Thomas Müller

Biographical sketch

As a Pediatrician I was privileged to encounter children with rare unknown liver and gastrointestinal disorders. In close collaboration with Andreas Janecke and Lukas Huber we further characterize these "new" diseases applying next generation sequencing and cell culture models including organoids derived from hepatic and intestinal stem cells. In the last years we are able to unravel the molecular basis of five congenital enteropathies such as Microvillus inclusion disease and congenital sodium diarrhea.

Curriculum vitae

Medical University of Innsbruck
Department of Pediatrics I
Anichstrasse 35
6020 Innsbruck
Austria
Tel: +43-512-504-26345
Web: https://kinderzentrum.tirol-kliniken.at/
Email: thomas.mueller@i-med.ac.at
ORCID: 0000-0001-8908-5589

Date of birth	6 July 1970
Place of birth	Hohenems, Austria
Citizenship	Austrian

Education

1988	Matura, Bundesrealgymnasium Reutte, Austria
1996	Medical doctor, University of Innsbruck, Austria

Career History

1998-2004	Resident at the Department of Pediatrics, Medical University of Innsbruck
2003	Venia legendi (Habilitation) for "Pediatrics"
since 2003	Associate Professor of Pediatrics, Department of Pediatrics I, Medical University of Innsbruck
2006	Appointment to the University of Bonn as a Professor of Pediatric Gastroenterology and Hepatology – DECLINED
2015	Primo loco as a Professor of Pediatrics, Medical University of Graz
2016	Appointment to the Medical University of Innsbruck as a Professor for Pediatrics Director of the Department of Pediatrics I, Medical University of Innsbruck

Fellowships

Department of Pathology, University of Innsbruck (1996-98)
Research fellow at the Institute of Medical Genetics (Prof. PL Pearson), University of Utrecht, The Netherlands (1998)
Clinical fellow im Children´s Hospital, University of Sheffield (Prof. MS Tanner) and The Liver Unit, Children´s Hospital, University of Birmingham (Prof. DA Kelly) (2003)

Awards

Research Award Innsbruck, Medical University (1996)
Research Award of the Austrian Society of Pediatrics (1997)
Research Award of the Princedom of Liechtenstein (2001)
Research Award of the Austrian Society of Pediatrics (2005)
Paul-Caspar-Tyrell-Price of the German Society for Pediatric Gastroenterology and Nutrition (2007)

Research Award of the Austrian Society of Pediatrics "Clemens von Pirquet Award" of the Austrian Society of Pediatrics (2013)
Research Award of the Austrian Society of Pediatrics (2014)

Publications Number of publication=79, h-index=29, cited>2568
Google Scholar link

Patents None

Other Functions Reviewer for: GUT, Human Mutation, Journal of Clinical Investigation, Journal of Inherited Metabolic Disease, Journal of Pediatric Gastroenterology and Nutrition, Journal of Pediatrics, PLoS One, Transplant International

Research Interests Identification and functional characterization of gene defects in patients with rare inborn errors of metabolism

Funds obtained (in €, 5 most important ones)

International Copper Association (ICA) Nr. TPT0551-99	95.000	1999-2001
Tiroler Wissenschaftsfonds (TWF): UNI-0404/142	33.000	2005-2007
Österreichische Nationalbank (OeNB) Jubiläumsfonds Nr.14496	99.800	2012-2013
Else Kröner-Fresenius-Stiftung Nr.: 2013_A230	99.950	2014-2015
Österreichische Nationalbank (OeNB) Jubiläumsfonds Nr.16678	150.000	2016-2018

PhD Students supervised by faculty since 2013

Name (PhD Program)	Title of PhD thesis	Start	Graduation	publications
Cornelia Thoeni (co-supervision with LAH), (MCB)	Molecular mechanisms in the pathogenesis of Microvillus Inclusion Disease (MVID)	2011	2013	4
Georg Friedrich Vogel (co-supervision with LAH) (MCB)	Myo5b-Syntaxin3 interplay drives cargo-selective apical exocytosis in enterocaytes	2010	2015	8
Iris Krainer (co-supervision with LAH) (MCBO)	Establishment and analyses of organoids from patients with congenital entheropathies and liver diseases	2015	ongoing	1
Katharina Klee (co-supervision with LAH) (MCBO)	Investigation of cargo-selective apical transport in polarized epithelial cells	2016	ongoing	1

International collaborators

	Project	**Joint public.**	**lab for stay abroad**
Hans Clevers (Hubrecht Institute, University Utrecht, NL)	Intestinal and liver organoids	2	yes
Mark Donowitz (Division of Gastroenterology, Johns Hopkins University, USA)	Sodium transport in intestinal organoids	1	yes
Paul Gissen (MRC Laboratory for Molecular Cell Biology, UC-London, UK)	VPS33B mutations affecting Rab Protein interaction	1	yes

Research Network: ENRICA (European Reference Network on Rare inherited and congenital diseases)

Thomas Müller; 10 most important scientific publications

1. Janecke AR, Heinz-Erian P, Yin J, Petersen BS, Franke A, Lechner S, Fuchs I, Melancon S, Uhlig HH, Travis S, Marinier E, Perisic V, Ristic N, Gerner P, Booth IW, Wedenoja S, Baumgartner N, Vodopiutz J, Frechette-Duval MC, De Lafollie J, Persad R, Warner N, Tse CM, Sud K, Zachos NC, Sarker R, Zhu X, Muise AM, Zimmer KP, Witt H, Zoller H, Donowitz M, **Müller T**. Reduced sodium/proton exchanger NHE3 activity causes congenital sodium diarrhea. **Hum Mol Genet**. 2015 Dec1;24(23):6614-23. doi: 10.1093/hmg/ddv367. PMID: 26358773;

2. **Müller T**, Rasool I, Heinz-Erian P, Mildenberger E, Hülstrunk C, Müller A, Michaud L, Koot BG, Ballauff A, Vodopiutz J, Rosipal S, Petersen BS, Franke A, Fuchs I, Witt H, Zoller H, Janecke AR, Visweswariah SS. Congenital secretory diarrhoea caused by activating germline mutations in GUCY2C. **Gut.** 2016 Aug;65(8):1306-13. doi: 10.1136/gutjnl-2015-309441. PMID: 25994218;

3. Wiegerinck CL, Janecke AR, Schneeberger K, Vogel GF, van Haaften-Visser DY, Escher JC, Adam R, Thöni CE, Pfaller K, Jordan AJ, Weis CA, Nijman IJ, Monroe GR, van Hasselt PM, Cutz E, Klumperman J, Clevers H, Nieuwenhuis EE, Houwen RH, van Haaften G, Hess MW, Huber LA, Stapelbroek JM, **Müller T***, Middendorp S*. Loss of syntaxin 3 causes variant microvillus inclusion disease. **Gastroenterology**. 2014 Jul;147(1):65-68.e10. doi: 10.1053/j.gastro.2014.04.002. PMID: 24726755. *shared last authors

4. **Müller T**, Mizumoto S, Suresh I, Komatsu Y, Vodopiutz J, Dundar M, Straub V, Lingenhel A, Melmer A, Lechner S, Zschocke J, Sugahara K, Janecke AR. Loss of dermatan sulfate epimerase (DSE) function results in musculocontractural Ehlers-Danlos syndrome. **Hum Mol Genet**. 2013 Sep 15;22(18):3761-72. doi: 10.1093/hmg/ddt227. PMID: 23704329.

5. Heinz-Erian P*, **Müller T***, Krabichler B, Schranz M, Becker C, Rüschendorf F, Nürnberg P, Rossier B, Vujic M, Booth IW, Holmberg C, Wijmenga C, Grigelioniene G, Kneepkens CM, Rosipal S, Mistrik M, Kappler M, Michaud L, Dóczy LC, Siu VM, Krantz M, Zoller H, Utermann G, Janecke AR. Mutations in SPINT2 cause a syndromic form of congenital sodium diarrhea. **Am J Hum Genet** 2009 Feb;84(2):188-96. doi: 10.1016/j.ajhg.2009.01.004. PMID: 19185281; *shared first authors

6. **Müller T**, Hess MW, Schiefermeier N, Pfaller K, Ebner HL, Heinz-Erian P, Ponstingl H, Partsch J, Röllinghoff B, Köhler H, Berger T, Lenhartz H, Schlenck B, Houwen RJ, Taylor CJ, Zoller H, Lechner S, Goulet O, Utermann G, Ruemmele FM, Huber LA, Janecke AR. MYO5B mutations cause microvillus inclusion disease and disrupt epithelial cell polarity. **Nat Genet** 2008 Oct;40(10):1163-5. doi: 10.1038/ng.225. PMID: 18724368.

7. **Müller T**, Langner C, Fuchsbichler A, Heinz-Erian P, Ellemunter H, Schlenck B, Bavdekar AR, Pradhan AM, Pandit A, Müller-Höcker J, Melter M, Kobayashi K, Nagasaka H, Kikuta H, Müller W, Tanner MS, Sternlieb I, Zatloukal K, Denk H. Immunohistochemical analysis of Mallory bodies in Wilsonian and non-Wilsonian hepatic copper toxicosis. **Hepatology** 2004 Apr;39(4):963-9. PMID: 15057900.

8. **Müller T**, van de Sluis B, Zhernakova A, van Binsbergen E, Janecke AR, Bavdekar A, Pandit A, Weirich-Schwaiger H, Witt H, Ellemunter H, Deutsch J, Denk H, Müller W, Sternlieb I, Tanner MS, Wijmenga C. The canine copper toxicosis gene MURR1 does not cause non-Wilsonian hepatic copper toxicosis. **J Hepatol** 2003 Feb;38(2):164-8. PMID: 12547404.

9. **Müller T**, Wijmenga C, Phillips AD, Janecke A, Houwen RH, Fischer H, Ellemunter H, Frühwirth M, Offner F, Hofer S, Müller W, Booth IW, Heinz-Erian P. Congenital sodium diarrhea is an autosomal recessive disorder of sodium/proton exchange but unrelated to known candidate genes. **Gastroenterology** 2000 Dec;119(6):1506-13. PMID: 11113072.

10. **Müller T**, Feichtinger H, Berger H, Muller W. Endemic Tyrolean infantile cirrhosis: an ecogenetic disorder. **Lancet** 1996 Mar 30;347(9005):877-80. PMID: 8622397.

Thomas Müller; all publications since 2013

1. van Rijn JM, Ardy RC, Kuloğlu Z, Härter B, van Haaften-Visser DY, van der Doef HPJ, van Hoesel M, Kansu A, van Vugt AHM, Ng M, Kokke FTM, Krolo A, Başaran MK, Kaya NG, Ünlüsoy Aksu A, Dalgıç B, Ozcay F, Baris Z, Kain R, Stigter ECA, Lichtenbelt KD, Massink MPG, Duran KJ, Verheij JBGM, Lugtenberg D, Nikkels PGJ, Brouwer HGF, Verkade HJ, Scheenstra R, Spee B, Nieuwenhuis EES, Coffer PJ, Janecke AR, van Haaften G, Houwen RHJ, **Müller T**, Middendorp S, Boztug K. Intestinal failure and aberrant lipid metabolism in patients with DGAT1 deficiency. **Gastroenterology.** 2018 Mar 28. pii: S0016-5085(18)30347-0. doi: 10.1053/j.gastro.2018.03.040. [Epub ahead of print] PubMed PMID: 29604290.

2. Danescu S, Has C, Baican C, **Müller T,** Baican A. A novel IKBKG mutation in a patient with incontinentia pigmenti and features of hepatic ciliopathy. **Australas J Dermatol.** 2018 Mar 8. doi: 10.1111/ajd.12805. [Epub ahead of print] PubMed PMID: 29520766.

3. Niederwanger C, Lechner S, König L, Janecke AR, Pototschnig C, Häussler B, Scholl-Bürgi S, **Müller T,** Heinz-Erian P. Isolated choanal and gut atresias: pathogenetic role of serine protease inhibitor type 2 (SPINT2) gene mutations unlikely. **Eur J Med Res.** 2018 Mar 2;23(1):13. doi: 10.1186/s40001-018-0312-2. PubMed PMID: 29499739; PubMed Central PMCID: PMC5834866.

4. Rosendahl J, Kirsten H, Hegyi E, Kovacs P, Weiss FU, Laumen H, Lichtner P, Ruffert C, Chen JM, Masson E, Beer S, Zimmer C, Seltsam K, Algül H, Bühler F, Bruno MJ, Bugert P, Burkhardt R, Cavestro GM, Cichoz-Lach H, Farré A, Frank J, Gambaro G, Gimpfl S, Grallert H, Griesmann H, Grützmann R, Hellerbrand C, Hegyi P, Hollenbach M, Iordache S, Jurkowska G, Keim V, Kiefer F, Krug S, Landt O, Leo MD, Lerch MM, Lévy P, Löffler M, Löhr M, Ludwig M, Macek M, Malats N, Malecka-Panas E, Malerba G, Mann K, Mayerle J, Mohr S, Te Morsche RHM, Motyka M, Mueller S, **Müller T**, Nöthen MM, Pedrazzoli S, Pereira SP, Peters A, Pfützer R, Real FX, Rebours V, Ridinger M, Rietschel M, Rösmann E, Saftoiu A, Schneider A, Schulz HU, Soranzo N, Soyka M, Simon P, Skipworth J, Stickel F, Strauch K, Stumvoll M, Testoni PA, Tönjes A, Werner L, Werner J, Wodarz N, Ziegler M, Masamune A, Mössner J, Férec C, Michl P, P H Drenth J, Witt H, Scholz M, Sahin-Tóth M; all members of the PanEuropean Working group on ACP. Genome-wide association study identifies inversion in the CTRB1-CTRB2 locus to modify risk for alcoholic and non-alcoholic chronic pancreatitis. **Gut.** 2017 Jul 28. pii: gutjnl-2017-314454. doi: 10.1136/gutjnl-2017-314454. [Epub ahead of print] PubMed PMID: 28754779.

5. Vogel GF, van Rijn JM, Krainer IM, Janecke AR, Posovszky C, Cohen M, Searle C, Jantchou P, Escher JC, Patey N, Cutz E, **Müller T,** Middendorp S, Hess MW, Huber LA. Disrupted apical exocytosis of cargo vesicles causes enteropathy in FHL5 patients with Munc18-2 mutations. **JCI Insight.** 2017 Jul 20;2(14). pii: 94564. doi: 10.1172/jci.insight.94564. [Epub ahead of print] PubMed PMID: 28724787; PubMed Central PMCID: PMC5518552.

6. Vogel GF, Janecke AR, Krainer IM, Gutleben K, Witting B, Mitton SG, Mansour S, Ballauff A, Roland JT, Engevik AC, Cutz E, **Müller T**, Goldenring JR, Huber LA, Hess MW. Abnormal Rab11-Rab8-vesicles cluster in enterocytes of patients with microvillus inclusion disease. **Traffic.** 2017 Jul;18(7):453-464. doi:10.1111/tra.12486. Epub 2017 May 17. PubMed PMID: 28407399; PubMed Central PMCID:PMC5693299.

7. Janecke AR, Xu R, Steichen-Gersdorf E, Waldegger S, Entenmann A, Giner T, Krainer I, Huber LA, Hess MW, Frishberg Y, Barash H, Tzur S, Schreyer-Shafir N, Sukenik-Halevy R, Zehavi T, Raas-Rothschild A, Mao C, **Müller T.** Deficiency of the sphingosine-1-phosphate lyase SGPL1 is associated with congenital nephrotic syndrome and congenital adrenal calcifications. **Hum Mutat.** 2017 Apr;38(4):365-372. doi: 10.1002/humu.23192. Epub 2017 Mar 6. PubMed PMID: 28181337; PubMed Central PMCID: PMC5384969.

8. Binder E, Loinger M, Mühlbacher A, Edlinger M, Steichen E, Meraner D, Loacker L, Weigel G, **Müller T,** Fröhlich-Reiterer E, Hofer SE. Genotyping of coeliac-specific human leucocyte antigen in children with type 1 diabetes: does this screening method make sense? **Arch Dis Child.** 2017 Jul;102(7):603-606. doi:10.1136/archdischild-2016-311610. Epub 2016 Dec 30. PubMed PMID: 28039141.

9. Gruber R, Rogerson C, Windpassinger C, Banushi B, Straatman-Iwanowska A, Hanley J, Forneris F, Strohal R, Ulz P, Crumrine D, Menon GK, Blunder S, Schmuth M, **Müller T,** Smith H, Mills K, Kroisel P, Janecke AR, Gissen P. Autosomal Recessive Keratoderma-Ichthyosis-Deafness (ARKID) Syndrome Is Caused by VPS33B Mutations Affecting Rab Protein Interaction and Collagen Modification. **J Invest Dermatol.** 2017 Apr;137(4):845-854. doi: 10.1016/j.jid.2016.12.010. Epub 2016 Dec 23. Review. PubMed PMID: 28017832.

10. Baumann M, Steichen-Gersdorf E, Krabichler B, Petersen BS, Weber U, Schmidt WM, Zschocke J, **Müller T**, Bittner RE, Janecke AR. Homozygous SYNE1 mutation causes congenital onset of muscular weakness with distal arthrogryposis: a genotype-phenotype correlation. **Eur J Hum Genet**. 2016 Oct 26. doi: 10.1038/ejhg.2016.144. [Epub ahead of print] PMID: 27782104.

11. Hagenbuchner J, Rupp M, Salvador C, Meister B, Kiechl-Kohlendorfer U, **Müller T**, Geiger K, Sergi C, Obexer P, Ausserlechner MJ. Nuclear FOXO3 predicts adverse clinical outcome and promotes tumor angiogenesis in neuroblastoma. **Oncotarget.** 2016 Oct 18. doi: 10.18632/oncotarget.12728. [Epub ahead of print] PMID: 27769056.

12. Vodopiutz J, Mizumoto S, Lausch E, Rossi A, Unger S, Janocha N, Costantini R, Seidl R, Greber-Platzer S, Yamada S, **Müller T**, Jilma B, Ganger R, Superti-Furga A, Ikegawa S, Sugahara K, Janecke AR. Chondroitin Sulfate N-acetylgalactosaminyltransferase-1 (CSGalNAcT-1) Deficiency Results in a Mild Skeletal Dysplasia and Joint Laxity. **Hum Mutat.** 2016 Sep 7. doi: 10.1002/humu.23070. [Epub ahead of print] PMID: 27599773.

13. Kopajtich R, Murayama K, Janecke AR, Haack TB, Breuer M, Knisely AS, Harting I, Ohashi T, Okazaki Y, Watanabe D, Tokuzawa Y, Kotzaeridou U, Kölker S, Sauer S, Carl M, Straub S, Entenmann A, Gizewski E, Feichtinger RG, Mayr JA, Lackner K, Strom TM, Meitinger T, **Müller T**, Ohtake A, Hoffmann GF, Prokisch H, Staufner C. Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. **Am J Hum Genet.** 2016 Aug 4;99(2):414-22. doi: 10.1016/j.ajhg.2016.05.027. PMID: 27426735.

14. Jansen EJ, Timal S, Ryan M, Ashikov A, van Scherpenzeel M, Graham LA, Mandel H, Hoischen A, Iancu TC, Raymond K, Steenbergen G, Gilissen C, Huijben K, van Bakel NH, Maeda Y, Rodenburg RJ, Adamowicz M, Crushell E, Koenen H, Adams D, Vodopiutz J, Greber-Platzer S, **Müller T**, Dueckers G, Morava E, Sykut-Cegielska J, Martens GJ, Wevers RA, Niehues T, Huynen MA, Veltman JA, Stevens TH, Lefeber DJ. ATP6AP1 deficiency causes an immunodeficiency with hepatopathy, cognitive impairment and abnormal protein glycosylation. **Nat Commun.** 2016 May 27;7:11600. doi: 10.1038/ncomms11600. PMID: 27231034.

15. Janecke AR, Heinz-Erian P, **Müller T**. Mechanisms Underlying Dysregulation of Electrolyte Absorption in Inflammatory Bowel Disease-Associated Diarrhea**. Inflamm Bowel Dis**. 2016 Jun;22(6):E17-8. doi: 10.1097/MIB.0000000000000778. PMID: 27104826.

16. Janecke AR, Heinz-Erian P, **Müller T**. Congenital Sodium Diarrhea: A Form of Intractable Diarrhea, With a Link to Inflammatory Bowel Disease. **J Pediatr Gastroenterol Nutr.** 2016 Aug;63(2):170-6. doi: 10.1097/MPG.0000000000001139. PMID: 26835907.

17. Vogel GF, Hess MW, Pfaller K, Huber LA, Janecke AR, **Müller T**. Towards understanding microvillus inclusion disease. **Mol Cell Pediatr**. 2016 Dec;3(1):3. doi: 10.1186/s40348-016-0031-0. PMID: 26830108.

18. Vogel GF, Klee KM, Janecke AR, **Müller T**, Hess MW, Huber LA. Cargo-selective apical exocytosis in epithelial cells is conducted by Myo5B, Slp4a, Vamp7, and Syntaxin 3. J Cell Biol. 2015 Nov 9;211(3):587-604. doi: 10.1083/jcb.201506112. PMID: 26553929.

19. Härter B, Fuchs I, **Müller T**, Akbulut UE, Cakir M, Janecke AR. Early Clinical Diagnosis of PC1/3 Deficiency in a Patient With a Novel Homozygous PCSK1 Splice-Site Mutation. **J Pediatr Gastroenterol Nutr**. 2016 Apr;62(4):577-80. doi: 10.1097/MPG.0000000000001018. PMID: 26488123.

20. Schneeberger K, Vogel GF, Teunissen H, van Ommen DD, Begthel H, El Bouazzaoui L, van Vugt AH, Beekman JM, Klumperman J, **Müller T**, Janecke A, Gerner P, Huber LA, Hess MW, Clevers H, van Es JH, Nieuwenhuis EE, Middendorp S. An inducible mouse model for microvillus inclusion disease reveals a role for myosin Vb in apical and basolateral trafficking. **Proc Natl Acad Sci** U S A. 2015 Oct 6;112(40):12408-13. doi: 10.1073/pnas.1516672112. PMID: 26392529.

21. Janecke AR, Li B, Boehm M, Krabichler B, Rohrbach M, **Müller T**, Fuchs I, Golas G, Katagiri Y, Ziegler SG, Gahl WA, Wilnai Y, Zoppi N, Geller HM, Giunta C, Slavotinek A, Steinmann B. The phenotype of the musculocontractural type of Ehlers-Danlos syndrome due to CHST14 mutations. **Am J Med Genet A**. 2016 Jan;170A(1):103-15. doi: 10.1002/ajmg.a.37383. PMID: 26373698.

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23. Vodopiutz J, Seidl R, Prayer D, Khan MI, Mayr JA, Streubel B, Steiß JO, Hahn A, Csaicsich D, Castro C, Assoum M, **Müller T,** Wieczorek D, Mancini GM, Sadowski CE, Lévy N, Mégarbané A, Godbole K, Schanze D, Hildebrandt F, Delague V, Janecke AR, Zenker M. WDR73 Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. **Hum Mutat.** 2015 Nov;36(11):1021-8. doi:10.1002/humu.22828. PMID: 26123727.

24. **Müller T,** Rasool I, Heinz-Erian P, Mildenberger E, Hülstrunk C, Müller A, Michaud L, Koot BG, Ballauff A, Vodopiutz J, Rosipal S, Petersen BS, Franke A, Fuchs I, Witt H, Zoller H, Janecke AR, Visweswariah SS. Congenital secretory diarrhoea caused by activating germline mutations in GUCY2C. **Gut.** 2016 Aug;65(8):1306-13. doi: 10.1136/gutjnl-2015-309441. PMID: 25994218.

25. Wiegerinck CL, Janecke AR, Schneeberger K, Vogel GF, van Haaften-Visser DY, Escher JC, Adam R, Thöni CE, Pfaller K, Jordan AJ, Weis CA, Nijman IJ, Monroe GR, van Hasselt PM, Cutz E, Klumperman J, Clevers H, Nieuwenhuis EE, Houwen RH, van Haaften G, Hess MW, Huber LA, Stapelbroek JM, **Müller T**, Middendorp S. Loss of syntaxin 3 causes variant microvillus inclusion disease. Gastroenterology. 2014 Jul;147(1):65-68.e10. doi: 10.1053/j.gastro.2014.04.002. PMID: 24726755.

26. Thoeni CE, Vogel GF, Tancevski I, Geley S, Lechner S, Pfaller K, Hess MW, **Müller T**, Janecke AR, Avitzur Y, Muise A, Cutz E, Huber LA. Microvillus inclusion disease: loss of Myosin vb disrupts intracellular traffic and cell polarity. **Traffic**. 2014 Jan;15(1):22-42. doi: 10.1111/tra.12131. PMID: 24138727.

27. Witt H, Beer S, Rosendahl J, Chen JM, Chandak GR, Masamune A, Bence M, Szmola R, Oracz G, Macek M Jr, Bhatia E, Steigenberger S, Lasher D, Bühler F, Delaporte C, Tebbing J, Ludwig M, Pilsak C, Saum K, Bugert P, Masson E, Paliwal S, Bhaskar S, Sobczynska-Tomaszewska A, Bak D, Balascak I, Choudhuri G, Nageshwar Reddy D, Rao GV, Thomas V, Kume K, Nakano E, Kakuta Y, Shimosegawa T, Durko L, Szabó A, Schnúr A, Hegyi P, Rakonczay Z Jr, Pfützer R, Schneider A, Groneberg DA, Braun M, Schmidt H, Witt U, Friess H, Algül H, Landt O, Schuelke M, Krüger R, Wiedenmann B, Schmidt F, Zimmer KP, Kovacs P, Stumvoll M, Blüher M, **Müller T**, Janecke A, Teich N, Grützmann R, Schulz HU, Mössner J, Keim V, Löhr M, Férec C, Sahin-Tóth M. Variants in CPA1 are strongly associated with early onset chronic pancreatitis. **Nat Genet.** 2013 Oct;45(10):1216-20. doi: 10.1038/ng.2730. PMID: 23955596.

28. **Müller T**, Mizumoto S, Suresh I, Komatsu Y, Vodopiutz J, Dundar M, Straub V, Lingenhel A, Melmer A, Lechner S, Zschocke J, Sugahara K, Janecke AR. Loss of dermatan sulfate epimerase (DSE) function results in musculocontractural Ehlers-Danlos syndrome. **Hum Mol Genet.** 2013 Sep 15;22(18):3761-72. doi: 10.1093/hmg/ddt227. PMID: 23704329.

29. Vodopiutz J, Zoller H, Fenwick AL, Arnhold R, Schmid M, Prayer D, **Müller T**, Repa A, Pollak A, Aufricht C, Wilkie AO, Janecke AR. Homozygous SALL1 mutation causes a novel multiple congenital anomaly-mental retardation syndrome. **J Pediatr.** 2013 Mar;162(3):612-7. doi: 10.1016/j.jpeds.2012.08.042. PMID: 23069192.