
92. Vilboux T, Malicdan MCV (Co-first author), Chang YM, Guo J, Zerfas PM, Stephen J, Cullinane AR, Bryant J, Fischer R, Brooks BP, Zein WM, Wiggs EA, Zalewski CK, Poretti A, Bryan MM, Vemulapalli M, Mullikin JC, Kirby M, Anderson SM, Program NCS, Huizing M, Toro C, Gahl WA, Gunay-Aygun M. Cystic cerebellar dysplasia and biallelic LAMA1 mutations: a lamininopathy associated with tics, obsessive compulsive traits and myopia due to cell adhesion and migration defects. **J Med Genet**. 2016;53(5):318-29. PubMed PMID: Medline:27095636. English.

93. Valkanas E, Schaffer K, Dunham C, Maduro V, du Souich C, Rupps R, Adams DR, Baradaran-Heravi A, Flynn E, Malicdan MC, Gahl WA, Toro C, Boerkoel CF. Phenotypic evolution of UNC80 loss of function. **Am J Med Genet A**. 2016;170(12):3106-14. PubMed PMID: Medline:27513830. English.
94. Stephen J, Vilboux T, Haberman Y, Pri-Chen H, Pode-Shakked B, Mazaheri S, Marek-Yagel D, Barel O, Di Segni A, Eyal E, Hout-Siloni G, Lahad A, Shalem T, Rechavi G, Malicdan MCV, Weiss B, Gahl WA, Anikster Y. Congenital protein losing enteropathy: an inborn error of lipid metabolism due to DGAT1 mutations. **Eur J Hum Genet**. 2016;24(9):1268-73. PubMed PMID: Medline:26883093. English.
95. O'Brien KJ, Lozier J, Cullinane AR, Osorio B, Nghiem K, Speransky V, Zein WM, Mullikin JC, Neff AT, Simon KL, Malicdan MCV, Gahl WA, Young LR, Gochuico BR. Identification of a novel mutation in HPS6 in a patient with hemophilia B and oculocutaneous albinism. **Mol Genet Metab**. 2016;119(3):284-7. PubMed PMID: Medline:27641950. English.
96. Mehrpour M, Gohari F, Dizaji MZ, Ahani A, Malicdan MCV, Behnam B. An ABCD1 Mutation (c.253dupC) Caused Diverse Phenotypes of Adrenoleukodystrophy in an Iranian Consanguineous Pedigree. **J Mol Genet Med**. 2016;10(2). PubMed PMID: Medline:27489563. English.
97. Maglic D, Stephen J, Malicdan MCV, Guo J, Fischer R, Konzman D, Program NCS, Mullikin JC, Gahl WA, Vilboux T, Gunay-Aygun M. TMEM231 Gene Conversion Associated with Joubert and Meckel-Gruber Syndromes in the Same Family. **Hum Mutat**. 2016;37(11):1144-8. PubMed PMID: Medline:27449316. English.
98. Maduro V, Pusey BN, Cherukuri PF, Atkins P, du Souich C, Rupps R, Limbos M, Adams DR, Bhatt SS, Eydoux P, Links AE, Lehman A, Malicdan MC, Mason CE, Morimoto M, Mullikin JC, Sear A, Van Karnebeek C, Stankiewicz P, Gahl WA, Toro C, Boerkoel CF. Complex translocation disrupting TCF4 and altering TCF4 isoform expression segregates as mild autosomal dominant intellectual disability. **Orphanet J Rare Dis**. 2016;11(1):62. PubMed PMID: Medline:27179618. English.
99. Paull D, Sevilla A, Zhou H, Hahn AK, Kim H, Napolitano C, Tsankov A, Shang L, Krumholz K, Jagadeesan P, Woodard CM, Sun B, Vilboux T, Zimmer M, Forero E, Moroziewicz DN, Martinez H, Malicdan MCV, Weiss KA, Vensand LB, Dusenberry CR, Polus H, Sy KTL, Kahler DJ, Gahl WA, Solomon SL, Chang S, Meissner A, Eggan K, Noggle SA. Automated, high-throughput derivation, characterization and differentiation of induced pluripotent stem cells. **Nat Methods**. 2015;12(9):885-92. PubMed PMID: Medline:26237226. English.
100. Marchegiani S, Davis T, Tessadori F, van Haaften G, Brancati F, Hoischen A, Huang H, Valkanas E, Pusey B, Schanze D, Venselaar H, Vulto-van Silfhout AT, Wolfe LA, Tifft CJ, Zerfas PM, Zambruno G, Kariminejad A, Sabbagh-Kermani F, Lee J, Tsokos MG, Lee C-CR, Ferraz V, da Silva EM, Stevens CA, Roche N, Bartsch O, Farndon P, Bermejo-Sanchez E, Brooks BP, Maduro V, Dallapiccola B, Ramos FJ, Chung H-YB, Le Caignec C, Martins F, Jacyk WK, Mazzanti L, Brunner HG, Bakkers J, Lin S, Malicdan MCV, Boerkoel CF, Gahl WA, de Vries BBA, van Haelst MM, Zenker M, Markello TC. Recurrent Mutations in the Basic Domain of TWIST2 Cause Ablepharon Macrostomia and Barber-Say Syndromes. **Am J Hum Genet**. 2015;97(1):99-110. PubMed PMID: Medline:26119818. English.
101. Malicdan MCV, Vilboux T, Stephen J, Maglic D, Mian L, Konzman D, Guo J, Yildirimli D, Bryant J, Fischer R, Zein WM, Snow J, Vemulapalli M, Mullikin JC, Toro C, Solomon BD, Niederhuber JE, Program NCS, Gahl WA, Gunay-Aygun M. Mutations in human homologue of chicken talpid3 gene (KIAA0586) cause a hybrid ciliopathy with overlapping features of Jeune and Joubert syndromes. **J Med Genet**. 2015;52(12):830-9. PubMed PMID: Medline:26386044. English.
102. Lam C, Golas GA, Davids M, Huizing M, Kane MS, Krasnewich DM, Malicdan MCV, Adams DR, Markello TC, Zein WM, Gropman AL, Lodish MB, Stratakis CA, Maric I, Rosenzweig SD, Baker EH, Ferreira CR, Danylchuk NR, Kahler S, Garnica AD, Bradley Schaefer G, Boerkoel CF, Gahl WA, Wolfe LA. Expanding the clinical and molecular characteristics of PIGT-CDG, a disorder of glycosylphosphatidylinositol anchors. **Mol Genet Metab**. 2015;115(2-3):128-40. PubMed PMID: Medline:25943031. English.
103. Yonekawa T, Malicdan MCV, Cho A, Hayashi YK, Nonaka I, Mine T, Yamamoto T, Nishino I, Noguchi S. Sialyllactose ameliorates myopathic phenotypes in symptomatic GNE myopathy model mice. **Brain**. 2014;137(Pt 10):2670-9. PubMed PMID: Medline:25062695. English.
104. Noguchi S, Ogawa M, Kawahara G, Malicdan MC, Nishino I. Allele-specific Gene Silencing of Mutant mRNA Restores Cellular Function in Ullrich Congenital Muscular Dystrophy Fibroblasts. **Mol Ther Nucleic Acids**. 2014;3:e171. PubMed PMID: Medline:24959844. English.

105. Leoyklang P, Malicdan MC, Yardeni T, Celeste F, Ciccone C, Li X, Jiang R, Gahl WA, Carrillo-Carrasco N, He M, Huizing M. Sialylation of Thomsen-Friedenreich antigen is a noninvasive blood-based biomarker for GNE myopathy. **Biomark Med**. 2014;8(5):641-52. PubMed PMID: Medline:25123033. English.
106. Huizing M, Carrillo-Carrasco N, Malicdan MCV, Noguchi S, Gahl WA, Mitrani-Rosenbaum S, Argov Z, Nishino I. GNE myopathy: new name and new mutation nomenclature. **Neuromuscul Disord**. 2014;24(5):387-9. PubMed PMID: Medline:24685570. English.
107. de Dios JKL, Shrader JA, Joe GO, McClean JC, Williams K, Evers R, Malicdan MCV, Ciccone C, Mankodi A, Huizing M, McKew JC, Bluemke DA, Gahl WA, Carrillo-Carrasco N. Atypical presentation of GNE myopathy with asymmetric hand weakness. **Neuromuscul Disord**. 2014;24(12):1063-7. PubMed PMID: Medline:25182749. English.
108. Celeste FV, Vilboux T, Ciccone C, de Dios JK, Malicdan MCV, Leoyklang P, McKew JC, Gahl WA, Carrillo-Carrasco N, Huizing M. Mutation update for GNE gene variants associated with GNE myopathy. **Hum Mutat**. 2014;35(8):915-26. PubMed PMID: Medline:24796702. English.
109. Vilboux T, Lev A, Malicdan MCV (Co-first author), Simon AJ, Jarvinen P, Racek T, Puchalka J, Sood R, Carrington B, Bishop K, Mullikin J, Huizing M, Garty BZ, Eyal E, Wolach B, Gavrieli R, Toren A, Soudack M, Atawneh OM, Babushkin T, Schiby G, Cullinane A, Avivi C, Polak-Charcon S, Barshack I, Amariglio N, Rechavi G, van der Werff ten Bosch J, Anikster Y, Klein C, Gahl WA, Somech R. A congenital neutrophil defect syndrome associated with mutations in VPS45. **N Engl J Med**. 2013;369(1):54-65. PubMed PMID: Medline:23738510. English.
110. Nesterova G, Malicdan MC, Yasuda K, Sakaki T, Vilboux T, Ciccone C, Horst R, Huang Y, Golas G, Introne W, Huizing M, Adams D, Boerkoel CF, Collins MT, Gahl WA. 1,25-(OH)2D-24 Hydroxylase (CYP24A1) Deficiency as a Cause of Nephrolithiasis. **Clin J Am Soc Nephrol**. 2013;8(4):649-57. PubMed PMID: Medline:23293122. English.
111. Mori-Yoshimura M, Okuma A, Oya Y, Fujimura-Kiyono C, Nakajima H, Matsuura K, Takemura A, Malicdan MCV, Hayashi YK, Nonaka I, Murata M, Nishino I. Clinicopathological features of centronuclear myopathy in Japanese populations harboring mutations in dynamin 2. **Clin Neurol Neurosurg**. 2012;114(6):678-83. PubMed PMID: Medline:22613877. English.
112. Mori-Yoshimura M, Monma K, Suzuki N, Aoki M, Kumamoto T, Tanaka K, Tomimitsu H, Nakano S, Sonoo M, Shimizu J, Sugie K, Nakamura H, Oya Y, Hayashi YK, Malicdan MCV, Noguchi S, Murata M, Nishino I. Heterozygous UDP-GlcNAc 2-epimerase and N-acetylmannosamine kinase domain mutations in the GNE gene result in a less severe GNE myopathy phenotype compared to homozygous N-acetylmannosamine kinase domain mutations. **J Neurol Sci**. 2012;318(1-2):100-5. PubMed PMID: Medline:22507750. English.
113. Momma K, Noguchi S, Malicdan MCV, Hayashi YK, Minami N, Kamakura K, Nonaka I, Nishino I. Rimmed vacuoles in Becker muscular dystrophy have similar features with inclusion myopathies. **PLoS One**. 2012;7(12):e52002. PubMed PMID: Medline:23251671. English.
114. Malicdan MCV, Noguchi S, Tokutomi T, Goto Y-i, Nonaka I, Hayashi YK, Nishino I. Peracetylated N-acetylmannosamine, a synthetic sugar molecule, efficiently rescues muscle phenotype and biochemical defects in mouse model of sialic acid-deficient myopathy. **J Biol Chem**. 2012;287(4):2689-705. PubMed PMID: Medline:22157763. English.
115. Malicdan MCV, Nishino I. Autophagy in lysosomal myopathies. **Brain Pathol**. 2012;22(1):82-8. PubMed PMID: Medline:22150923. English.
116. Malicdan MC, Noguchi S, Tokutomi T, Goto Y, Nonaka I, Hayashi YK, Nishino I. Malicdan MC, Nishino I. Autophagy in lysosomal myopathies. Brain Pathol. 2012 Jan;22(1):82-8. PubMed PMID: 22150923. Epub 2011/12/14.
117. Noguchi S, Malicdan MCV, Nishino I. [Animal model of distal myopathy with rimmed vacuoles/hereditary inclusion body myopathy and preclinical trial with sugar compounds]. **Brain Nerve**. 2010;62(6):601-7. PubMed PMID: Medline:20548120. Japanese.
118. Malicdan MCV, Noguchi S, Nishino I. A preclinical trial of sialic acid metabolites on distal myopathy with rimmed vacuoles/hereditary inclusion body myopathy, a sugar-deficient myopathy: a review. **Ther Adv Neurol Disord**. 2010;3(2):127-35. PubMed PMID: Medline:21179605. English.

119. Ohkuma A, Noguchi S, Sugie H, Malicdan MCV, Fukuda T, Shimazu K, Lopez LC, Hirano M, Hayashi YK, Nonaka I, Nishino I. Clinical and genetic analysis of lipid storage myopathies. **Muscle Nerve**. 2009;39(3):333-42. PubMed PMID: Medline:19208393. English.
120. Nishino I, Malicdan MCV, Noguchi S. [Development of therapy for distal myopathy with rimmed vacuoles]. **Rinsho Shinkeigaku**. 2009;49(11):852-5. PubMed PMID: Medline:20030229. Japanese.
121. Malicdan MCV, Noguchi S, Nishino I. Monitoring autophagy in muscle diseases. **Methods Enzymo**l. 2009;453:379-96. PubMed PMID: Medline:19216917. English.
122. Malicdan MCV, Noguchi S, Hayashi YK, Nonaka I, Nishino I. Prophylactic treatment with sialic acid metabolites precludes the development of the myopathic phenotype in the DMRV-hIBM mouse model. **Nat Med**. 2009;15(6):690-5. PubMed PMID: Medline:19448634. English.
123. Ohkuma A, Nonaka I, Malicdan MCV, Noguchi S, Ohji S, Nomura K, Sugie H, Hayashi YK, Nishino I. Distal lipid storage myopathy due to PNPLA2 mutation. **Neuromuscul Disord**. 2008;18(8):671-4. PubMed PMID: Medline:18657972. English.
124. Malicdan MCV, Nonaka I. Distal myopathies a review: highlights on distal myopathies with rimmed vacuoles. **Neurol India**. 2008;56(3):314-24. PubMed PMID: Medline:18974558. English.
125. Malicdan MCV, Noguchi S, Nishino I. Recent advances in distal myopathy with rimmed vacuoles (DMRV) or hIBM: treatment perspectives. **Curr Opin Neurol**. 2008;21(5):596-600. PubMed PMID: Medline:18769255. English.
126. Malicdan MCV, Noguchi S, Hayashi YK, Nishino I. Muscle weakness correlates with muscle atrophy and precedes the development of inclusion body or rimmed vacuoles in the mouse model of DMRV/hIBM. **Physiol Genomics**. 2008;35(1):106-15. PubMed PMID: Medline:18628337. English.
127. Malicdan MC, Noguchi S, Nonaka I, Saftig P, Nishino I. Lysosomal myopathies: an excessive build-up in autophagosomes is too much to handle. **Neuromuscul Disord**. 2008;18(7):521-9. PubMed PMID: Medline:18502640. English.
128. Kawahara G, Ogawa M, Okada M, Malicdan MCV, Goto Y-I, Hayashi YK, Noguchi S, Nishino I. Diminished binding of mutated collagen VI to the extracellular matrix surrounding myocytes. **Muscle Nerve**. 2008;38(3):1192-5. PubMed PMID: Medline:18642359. English.
129. Kawahara G, Ogawa M, Okada M, Malicdan MC, Goto Y, Hayashi YK, Noguchi S, Nishino I. Diminished binding of mutated collagen VI to the extracellular matrix surrounding myocytes. **Muscle Nerve**. 2008 Sep;38(3):1192-5. PubMed PMID: 18642359. Epub 2008/07/22.
130. Delaunay A, Bromberg KD, Hayashi Y, Mirabella M, Burch D, Kirkwood B, Serra C, Malicdan MC, Mizisin AP, Morosetti R, Broccolini A, Guo LT, Jones SN, Lira SA, Puri PL, Shelton GD, Ronai Ze. The ER-bound RING finger protein 5 (RNF5/RMA1) causes degenerative myopathy in transgenic mice and is deregulated in inclusion body myositis. **PLoS One**. 2008;3(2):e1609. PubMed PMID: Medline:18270596. English.
131. Malicdan MCV, Noguchi S, Nonaka I, Hayashi YK, Nishino I. A Gne knockout mouse expressing human GNE D176V mutation develops features similar to distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy. **Hum Mol Genet**. 2007;16(22):2669-82. PubMed PMID: Medline:17704511. English.
132. Malicdan MCV, Noguchi S, Nishino I. Autophagy in a mouse model of distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy. **Autophagy**. 2007;3(4):396-8. PubMed PMID: Medline:17471014. English.
133. Malicdan MCV, Noguchi S, Nishino I. Perspectives on distal myopathy with rimmed vacuoles or hereditary inclusion body myopathy: contributions from an animal model. Lack of sialic acid, a central determinant in sugar chains, causes myopathy? **Acta Myol**. 2007;26(3):171-5. PubMed PMID: Medline:18646567. English.
134. Wu S, Ibarra MCA, Malicdan MCV, Murayama K, Ichihara Y, Kikuchi H, Nonaka I, Noguchi S, Hayashi YK, Nishino I. Central core disease is due to RYR1 mutations in more than 90% of patients. **Brain**. 2006;129(Pt 6):1470-80. PubMed PMID: Medline:16621918. English.
135. Nishino I, Noguchi S, Murayama K, Ohkuma A, Kasahata N, Malicdan MCV, Hayashi YK, Nonaka I. [Molecular pathomechanism of distal myopathy with rimmed vacuoles]. **Rinsho Shinkeigaku**. 2005;45(11):943-5. PubMed PMID: Medline:16447769. Japanese.
136. Nishino I, Malicdan MCV, Murayama K, Nonaka I, Hayashi YK, Noguchi S. Molecular pathomechanism of distal myopathy with rimmed vacuoles. **Acta Myol. 2005**;24(2):80-3. PubMed PMID: Medline:16550921. English.

137. Malicdan MC, Rosales RL, Delgado MMS, Balingit E. The association of anthropometric measurements, body mass index, and diabetes mellitus in the occurrence of carpal tunnel syndrome. **Philipp J Neurol** 2005; 9:1-6.
138. Barja RA, Rosales R, Malicdan MC, Ramos R. Miyoshi type distal myopathy with dysferlin mutation in a Filipino patient. **Philipp J Neurol** 2005; 9:33-37.
139. Ty AU, Rosales RL, Delgado MMS, Malicdan MC. Detection of subclinical diabetic neuropathy: potential factors affecting electrodiagnostic parameters. **Philipp J Neurol** 2004; 8:41-45.
140. Corrales MLP, PeBenito R, Malicdan MC, Moral-Valencia MAL, Lat AB. Thymectomy in a 4-year old Filipino girl with ocular myasthenia gravis: a case report. **Philipp J Neurol** 2003; 7:39-42.
141. Malicdan MC; Moral-Valencia MA, Pica EC, Conde JL, Diaz AF. Isaac's syndrome in a 15- year old Filipino boy: a case report. **Philipp J Neurol** 2002; 6:79-87.

Consortium Publications

142. Borja N, Bivona S, Peart LS, Johnson B, Gonzalez J, Barbouth D, Moore H, Guo S; Undiagnosed Disease Network (…**Malicdan MCV**), Bademci G, Tekin M. Genome sequencing reveals novel noncoding variants in PLA2G6 and LMNB1 causing progressive neurologic disease. **Mol Genet Genomic Med**. 2022 Apr;10(4):e1892. doi: 10.1002/mgg3.1892. PMID: 35247231.
143. Brokamp E, Koziura ME, Phillips JA 3rd, Tang LA, Cogan JD, Rives LC, Robertson AK, Duncan L, Bican A, Peterson JF, Newman JH, Hamid R, Bastarache L; Undiagnosed Diseases Network (…**Malicdan MCV**). One is the loneliest number: genotypic matchmaking using the electronic health record. **Genet Med**. 2021 Oct;23(10):1830-1832. doi: 10.1038/s41436-021-01179-w. PMID: 34230636.
144. Shankar SP, Grimsrud K, Lanoue L, Egense A, Willis B, Hörberg J, AlAbdi L, Mayer K, Ütkür K, Monaghan KG, Krier J, Stoler J, Alnemer M, Shankar PR, Schaffrath R, Alkuraya FS, Brinkmann U, Eriksson LA, Lloyd K, Rauen KA; Undiagnosed Diseases Network (…**Malicdan MCV**). A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. Genet Med. 2022 Jul;24(7):1567-1582. doi: 10.1016/j.gim.2022.03.014. PMID: 35482014.
145. Baldridge D, Wangler MF, Bowman AN, Yamamoto S, Undiagnosed Diseases N, Schedl T, Pak SC, Postlethwait JH, Shin J, Solnica-Krezel L, Bellen HJ, Westerfield M, Undiagnosed Diseases Network (…**Malicdan MCV**),. Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. **Orphanet J Rare Dis** 2021;16(1):206.
146. Cope H, Barseghyan H, Bhattacharya S, Fu Y, Hoppman N, Marcou C, Walley N, Rehder C, Deak K, Alkelai A, Undiagnosed Diseases N, Vilain E, Shashi V, Acosta MT, Adam M, Adams DR, Agrawal PB, Alejandro ME, Alvey J, Amendola L, Andrews A, Ashley EA, Azamian MS, Bacino CA, Bademci G, Baker E, Balasubramanyam A, Baldridge D, Bale J, Bamshad M, Barbouth D, Bayrak-Toydemir P, Beck A, Beggs AH, Behrens E, Bejerano G, Bennet J, Berg-Rood B, Bernstein JA, Berry GT, Bican A, Bivona S, Blue E, Bohnsack J, Bonnenmann C, Bonner D, Botto L, Boyd B, Briere LC, Brokamp E, Brown G, Burke EA, Burrage LC, Butte MJ, Byers P, Byrd WE, Carey J, Carrasquillo O, Chang TCP, Chanprasert S, Chao H-T, Clark GD, Coakley TR, Cobban LA, Cogan JD, Coggins M, Sessions Cole F, Colley HA, Cooper CM, Craigen WJ, Crouse AB, Cunningham M, D'Souza P, Dai H, Dasari S, Davis J, Dayal JG, Deardorff M, Dell'Angelica EC, Dhar SU, Dipple K, Doherty D, Dorrani N, Doss AL, Douine ED, Draper DD, Duncan L, Earl D, Eckstein DJ, Emrick LT, Eng CM, Esteves C, Falk M, Fernandez L, Ferreira C, Fieg EL, Findley LC, Fisher PG, Fogel BL, Forghani I, Fresard L, Gahl WA, Glass I, Gochuico B, Godfrey RA, Golden-Grant K, Goldman AM, Goldrich MP, Goldstein DB, Grajewski A, Groden CA, Gutierrez I, Hahn S, Hamid R, Hanchard NA, Hassey K, Hayes N, High F, Hing A, Hisama FM, Holm IA, Hom J, Horike-Pyne M, Huang A, Huang Y, Huryn L, Isasi R, Jamal F, Jarvik GP, Jarvik J, Jayadev S, Karaviti L, Kennedy J, Kiley D, Kohane IS, Kohler JN, Krakow D, Krasnewich DM, Kravets E, Korrick S, Koziura M, Krier JB, Lalani SR, Lam B, Lam C, LaMoure GL, Lanpher BC, Lanza IR, Latham L, LeBlanc K, Lee BH, Lee H, Levitt R, Lewis RA, Lincoln SA, Liu P, Longo N, Loo SK, Loscalzo J, Maas RL, MacDowall J, Macnamara EF, MacRae CA, Maduro VV, Majcherska MM, Mak BC, **Malicdan MCV**, Mamounas LA, Manolio TA, Mao R, Maravilla K, Markello TC, Marom R, Marth G, Martin BA, Martin MG, Martinez-Agosto JA, Marwaha S, McCauley J, McConkie-Rosell A, McCormack CE, McCray AT, McGee E, Mefford H, Merritt JL, Might M, Mirzaa G, Morava E, Moretti PM, Mosbrook-Davis D, Mulvihill JJ, Murdock DR, Nagy A, Nakano-Okuno M, Nath A, Nelson SF, Newman JH, Nicholas SK, Nickerson D, Nieves-Rodriguez S, Novacic D, Oglesbee D, Orengo JP, Pace L, Pak S,

Pallais JC, Palmer CGS, Papp JC, Parker NH, Posey JE, Potocki L, Power B, Pusey BN, Quinlan A, Raskind W, Raja AN, Rao DA, Renteria G, Reuter CM, Rives L, Robertson AK, Rodan LH, Rosenfeld JA, Rosenwasser N, Rossignol F, Ruzhnikov M, Sacco R, Sampson JB, Samson SL, Saporta M, Scott CR, Schaechter J, Schedl T, Schoch K, Scott DA, Shin J, Signer R, Silverman EK, Sinsheimer JS, Sisco K, Smith EC, Smith KS, Solem E, Solnica-Krezel L, Solomon B, Spillmann RC, Stoler JM, Sullivan JA, Sullivan K, Sun A, Sutton S, Sweetser DA, Sybert V, Tabor HK, Tan ALM, Tan QKG, Tekin M, Telischi F, Thorson W, Thurm A, Tifft CJ, Toro C, Tran AA, Tucker BM, Urv TK, Vanderver A, Velinder M, Viskochil D, Vogel TP, Wahl CE, Wallace S, Walsh CA, Walker M, Wambach J, Wan J, Wang L-K, Wangler MF, Ward PA, Wegner D, Wener M, Wenger T, Perry KW, Westerfield M, Wheeler MT, Whitlock J, Wolfe LA, Woods JD, Yamamoto S, Yang J, Yousef M, Zastrow DB, Zein W, Zhao C, Zuchner S. Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. **Molecular genetics & genomic medicine** 2021:e1665.

147. Ferdinandusse S, McWalter K, Te Brinke H, Ijlst L, Mooijer PM, Ruiter JPN, van Lint AEM, Pras-Raves M, Wever E, Millan F, Guillen Sacoto MJ, Begtrup A, Tarnopolsky M, Brady L, Ladda RL, Sell SL, Nowak CB, Douglas J, Tian C, Ulm E, Perlman S, Drack AV, Chong K, Martin N, Brault J, Brokamp E, Toro C, Gahl WA, Macnamara EF, Wolfe L, Undiagnosed Diseases N, Waisfisz Q, Zwijnenburg PJG, Ziegler A, Barth M, Smith R, Ellingwood S, Gaebler-Spira D, Bakhtiari S, Kruer MC, van Kampen AHC, Wanders RJA, Waterham HR, Cassiman D, Vaz FM, Alejandro ME, Azamian MS, Bacino CA, Balasubramanyam A, Burrage LC, Chao H-T, Clark GD, Craigen WJ, Dai H, Dhar SU, Emrick LT, Goldman AM, Hanchard NA, Jamal F, Karaviti L, Lalani SR, Lee BH, Lewis RA, Marom R, Moretti PM, Murdock DR, Nicholas SK, Orengo JP, Posey JE, Potocki L, Rosenfeld JA, Samson SL, Scott DA, Tran AA, Vogel TP, Wangler MF, Yamamoto S, Eng CM, Liu P, Ward PA, Behrens E, Deardorff M, Falk M, Hassey K, Sullivan K, Vanderver A, Goldstein DB, Cope H, McConkie-Rosell A, Schoch K, Shashi V, Smith EC, Spillmann RC, Sullivan JA, Tan QKG, Walley NM, Agrawal PB, Beggs AH, Berry GT, Briere LC, Cobban LA, Coggins M, Cooper CM, Fieg EL, High F, Holm IA, Korrick S, Krier JB, Lincoln SA, Loscalzo J, Maas RL, MacRae CA, Pallais JC, Rao DA, Rodan LH, Silverman EK, Stoler JM, Sweetser DA, Walker M, Walsh CA, Esteves C, Kelley EG, Kohane IS, LeBlanc K, McCray AT, Nagy A, Dasari S, Lanpher BC, Lanza IR, Morava E, Oglesbee D, Bademci G, Barbouth D, Bivona S, Carrasquillo O, Chang TCP, Forghani I, Grajewski A, Isasi R, Lam B, Levitt R, Liu XZ, McCauley J, Sacco R, Saporta M, Schaechter J, Tekin M, Telischi F, Thorson W, Zuchner S, Colley HA, Dayal JG, Eckstein DJ, Findley LC, Krasnewich DM, Mamounas LA, Manolio TA, Mulvihill JJ, LaMoure GL, Goldrich MP, Urv TK, Doss AL, Acosta MT, Bonnenmann C, D'Souza P, Draper DD, Ferreira C, Godfrey RA, Groden CA, Macnamara EF, Maduro VV, Markello TC, Nath A, Novacic D, Pusey BN, Toro C, Wahl CE, Baker E, Burke EA, Adams DR, Gahl WA, Malicdan MCV, Tifft CJ, Wolfe LA, Yang J, Power B, Gochuico B, Huryn L, Latham L, Davis J, Mosbrook-Davis D, Rossignol F, Solomon B, MacDowall J, Thurm A, Zein W, Yousef M, Adam M, Amendola L, Bamshad M, Beck A, Bennett J, Berg-Rood B, Blue E, Boyd B, Byers P, Chanprasert S, Cunningham M, Dipple K, Doherty D, Earl D, Glass I, Golden-Grant K, Hahn S, Hing A, Hisama FM, Horike-Pyne M, Jarvik GP, Jarvik J, Jayadev S, Lam C, Maravilla K, Mefford H, Merritt JL, Mirzaa G, Nickerson D, Raskind W, Rosenwasser N, Scott CR, Sun A, Sybert V, Wallace S, Wener M, Wenger T, Ashley EA, Bejerano G, Bernstein JA, Bonner D, Coakley TR, Fernandez L, Fisher PG, Fresard L, Hom J, Huang Y, Kohler JN, Kravets E, Majcherska MM, Martin BA, Marwaha S, McCormack CE, Raja AN, Reuter CM, Ruzhnikov M, Sampson JB, Smith KS, Sutton S, Tabor HK, Tucker BM, Wheeler MT, Zastrow DB, Zhao C, Byrd WE, Crouse AB, Might M, Nakano-Okuno M, Whitlock J, Brown G, Butte MJ, Dell'Angelica EC, Dorrani N, Douine ED, Fogel BL, Gutierrez I, Huang A, Krakow D, Lee H, Loo SK, Mak BC, Martin MG, Martinez-Agosto JA, McGee E, Nelson SF, Nieves-Rodriguez S, Palmer CGS, Papp JC, Parker NH, Renteria G, Signer RH, Sinsheimer JS, Wan J, Wang L-K, Perry KW, Woods JD, Alvey J, Andrews A, Bale J, Bohnsack J, Botto L, Carey J, Pace L, Longo N, Marth G, Moretti P, Quinlan A, Velinder M, Viskochil D, Bayrak-Toydemir P, Mao R, Westerfield M, Bican A, Brokamp E, Duncan L, Hamid R, Kennedy J, Kozuira M, Newman JH, Phillips JA, 3rd, Rives L, Robertson AK, Solem E, Cogan JD, Cole FS, Hayes N, Kiley D, Sisco K, Wambach J, Wegner D, Baldridge D, Pak S, Schedl T, Shin J, Solnica-Krezel L. An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. **Genetics in medicine : official journal of the American College of Medical Genetics** 2021;23(4):740-50.

148. Kobren SN, Baldridge D, Velinder M, Krier JB, LeBlanc K, Esteves C, Pusey BN, Zuchner S, Blue E, Lee H, Huang A, Bastarache L, Bican A, Cogan J, Marwaha S, Alkelai A, Murdock DR, Liu P, Wegner DJ, Paul AJ,

Undiagnosed Diseases N, Sunyaev SR, Kohane IS, Acosta MT, Adam M, Adams DR, Agrawal PB, Alejandro ME, Alvey J, Amendola L, Andrews A, Ashley EA, Azamian MS, Bacino CA, Bademci G, Baker E, Balasubramanyam A, Baldridge D, Bale J, Bamshad M, Barbouth D, Bayrak-Toydemir P, Beck A, Beggs AH, Behrens E, Bejerano G, Bennett J, Berg-Rood B, Bernstein JA, Berry GT, Bican A, Bivona S, Blue E, Bohnsack J, Bonnenmann C, Bonner D, Botto L, Boyd B, Briere LC, Brokamp E, Brown G, Burke EA, Burrage LC, Butte MJ, Byers P, Byrd WE, Carey J, Carrasquillo O, Chang TCP, Chanprasert S, Chao H-T, Clark GD, Coakley TR, Cobban LA, Cogan JD, Coggins M, Cole FS, Colley HA, Cooper CM, Cope H, Craigen WJ, Crouse AB, Cunningham M, D'Souza P, Dai H, Dasari S, Davis J, Daya JG, Deardorff M, Dell'Angelica EC, Dhar SU, Dipple K, Doherty D, Dorrani N, Doss AL, Douine ED, Draper DD, Duncan L, Earl D, Eckstein DJ, Emrick LT, Eng CM, Esteves C, Falk M, Fernandez L, Ferreira C, Fieg EL, Findley LC, Fisher PG, Fogel BL, Forghani I, Fresard L, Gahl WA, Glass I, Gochuico B, Godfrey RA, Golden-Grant K, Goldman AM, Goldrich MP, Goldstein DB, Grajewski A, Groden CA, Gutierrez I, Hahn S, Hamid R, Hanchard NA, Hassey K, Hayes N, High F, Hing A, Hisama FM, Holm IA, Hom J, Horike-Pyne M, Huang A, Huang Y, Huryn L, Isasi R, Jamal F, Jarvik GP, Jarvik J, Jayadev S, Karaviti L, Kennedy J, Kiley D, Kohane IS, Kohler JN, Korrick S, Kozuira M, Krakow D, Krasnewich DM, Kravets E, Krier JB, LaMoure GL, Lalani SR, Lam B, Lam C, Lanpher BC, Lanza IR, Latham L, LeBlanc K, Lee BH, Lee H, Levitt R, Lewis RA, Lincoln SA, Liu P, Liu XZ, Longo N, Loo SK, Loscalzo J, Maas RL, MacDowall J, MacRae CA, Macnamara EF, Maduro VV, Majcherska MM, Mak BC, Malicdan MCV, Mamounas LA, Manolio TA, Mao R, Maravilla K, Markello TC, Marom R, Marth G, Martin BA, Martin MG, Martinez-Agosto JA, Marwaha S, McCauley J, McConkie-Rosell A, McCormack CE, McCray AT, McGee E, Mefford H, Merritt JL, Might M, Mirzaa G, Morava E, Moretti PM, Moretti P, Mosbrook-Davis D, Mulvihill JJ, Murdock DR, Nagy A, Nakano-Okuno M, Nath A, Nelson SF, Newman JH, Nicholas SK, Nickerson D, Nieves-Rodriguez S, Novacic D, Oglesbee D, Orengo JP, Pace L, Pak S, Pallais JC, Palmer CGS, Papp JC, Parker NH, Phillips JA, 3rd, Posey JE, Potocki L, Power B, Pusey BN, Quinlan A, Raja AN, Rao DA, Raskind W, Renteria G, Reuter CM, Rives L, Robertson AK, Rodan LH, Rosenfeld JA, Rosenwasser N, Rossignol F, Ruzhnikov M, Sacco R, Sampson JB, Samson SL, Saporta M, Schaechter J, Schedl T, Schoch K, Scott CR, Scott DA, Shashi V, Shin J, Signer RH, Silverman EK, Sinsheimer JS, Sisco K, Smith EC, Smith KS, Solem E, Solnica-Krezel L, Ben S, Spillmann RC, Stoler JM, Sullivan JA, Sullivan K, Sun A, Sutton S, Sweetser DA, Sybert V, Tabor HK, Tan ALM, Tan QKG, Tekin M, Telischi F, Thorson W, Thurm A, Tifft CJ, Toro C, Tran AA, Tucker BM, Urv TK, Vanderver A, Velinder M, Viskochil D, Vogel TP, Wahl CE, Walker M, Wallace S, Walley NM, Walsh CA, Wambach J, Wan J, Wang L-K, Wangler MF, Ward PA, Wegner D, Wener M, Wenger T, Perry KW, Westerfield M, Wheeler MT, Whitlock J, Wolfe LA, Woods JD, Yamamoto S, Yang J, Yousef M, Zastrow DB, Zein W, Zhao C, Zuchner S. Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. **Genetics in medicine : official journal of the American College of Medical Genetics** 2021.

149. Kyle JE, Stratton KG, Zink EM, Kim Y-M, Bloodsworth KJ, Monroe ME, Undiagnosed Diseases N, Waters KM, Webb-Robertson B-JM, Koeller DM, Metz TO, Bacino CA, Hanchard NA, Lewis RA, Rosenfeld JA, Scott DA, Tran AA, Ward PA, Burrage LC, Clark GD, Alejandro ME, Posey JE, Wangler MF, Lee BH, Craigen WJ, Bellen HJ, Nicholas SK, Bostwick BL, Samson SL, Goldman AM, Moretti PM, Eng CM, Muzny DM, Orengo JP, Vogel TP, Lalani SR, Murdock DR, Azamian MS, Orange JS, Emrick LT, Dhar SU, Balasubramanyam A, Potocki L, Yamamoto S, Yang Y, Chen S, Jamal F, Karaviti L, Marom R, Lincoln SA, Walsh CA, Beggs AH, Rodan LH, Stoler JM, Berry GT, Cobban LA, MacRae CA, Krier JB, Silverman EK, Fieg EL, Maas RL, Loscalzo J, Aday A, Korrick S, Goldstein DB, Stong N, Sullivan JA, Spillmann RC, Pena LDM, Tan QKG, Walley NM, Jiang Y-H, McConkie-Rosell A, Schoch K, Shashi V, Cope H, Holm IA, Kohane IS, McCray AT, Esteves C, LeBlanc K, Might M, Kelley E, Worthey EA, Dorset DC, Boone BE, Levy SE, Birch CL, Jones AL, Brown DM, Bick DP, Newberry JS, Lazar J, May T, Sweetser DA, Briere LC, Pallais JC, Cooper CM, High F, Walker M, Colley HA, Mamounas LA, Manolio TA, Burke EA, Godfrey RA, Groden CA, Gahl WA, Wolfe LA, Markello TC, Lau CC, Draper DD, Gould SE, Nehrebecky ME, Wahl CE, Batzli GF, Macnamara EF, Dayal JG, Eckstein DJ, Mulvihill JJ, Tifft CJ, Urv TK, Wise AL, Murphy JL, Gropman AL, Howerton EM, Krasnewich DM, Johnston JM, Pusey BN, Adams DR, Maduro VV, Malicdan MCV, Davids M, Estwick T, Novacic D, Sharma P, Toro C, Yu G, Behnam B, D'Souza P, Ferreira C, Morimoto M, Baker EH, Yang J, Gourdine J-PF, Brush M, Haendel M, Ashley EA, Bernstein JA, Sampson JB, Zastrow DB, Friedman ND, Merker JD, McCormack CE, Fisher PG, Davidson JM, Dries AM, Enns GM, Majcherska MM, Reuter CM, Waggott DM, Kohler JN, Coakley TR, Smith KS,

Wheeler MT, Bonner D, Fernandez L, Hom J, Huang Y, Marwaha S, Zhao C, Martinez-Agosto JA, Dell'Angelica EC, Papp JC, Douine ED, Nelson SF, Martin MG, Palmer CG, Parker NH, Butte MJ, Yoon AJ, Loo SK, Fogel BL, Dipple KM, Sinsheimer JS, Allard P, Barseghyan H, Dorrani N, Lee H, Vilain E, Eskin A, Renteria G, Signer R, Wan J, Zheng A, Westerfield M, Phillips JA, 3rd, Cogan JD, Newman JH, Robertson AK, Hamid R, Bican A, Brokamp E, Duncan L, Kozuira M, Rives L, Shakachite L. A resource of lipidomics and metabolomics data from individuals with undiagnosed diseases. **Sci Data** 2021;8(1):114.

150. Marbach F, Stoyanov G, Erger F, Stratakis CA, Settas N, London E, Rosenfeld JA, Torti E, Haldeman-Englert C, Sklirou E, Kessler E, Ceulemans S, Nelson SF, Martinez-Agosto JA, Palmer CGS, Signer RH, Undiagnosed Diseases N, (… Malicdan MCV). Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. **Genetics in medicine,** 2021.

151. Studwell CM, Kelley EG, Undiagnosed Diseases N, Sinsheimer JS, Palmer CGS, LeBlanc K, Acosta MT, Adam M, Adams DR, Agrawal PB, Alejandro ME, Alvey J, Amendola L, Andrews A, Ashley EA, Azamian MS, Bacino CA, Bademci G, Baker E, Balasubramanyam A, Baldridge D, Bale J, Bamshad M, Barbouth D, Bayrak-Toydemir P, Beck A, Beggs AH, Behrens E, Bejerano G, Bennet J, Berg-Rood B, Bernstein JA, Berry GT, Bican A, Bivona S, Blue E, Bohnsack J, Bonnenmann C, Bonner D, Botto L, Boyd B, Briere LC, Brokamp E, Brown G, Burke EA, Burrage LC, Butte MJ, Byers P, Byrd WE, Carey J, Carrasquillo O, Chang TCP, Chanprasert S, Chao H-T, Clark GD, Coakley TR, Cobban LA, Cogan JD, Coggins M, Cole FS, Colley HA, Cooper CM, Cope H, Craigen WJ, Crouse AB, Cunningham M, D'Souza P, Dai H, Dasari S, Davids M, Dayal JG, Deardorff M, Dell'Angelica EC, Dhar SU, Dipple K, Doherty D, Dorrani N, Douine ED, Draper DD, Duncan L, Earl D, Eckstein DJ, Emrick LT, Eng CM, Esteves C, Estwick T, Falk M, Fernandez L, Ferreira C, Fieg EL, Findley LC, Fisher PG, Fogel BL, Forghani I, Fresard L, Gahl WA, Glass I, Godfrey RA, Golden-Grant K, Goldman AM, Goldstein DB, Grajewski A, Groden CA, Gropman AL, Gutierrez I, Hahn S, Hamid R, Hanchard NA, Hassey K, Hayes N, High F, Hing A, Hisama FM, Holm IA, Hom J, Horike-Pyne M, Huang A, Huang Y, Isasi R, Jamal F, Jarvik GP, Jarvik J, Jayadev S, Johnston JM, Karaviti L, Kelley EG, Kennedy J, Kiley D, Kohane IS, Kohler JN, Krakow D, Krasnewich DM, Kravets E, Korrick S, Koziura M, Krier JB, Lalani SR, Lam B, Lam C, Lanpher BC, Lanza IR, Lau CC, LeBlanc K, Lee BH, Lee H, Levitt R, Lewis RA, Lincoln SA, Liu P, Liu XZ, Longo N, Loo SK, Loscalzo J, Maas RL, Macnamara EF, MacRae CA, Maduro VV, Majcherska MM, Malicdan MCV, Mamounas LA, Manolio TA, Mao R, Maravilla K, Markello TC, Marom R, Marth G, Martin BA, Martin MG, Martinez-Agosto JA, Marwaha S, McCauley J, McConkie-Rosell A, McCormack CE, McCray AT, McGee E, Mefford H, Merritt JL, Might M, Mirzaa G, Morava E, Moretti PM, Morimoto M, Mulvihill JJ, Murdock DR, Nakano-Okuno M, Nath A, Nelson SF, Newman JH, Nicholas SK, Nickerson D, Novacic D, Oglesbee D, Orengo JP, Pace L, Pak S, Pallais JC, Palmer CGS, Papp JC, Parker NH, Phillips JA, 3rd, Posey JE, Potocki L, Pusey BN, Quinlan A, Raskind W, Raja AN, Rao DA, Renteria G, Reuter CM, Rives L, Robertson AK, Rodan LH, Rosenfeld JA, Rosenwasser N, Ruzhnikov M, Sacco R, Sampson JB, Samson SL, Saporta M, Scott CR, Schaechter J, Schedl T, Schoch K, Scott DA, Sharma P, Shashi V, Shin J, Signer R, Sillari CH, Silverman EK, Sinsheimer JS, Sisco K, Smith EC, Smith KS, Solem E, Solnica-Krezel L, Spillmann RC, Stoler JM, Stong N, Sullivan JA, Sullivan K, Sun A, Sutton S, Sweetser DA, Sybert V, Tabor HK, Tamburro CP, Tan QKG, Tekin M, Telischi F, Thorson W, Tifft CJ, Toro C, Tran AA, Tucker BM, Urv TK, Vanderver A, Velinder M, Viskochil D, Vogel TP, Wahl CE, Wallace S, Walley NM, Walsh CA, Wener M, Wenger T, Perry KW, Westerfield M, Wheeler MT, Whitlock J, Wolfe LA, Woods JD, Yamamoto S, Yang J, Yu G, Zastrow DB, Zhao C, Zuchner S. Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. **J Genet Couns** 2021;30(2):439-47.

152. Sturrock BRH, Macnamara EF, McGuire P, Kruk S, Yang I, Murphy J, Undiagnosed Diseases N, Tifft CJ, Gordon-Lipkin E, Undiagnosed Diseases Network (… Malicdan MCV). Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in SDHA: A Counseling Conundrum. **Molecular genetics & genomic medicine** 2021:e1692.

153. Schneeberger PE, Kortüm F, Korenke GC, Alawi M, Santer R, Woidy M, Buhas D, Fox S, Juusola J, Alfadhel M, Webb BD, Coci EG, Abou Jamra R, Siekmeyer M, Biskup S, Heller C, Maier EM, Javaher-Haghighi P, Bedeschi MF, Ajmone PF, Iascone M, Peeters H, Ballon K, Jaeken J, Rodríguez Alonso A, Palomares-Bralo M, Santos-Simarro F, Meuwissen MEC, Beysen D, Kooy RF, Houlden H, Murphy D, Doosti M, Karimiani EG, Mojarrad M, Maroofian R, Noskova L, Kmoch S, Honzik T, Cope H, Sanchez-Valle A; Undiagnosed Diseases Network (…Malicdan MC), Gelb BD, Kurth I, Hempel M, Kutsche K. Biallelic MADD variants cause a phenotypic

spectrum ranging from developmental delay to a multisystem disorder. Brain. 2020 Aug 6. pii: awaa204. doi: 10.1093/brain/awaa204. [Epub ahead of print] PubMed [citation]. PMID: 32761064

154. Guillen Sacoto MJ, Tchasovnikarova IA, Torti E, Forster C, Andrew EH, Anselm I, Baranano KW, Briere LC, Cohen JS, Craigen WJ, Cytrynbaum C, Ekhilevitch N, Elrick MJ, Fatemi A, Fraser JL, Gallagher RC, Guerin A, Haynes D, High FA, Inglese CN, Kiss C, Koenig MK, Krier J, Lindstrom K, Marble M, Meddaugh H, Moran ES, Morel CF, Mu W, Muller EA 2nd, Nance J, Natowicz MR, Numis AL, Ostrem B, Pappas J, Stafstrom CE, Streff H, Sweetser DA, Szybowska M; Undiagnosed Diseases Network (…Malicdan MC), Walker MA, Wang W, Weiss K, Weksberg R, Wheeler PG, Yoon G, Kingston RE, Juusola J. De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. Am J Hum Genet. 2020 Aug 6;107(2):352-363. doi: 10.1016/j.ajhg.2020.06.013. Epub 2020 Jul 20. PubMed [citation] PMID: 32693025, PMCID: PMC7413887

155. Ng BG, Eklund EA, Shiryaev SA, Dong YY, Abbott MA, Asteggiano C, Bamshad MJ, Barr E, Bernstein JA, Chelakkadan S, Christodoulou J, Chung WK, Ciliberto MA, Cousin J, Gardiner F, Ghosh S, Graf WD, Grunewald S, Hammond K, Hauser NS, Hoganson GE, Houck KM, Kohler JN, Morava E, Larson AA, Liu P, Madathil S, McCormack C, Meeks NJL, Miller R, Monaghan KG, Nickerson DA, Palculict TB, Papazoglu GM, Pletcher BA, Scheffer IE, Schenone AB, Schnur RE, Si Y, Rowe LJ, Serrano Russi AH, Russo RS, Thabet F, Tuite A, Villanueva MM, Wang RY, Webster RI, Wilson D, Zalan A; Undiagnosed Diseases Network (…Malicdan MC), University of Washington Center for Mendelian Genomics (UW-CMG), Wolfe LA, Rosenfeld JA, Rhodes L, Freeze HH. Predominant and novel de novo variants in 29 individuals with ALG13 deficiency: Clinical description, biomarker status, biochemical analysis, and treatment suggestions. J Inherit Metab Dis. 2020 Jul 18. doi: 10.1002/jimd.12290. [Epub ahead of print] PubMed [citation] PMID: 32681751

156. Macnamara EF, D'Souza P; Undiagnosed Diseases Network (…Malicdan MC), Tifft CJ. The undiagnosed diseases program: Approach to diagnosis. Transl Sci Rare Dis. 2020 Apr 13;4(3-4):179-188. doi: 10.3233/TRD-190045. PubMed [citation] PMID: 32477883, PMCID: PMC7250153

157. Castilla-Vallmanya L, Selmer KK, Dimartino C, Rabionet R, Blanco-Sánchez B, Yang S, Reijnders MRF, van Essen AJ, Oufadem M, Vigeland MD, Stadheim B, Houge G, Cox H, Kingston H, Clayton-Smith J, Innis JW, Iascone M, Cereda A, Gabbiadini S, Chung WK, Sanders V, Charrow J, Bryant E, Millichap J, Vitobello A, Thauvin C, Mau-Them FT, Faivre L, Lesca G, Labalme A, Rougeot C, Chatron N, Sanlaville D, Christensen KM, Kirby A, Lewandowski R, Gannaway R, Aly M, Lehman A, Clarke L, Graul-Neumann L, Zweier C, Lessel D, Lozic B, Aukrust I, Peretz R, Stratton R, Smol T, Dieux-Coëslier A, Meira J, Wohler E, Sobreira N, Beaver EM, Heeley J, Briere LC, High FA, Sweetser DA, Walker MA, Keegan CE, Jayakar P, Shinawi M, Kerstjens-Frederikse WS, Earl DL, Siu VM, Reesor E, Yao T, Hegele RA, Vaske OM, Rego S; Undiagnosed Diseases Network (…Malicdan MC), Care4Rare Canada Consortium, Shapiro KA, Wong B, Gambello MJ, McDonald M, Karlowicz D, Colombo R, Serretti A, Pais L, O'Donnell-Luria A, Wray A, Sadedin S, Chong B, Tan TY, Christodoulou J, White SM, Slavotinek A, Barbouth D, Morel Swols D, Parisot M, Bole-Feysot C, Nitschké P, Pingault V, Munnich A, Cho MT, Cormier-Daire V, Balcells S, Lyonnet S, Grinberg D, Amiel J, Urreizti R, Gordon CT. Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genet Med. 2020 Jul;22(7):1215-1226. doi: 10.1038/s41436-020-0792-7. Epub 2020 May 7. PubMed [citation] PMID: 32376980

158. Schoch K, Tan QK, Stong N, Deak KL, McConkie-Rosell A, McDonald MT; Undiagnosed Diseases Network(…Malicdan MC), Goldstein DB, Jiang YH, Shashi V. Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses. Genet Med. 2020 Jul;22(7):1269-1275. doi: 10.1038/s41436-020-0781-x. Epub 2020 May 5. PubMed [citation] PMID: 32366967, PMCID: PMC7335342

159. Chung HL, Mao X, Wang H, Park YJ, Marcogliese PC, Rosenfeld JA, Burrage LC, Liu P, Murdock DR, Yamamoto S, Wangler MF; Undiagnosed Diseases Network (…Malicdan MC)., Chao HT, Long H, Feng L, Bacino CA, Bellen HJ, Xiao B. De Novo Variants in CDK19 Are Associated with a Syndrome Involving

Intellectual Disability and Epileptic Encephalopathy. Am J Hum Genet. 2020 May 7;106(5):717-725. doi:10.1016/j.ajhg.2020.04.001. Epub 2020 Apr 23. PubMed [citation] PMID: 32330417, PMCID: PMC7212481

160. Béziat V, Tavernier SJ, Chen YH, Ma CS, Materna M, Laurence A, Staal J, Aschenbrenner D, Roels L, Worley L, Claes K, Gartner L, Kohn LA, De Bruyne M, Schmitz-Abe K, Charbonnier LM, Keles S, Nammour J, Vladikine N, Maglorius Renkilaraj MRL, Seeleuthner Y, Migaud M, Rosain J, Jeljeli M, Boisson B, Van Braeckel E, Rosenfeld JA, Dai H, Burrage LC, Murdock DR, Lambrecht BN, Avettand-Fenoel V, Vogel TP; Undiagnosed Diseases Network (…Malicdan MC), Esther CR, Haskologlu S, Dogu F, Ciznar P, Boutboul D, Ouachée-Chardin M, Amourette J, Lebras MN, Gauvain C, Tcherakian C, Ikinciogullari A, Beyaert R, Abel L, Milner JD, Grimbacher B, Couderc LJ, Butte MJ, Freeman AF, Catherinot É, Fieschi C, Chatila TA, Tangye SG, Uhlig HH, Haerynck F, Casanova JL, Puel A. Dominant-negative mutations in human IL6ST underlie hyper-IgE syndrome. J Exp Med. 2020 Jun 1;217(6). pii: e20191804. doi: 10.1084/jem.20191804. PubMed [citation] PMID: 32207811

161. Mao D, Reuter CM, Ruzhnikov MRZ, Beck AE, Farrow EG, Emrick LT, Rosenfeld JA, Mackenzie KM, Robak L, Wheeler MT, Burrage LC, Jain M, Liu P, Calame D, Küry S, Sillesen M, Schmitz-Abe K, Tonduti D, Spaccini L, Iascone M, Genetti CA, Koenig MK, Graf M, Tran A, Alejandro M; Undiagnosed Diseases Network (…Malicdan MC), Lee BH, Thiffault I, Agrawal PB, Bernstein JA, Bellen HJ, Chao HT. De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. Am J Hum Genet. 2020 Apr 2;106(4):570-583. doi: 10.1016/j.ajhg.2020.02.016. Epub 2020 Mar 19. PubMed [citation] PMID: 32197074, PMCID: PMC7118694

162. Burdick KJ, Cogan JD, Rives LC, Robertson AK, Koziura ME, Brokamp E, Duncan L, Hannig V, Pfotenhauer J, Vanzo R, Paul MS, Bican A, Morgan T, Duis J, Newman JH, Hamid R, Phillips JA 3rd; Undiagnosed Diseases Network (…Malicdan MC). Limitations of exome sequencing in detecting rare and undiagnosed diseases. Am J Med Genet A. 2020 Jun;182(6):1400-1406. doi: 10.1002/ajmg.a.61558. Epub 2020 Mar 19. PubMed [citation] PMID: 32190976

163. Chung HL, Wangler MF, Marcogliese PC, Jo J, Ravenscroft TA, Zuo Z, Duraine L, Sadeghzadeh S, Li-Kroeger D, Schmidt RE, Pestronk A, Rosenfeld JA, Burrage L, Herndon MJ, Chen S; Members of Undiagnosed Diseases Network (…Malicdan MC), Shillington A, Vawter-Lee M, Hopkin R, Rodriguez-Smith J, Henrickson M, Lee B, Lee B, Moser AB, Jones RO, Watkins P, Yoo T, Mar S, Choi M, Bucelli RC, Yamamoto S, Lee HK, Prada CE, Chae JH, Vogel TP, Bellen HJ. Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron. 2020 May 20;106(4):589-606.e6. doi: 10.1016/j.neuron.2020.02.021. Epub 2020 Mar 12. PubMed [citation] PMID: 32169171, PMCID: PMC7289150

164. Woods JD, Khanlou N, Lee H, Signer R, Shieh P, Chen J, Herzog M, Palmer C, Martinez-Agosto J; Undiagnosed Diseases Network (…Malicdan MC), Nelson SF. Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing. Neuropathology. 2020 Jun;40(3):302-307. doi: 10.1111/neup.12641. Epub 2020 Feb 9. PubMed [citation] PMID: 32037607, PMCID: PMC7317439

165. Shieh C, Jones N, Vanle B, Au M, Huang AY, Silva APG, Lee H, Douine ED, Otero MG, Choi A, Grand K, Taff IP, Delgado MR, Hajianpour MJ, Seeley A, Rohena L, Vernon H, Gripp KW, Vergano SA, Mahida S, Naidu S, Sousa AB, Wain KE, Challman TD, Beek G, Basel D, Ranells J, Smith R, Yusupov R, Freckmann ML, Ohden L, Davis-Keppen L, Chitayat D, Dowling JJ, Finkel R, Dauber A, Spillmann R, Pena LDM; Undiagnosed Diseases Network (…Malicdan MC), Metcalfe K, Splitt M, Lachlan K, McKee SA, Hurst J, Fitzpatrick DR, Morton JEV, Cox H, Venkateswaran S, Young JI, Marsh ED, Nelson SF, Martinez JA, Graham JM Jr, Kini U, Mackay JP, Pierson TM. GATAD2B-associated neurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-related disorder. Genet Med. 2020 May;22(5):878-888. doi: 10.1038/s41436-019-0747-z. Epub 2020 Jan 17. Erratum in: Genet Med. 2020 Feb 12;:. PubMed [citation] PMID: 31949314

166. Mirzaa GM, Chong JX, Piton A, Popp B, Foss K, Guo H, Harripaul R, Xia K, Scheck J, Aldinger KA, Sajan SA, Tang S, Bonneau D, Beck A, White J, Mahida S, Harris J, Smith-Hicks C, Hoyer J, Zweier C, Reis A, Thiel CT, amra RA, Zeid N, Yang A, Farach LS, Walsh L, Payne K, Rohena L, Velinov M, Ziegler A, Schaefer E, Gatinois

V, Geneviève D, Simon MEH, Kohler J, Rotenberg J, Wheeler P, Larson A, Ernst ME, Akman CI, Westman R, Blanchet P, Schillaci LA, Vincent-Delorme C, Gripp KW, Mattioli F, Guyader GL, Gerard B, Mathieu-Dramard M, Morin G, Sasanfar R, Ayub M, Vasli N, Yang S, Person R, Monaghan KG, Nickerson DA, van Binsbergen E, Enns GM, Dries AM, Rowe LJ, Tsai ACH, Svihovec S, Friedman J, Agha Z, Qamar R, Rodan LH, Martinez-Agosto J, Ockeloen CW, Vincent M, Sunderland WJ, Bernstein JA; Undiagnosed Diseases Network (…Malicdan MC), Eichler EE, Vincent JB; University of Washington Center for Mendelian Genomics (UW-CMG), Bamshad MJ. De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genet Med. 2020 Mar;22(3):538-546. doi: 10.1038/s41436-019-0693-9. Epub 2019 Nov 14. PubMed [citation] PMID: 31723249, PMCID: PMC7060121

167. Holt JM, Wilk B, Birch CL, Brown DM, Gajapathy M, Moss AC, Sosonkina N, Wilk MA, Anderson JA, Harris JM, Kelly JM, Shaterferdosian F, Uno-Antonison AE, Weborg A; Undiagnosed Diseases Network (…Malicdan MC), Worthey EA. VarSight: prioritizing clinically reported variants with binary classification algorithms. BMC Bioinformatics. 2019 Oct 15;20(1):496. doi: 10.1186/s12859-019-3026-8. PubMed [citation] PMID: 31615419, PMCID: PMC6792253

168. Lee H, Huang AY, Wang LK, Yoon AJ, Renteria G, Eskin A, Signer RH, Dorrani N, Nieves-Rodriguez S, Wan J, Douine ED, Woods JD, Dell'Angelica EC, Fogel BL, Martin MG, Butte MJ, Parker NH, Wang RT, Shieh PB, Wong DA, Gallant N, Singh KE, Tavyev Asher YJ, Sinsheimer JS, Krakow D, Loo SK, Allard P, Papp JC; Undiagnosed Diseases Network (…Malicdan MC), Palmer CGS, Martinez-Agosto JA, Nelson SF. Diagnostic utility of transcriptome sequencing for rare Mendelian diseases. Genet Med. 2020 Mar;22(3):490-499. doi: 10.1038/s41436-019-0672-1. Epub 2019 Oct 14. PubMed [citation] PMID: 31607746, PMCID: PMC7405636

169. Accogli A, Calabretta S, St-Onge J, Boudrahem-Addour N, Dionne-Laporte A, Joset P, Azzarello-Burri S, Rauch A, Krier J, Fieg E, Pallais JC; Undiagnosed Diseases Network., McConkie-Rosell A, McDonald M, Freedman SF, Rivière JB, Lafond-Lapalme J, Simpson BN, Hopkin RJ, Trimouille A, Van-Gils J, Begtrup A, Tavyev Asher YJ, Sinsheimer JS, Krakow D, Loo SK, Allard P, Papp JC; Undiagnosed Diseases Network (…Malicdan MC), Palmer CGS, Martinez-Agosto JA, Nelson SF. De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Collosum, Axon, Cardiac, Ocular, and Genital Defects. Am J Hum Genet. 2019 Oct 3;105(4):854-868. doi: 10.1016/j.ajhg.2019.09.005. PubMed [citation] PMID: 31585109, PMCID: PMC6817525

170. Reuter CM, Kohler JN, Bonner D, Zastrow D, Fernandez L, Dries A, Marwaha S, Davidson J, Brokamp E, Herzog M, Hong J, Macnamara E, Rosenfeld JA, Schoch K, Spillmann R; Undiagnosed Diseases Network (…Malicdan MC), Loscalzo J, Krier J, Stoler J, Sweetser D, Palmer CGS, Phillips JA, Shashi V, Adams DA, Yang Y, Ashley EA, Fisher PG, Mulvihill JJ, Bernstein JA, Wheeler MT. Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. J Genet Couns. 2019 Dec;28(6):1107-1118. doi: 10.1002/jgc4.1161. Epub 2019 Sep 3. PubMed [citation] PMID: 31478310, PMCID: PMC6901723

171. McConkie-Rosell A, Schoch K, Sullivan J, Cope H, Spillmann R, Palmer CGS, Pena L, Jiang YH, Daniels N, Walley N, Tan KG; Undiagnosed Diseases Network (…Malicdan MC), Hooper SR, Shashi V. The genome empowerment scale: An assessment of parental empowerment in families with undiagnosed disease. Clin Genet. 2019 Dec;96(6):521-531. doi: 10.1111/cge.13635. Epub 2019 Oct 8. PubMed [citation] PMID: 31448412, PMCID: PMC6983919

172. Johnson BV, Kumar R, Oishi S, Alexander S, Kasherman M, Vega MS, Ivancevic A, Gardner A, Domingo D, Corbett M, Parnell E, Yoon S, Oh T, Lines M, Lefroy H, Kini U, Van Allen M, Grønborg S, Mercier S, Küry S, Bézieau S, Pasquier L, Raynaud M, Afenjar A, Billette de Villemeur T, Keren B, Désir J, Van Maldergem L, Marangoni M, Dikow N, Koolen DA, VanHasselt PM, Weiss M, Zwijnenburg P, Sa J, Reis CF, López-Otín C, Santiago-Fernández O, Fernández-Jaén A, Rauch A, Steindl K, Joset P, Goldstein A, Madan-Khetarpal S, Infante E, Zackai E, Mcdougall C, Narayanan V, Ramsey K, Mercimek-Andrews S, Pena L, Shashi V; Undiagnosed Diseases Network (…Malicdan MC), Schoch K, Sullivan JA, Pinto E Vairo F, Pichurin PN, Ewing SA, Barnett SS, Klee EW, Perry MS, Koenig MK, Keegan CE, Schuette JL, Asher S, Perilla-Young Y, Smith LD, Rosenfeld JA, Bhoj E, Kaplan P, Li D, Oegema R, van Binsbergen E, van der Zwaag B, Smeland MF, Cutcutache I, Page M,

Armstrong M, Lin AE, Steeves MA, Hollander ND, Hoffer MJV, Reijnders MRF, Demirdas S, Koboldt DC, Bartholomew D, Mosher TM, Hickey SE, Shieh C, Sanchez-Lara PA, Graham JM Jr, Tezcan K, Schaefer GB, Danylchuk NR, Asamoah A, Jackson KE, Yachelevich N, Au M, Pérez-Jurado LA, Kleefstra T, Penzes P, Wood SA, Burne T, Pierson TM, Piper M, Gécz J, Jolly LA. Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor β Signaling. Biol Psychiatry. 2020 Jan 15;87(2):100-112. doi: 10.1016/j.biopsych.2019.05.028. Epub 2019 Jun 29. PubMed [citation] PMID: 31443933, PMCID: PMC6925349

173. Geng LN, Kohler JN, Levonian P; Members of the Undiagnosed Diseases Network (…Malicdan MC), Bernstein JA, Ford JM, Ahuja N, Witteles R, Hom J, Wheeler M. Genomics in medicine: a novel elective rotation for internal medicine residents. Postgrad Med J. 2019 Oct;95(1128):569-572. doi: 10.1136/postgradmedj-2018-136355. Epub 2019 Aug 22. PubMed [citation] PMID: 31439813

174. Bhatia A, Mobley BC, Cogan J, Koziura ME, Brokamp E, Phillips J, Newman J; Undiagnosed Diseases Network (UDN)., Moore SA, Hamid R; Members of the Undiagnosed Diseases Network (…Malicdan MC). Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clin Imaging. 2019 Nov - Dec;58:108-113. doi: 10.1016/j.clinimag.2019.06.010. Epub 2019 Jun 21. PubMed [citation] PMID: 31299614, PMCID: PMC6893088

175. Shashi V, Geist J, Lee Y, Yoo Y, Shin U, Schoch K, Sullivan J, Stong N, Smith E, Jasien J, Kranz P; Undiagnosed Diseases Network (…Malicdan MC), Lee Y, Shin YB, Wright NT, Choi M, Kontrogianni-Konstantopoulos A. Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Hum Mutat. 2019 Aug;40(8):1115-1126. doi: 10.1002/humu.23760. Epub 2019 May 5. PubMed [citation] PMID: 31264822, PMCID: PMC6688907

176. Frésard L, Smail C, Ferraro NM, Teran NA, Li X, Smith KS, Bonner D, Kernohan KD, Marwaha S, Zappala Z, Balliu B, Davis JR, Liu B, Prybol CJ, Kohler JN, Zastrow DB, Reuter CM, Fisk DG, Grove ME, Davidson JM, Hartley T, Joshi R, Strober BJ, Utiramerur S; Undiagnosed Diseases Network (…. Malicdan MCV …); Care4Rare Canada Consortium, Lind L, Ingelsson E, Battle A, Bejerano G, Bernstein JA, Ashley EA, Boycott KM, Merker JD, Wheeler MT, Montgomery SB. Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. **Nat Med**. 2019 Jun;25(6):911-919. doi: 10.1038/s41591-019-0457-8. Epub 2019 Jun 3. PMID: 31160820.

177. Newman JH, Shaver A, Sheehan JH, Mallal S, Stone JH, Pillai S, Bastarache L, Riebau D, Allard-Chamard H, Stone WM, Perugino C, Pilkinton M, Smith SA, McDonnell WJ, Capra JA, Meiler J, Cogan J, Xing K, Mahajan VS, Mattoo H, Hamid R, Phillips JA 3rd; Undiagnosed Disease Network (… Malicdan MCV …). IgG4-related disease: Association with a rare gene variant expressed in cytotoxic T cells. **Mol Genet Genomic Med**. 2019 Apr 16:e686. doi: 10.1002/mgg3.686. [Epub ahead of print]. PMID: 30993913.

178. Walley NM, Pena LDM, Hooper SR, Cope H, Jiang Y-H, McConkie-Rosell A, Sanders C, Schoch K, Spillmann RC, Strong K, McCray AT, Mazur P, Esteves C, LeBlanc K, Undiagnosed Diseases N (… Malicdan MCV …), Wise AL, Shashi V,. Characteristics of undiagnosed diseases network applicants: implications for referring providers. **BMC Health Serv Res**. 2018;18(1):652. PubMed PMID: Medline:30134969. English.

179. Zastrow DB, Kohler JN, Bonner D, Reuter CM, Fernandez L, Grove ME, Fisk DG; Undiagnosed Diseases Network, Yang Y, Eng CM, Ward PA, Bick D, Worthey EA, Fisher PG, Ashley EA, Bernstein JA, Wheeler MT. A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing. **J Genet Couns**. PMID: 30964584

180. Burrage LC, Reynolds JJ, Baratang NV, Phillips JB, Wegner J, McFarquhar A, Higgs MR, Christiansen AE, Lanza DG, Seavitt JR, Jain M, Li X, Parry DA, Raman V, Chitayat D, Chinn IK, Bertuch AA, Karaviti L, Schlesinger AE, Earl D, Bamshad M, Savarirayan R, Doddapaneni H, Muzny D, Jhangiani SN, Eng CM, Gibbs RA, Bi W, Emrick L, Rosenfeld JA, Postlethwait J, Westerfield M, Dickinson ME, Beaudet AL, Ranza E, Huber C, Cormier-Daire V, Shen W, Mao R, Heaney JD, Orange JS; University of Washington Center for Mendelian Genomics; Undiagnosed Diseases Network (… Malicdan MCV …), Bertola D, Yamamoto GL, Baratela WAR, Butler MG,

Ali A, Adeli M, Cohn DH, Krakow D, Jackson AP, Lees M, Offiah AC, Carlston CM, Carey JC, Stewart GS, Bacino CA, Campeau PM, Lee B. Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. **Am J Hum Genet**. 2019 Mar 7;104(3):422-438. doi: 10.1016/j.ajhg.2019.01.007. Epub 2019 Feb 14. PMID: 30773277

181. Grove ME, White S, Fisk DG, Rego S, Dagan-Rosenfeld O, Kohler JN, Reuter CM, Bonner D; Undiagnosed Diseases Network (… Malicdan MCV …), Wheeler MT, Bernstein JA, Ormond KE, Hanson-Kahn AK. Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students. **J Genet Couns**. 2019 Apr;28(2):466-476. doi: 10.1002/jgc4.1094. Epub 2019 Feb 1. PMID: 30706981.
182. Kelly M, Park M, Mihalek I, Rochtus A, Gramm M, Pérez-Palma E, Axeen ET, Hung CY, Olson H, Swanson L, Anselm I, Briere LC, High FA, Sweetser DA; Undiagnosed Diseases Network (… Malicdan MCV …), Kayani S, Snyder M, Calvert S, Scheffer IE, Yang E, Waugh JL, Lal D, Bodamer O, Poduri A. Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region. **Epilepsia**. 2019 Mar;60(3):406-418. doi: 10.1111/epi.14653. Epub 2019 Jan 25. PMID: 30682224.
183. Machol K, Rousseau J, Ehresmann S, Garcia T, Nguyen TTM, Spillmann RC, Sullivan JA, Shashi V, Jiang YH, Stong N, Fiala E, Willing M, Pfundt R, Kleefstra T, Cho MT, McLaughlin H, Rosello Piera M, Orellana C, Martínez F, Caro-Llopis A, Monfort S, Roscioli T, Nixon CY, Buckley MF, Turner A, Jones WD, van Hasselt PM, Hofstede FC, van Gassen KLI, Brooks AS, van Slegtenhorst MA, Lachlan K, Sebastian J, Madan-Khetarpal S, Sonal D, Sakkubai N, Thevenon J, Faivre L, Maurel A, Petrovski S, Krantz ID, Tarpinian JM, Rosenfeld JA, Lee BH; Undiagnosed Diseases Network (… Malicdan MCV …), Campeau PM. Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. Am J Hum Genet. 2019 Jan 3;104(1):164-178. doi: 10.1016/j.ajhg.2018.11.007. Epub 2018 Dec 20. PMID: 30580808
184. Tan QKG, Cope H, Spillmann RC, Stong N, Jiang Y-H, McDonald MT, Rothman JA, Butler MW, Frush DP, Lachman RS, Lee B, Bacino CA, Bonner MJ, McCall CM, Pendse AA, Walley N, Undiagnosed Diseases N (… Malicdan MCV …), Shashi V, Pena LDM. Further evidence for the involvement of EFL1 in a Shwachman-Diamond-like syndrome and expansion of the phenotypic features. **Cold Spring Harb Mol Case Stud**. 2018;4(5). PubMed PMID: Medline:29970384. English.
185. Splinter K, Adams DR, Bacino CA, Bellen HJ, Bernstein JA, Cheatle-Jarvela AM, Eng CM, Esteves C, Gahl WA, Hamid R, Jacob HJ, Kikani B, Koeller DM, Kohane IS, Lee BH, Loscalzo J, Luo X, McCray AT, Metz TO, Mulvihill JJ, Nelson SF, Palmer CGS, Phillips JA, 3rd, Pick L, Postlethwait JH, Reuter C, Shashi V, Sweetser DA, Tifft CJ, Walley NM, Wangler MF, Westerfield M, Wheeler MT, Wise AL, Worthey EA, Yamamoto S, Ashley EA, Undiagnosed Diseases N (… Malicdan MCV …). Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. **N Engl J Med. 2018**;379(22):2131-9. PubMed PMID: Medline:30304647. English.
186. Shashi V, Schoch K, Spillmann R, Cope H, Tan QKG, Walley N, Pena L, McConkie-Rosell A, Jiang Y-H, Stong N, Need AC, Goldstein DB, Undiagnosed Diseases N (… Malicdan MCV …). A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. **Genetics in medicine** : official journal of the American College of Medical Genetics. 2018. PubMed PMID: Medline:29907797. English.
187. Pena LDM, Jiang Y-H, Schoch K, Spillmann RC, Walley N, Stong N, Rapisardo Horn S, Sullivan JA, McConkie-Rosell A, Kansagra S, Smith EC, El-Dairi M, Bellet J, Keels MA, Jasien J, Kranz PG, Noel R, Nagaraj SK, Lark RK, Wechsler DSG, Del Gaudio D, Leung ML, Hendon LG, Parker CC, Jones KL, Undiagnosed Diseases Network M (… Malicdan MCV …), Goldstein DB, Shashi V. Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. **Genetics in medicine** : official journal of the American College of Medical Genetics. 2018;20(4):464-9. PubMed PMID: Medline:28914269. English.
188. Olahova M, Yoon WH, Thompson K, Jangam S, Fernandez L, Davidson JM, Kyle JE, Grove ME, Fisk DG, Kohler JN, Holmes M, Dries AM, Huang Y, Zhao C, Contrepois K, Zappala Z, Fresard L, Waggott D, Zink EM, Kim Y-M, Heyman HM, Stratton KG, Webb-Robertson B-JM, Undiagnosed Diseases N (… Malicdan MCV …), Snyder M, Merker JD, Montgomery SB, Fisher PG, Feichtinger RG, Mayr JA, Hall J, Barbosa IA, Simpson MA, Deshpande C, Waters KM, Koeller DM, Metz TO, Morris AA, Schelley S, Cowan T, Friederich MW, McFarland R, Van Hove JLK, Enns GM, Yamamoto S, Ashley EA, Wangler MF, Taylor RW, Bellen HJ, Bernstein JA,

Wheeler MT. Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. **Am J Hum Genet**. 2018;102(3):494-504. PubMed PMID: Medline:29478781. English.

189. Marcogliese PC, Shashi V, Spillmann RC, Stong N, Rosenfeld JA, Koenig MK, Martinez-Agosto JA, Herzog M, Chen AH, Dickson PI, Lin HJ, Vera MU, Salamon N, Graham JM, Jr., Ortiz D, Infante E, Steyaert W, Dermaut B, Poppe B, Chung H-L, Zuo Z, Lee P-T, Kanca O, Xia F, Yang Y, Smith EC, Jasien J, Kansagra S, Spiridigliozzi G, El-Dairi M, Lark R, Riley K, Koeberl DD, Golden-Grant K, Program for Undiagnosed D, Undiagnosed Diseases N (… Malicdan MCV …), Yamamoto S, Wangler MF, Mirzaa G, Hemelsoet D, Lee B, Nelson SF, Goldstein DB, Bellen HJ, Pena LDM.. IRF2BPL Is Associated with Neurological Phenotypes. **Am J Hum Genet**. 2018;103(3):456. PubMed PMID: Medline:30193138. English.
190. Machol K, Rousseau J, Ehresmann S, Garcia T, Nguyen TTM, Spillmann RC, Sullivan JA, Shashi V, Jiang Y-H, Stong N, Fiala E, Willing M, Pfundt R, Kleefstra T, Cho MT, McLaughlin H, Rosello Piera M, Orellana C, Martinez F, Caro-Llopis A, Monfort S, Roscioli T, Nixon CY, Buckley MF, Turner A, Jones WD, van Hasselt PM, Hofstede FC, van Gassen KLI, Brooks AS, van Slegtenhorst MA, Lachlan K, Sebastian J, Madan-Khetarpal S, Sonal D, Sakkubai N, Thevenon J, Faivre L, Maurel A, Petrovski S, Krantz ID, Tarpinian JM, Rosenfeld JA, Lee BH, Undiagnosed Diseases N (… Malicdan MCV …), Campeau PM. Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. **Am J Hum Genet**. 2018. PubMed PMID: Medline:30580808. English.
191. Wang J, Al-Ouran R, Hu Y, Kim S-Y, Wan Y-W, Wangler MF, Yamamoto S, Chao H-T, Comjean A, Mohr SE, Udn, Perrimon N, Liu Z, Bellen HJ, and Undiagnosed Diseases Network collaborators (… Malicdan MCV …). MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. **Am J Hum Genet**. 2017;100(6):843-53. PubMed PMID: Medline:28502612. English.
192. Spillmann RC, McConkie-Rosell A, Pena L, Jiang Y-H, Undiagnosed Diseases N (… Malicdan MCV …), Schoch K, Walley N, Sanders C, Sullivan J, Hooper SR, Shashi V.. A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network. **Orphanet J Rare Dis**. 2017;12(1):71. PubMed PMID: Medline:28416019. English.
193. Schoch K, Meng L, Szelinger S, Bearden DR, Stray-Pedersen A, Busk OL, Stong N, Liston E, Cohn RD, Scaglia F, Rosenfeld JA, Tarpinian J, Skraban CM, Deardorff MA, Friedman JN, Akdemir ZC, Walley N, Mikati MA, Kranz PG, Jasien J, McConkie-Rosell A, McDonald M, Wechsler SB, Freemark M, Kansagra S, Freedman S, Bali D, Millan F, Bale S, Nelson SF, Lee H, Dorrani N, Center UCG, Undiagnosed Diseases N (… Malicdan MCV …), Goldstein DB, Xiao R, Yang Y, Posey JE, Martinez-Agosto JA, Lupski JR, Wangler MF, Shashi V. A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. **Am J Hum Genet**. 2017;100(2):343-51. PubMed PMID: Medline:28132692. English.
194. Ramoni RB, Mulvihill JJ, Adams DR, Allard P, Ashley EA, Bernstein JA, Gahl WA, Hamid R, Loscalzo J, McCray AT, Shashi V, Tifft CJ, Undiagnosed Diseases N (… Malicdan MCV …), Wise AL. The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. **Am J Hum Genet**. 2017;100(2):185-92. PubMed PMID: Medline:28157539. English.
195. Shashi V, Pena LDM, Kim K, Burton B, Hempel M, Schoch K, Walkiewicz M, McLaughlin HM, Cho M, Stong N, Hickey SE, Shuss CM, Undiagnosed Diseases N (… Malicdan MCV …), Freemark MS, Bellet JS, Keels MA, Bonner MJ, El-Dairi M, Butler M, Kranz PG, Stumpel CTRM, Klinkenberg S, Oberndorff K, Alawi M, Santer R, Petrovski S, Kuismin O, Korpi-Heikkila S, Pietilainen O, Aarno P, Kurki MI, Hoischen A, Need AC, Goldstein DB, Kortum F. De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. **Am J Hum Genet**. 2016;99(4):991-9. PubMed PMID: Medline:27693232. English.
196. Klionsky DJ, Abdalla FC, Abeliovich H, Abraham RT, Acevedo-Arozena A, … Malicdan MCV … Zuckerbraun B. Guidelines for the use and interpretation of assays for monitoring autophagy. **Autophagy**. 2012;8(4):445-544. PubMed PMID: Medline:22966490. English.

PATENTS

- Patent Application No. US 20120264928 A1, Satoru Noguchi, May Christine Malicdan, Ichizo Nishino (Inventors). Therapeutic pharmaceutical agent for diseases associated with decrease in function of gne protein, food composition, and food additive.

- Gne-/-hGNED176VTg as a mouse model for DMRV/HIBM (Japanese patent-2010). Satoru Noguchi, Nishino Kazumi, May Christine Malicdan (Inventors)
- NIH provisional patent No. 62/120,742 (filed Feb 2015) “Sialylation-Increasing Therapies for Diseases Associated with Oxidative Stress”
- Patent Application No. 15/061829, filed on March 4, 2016, entitled “CANNABINOID RECEPTOR MEDIATING COMPOUNDS.