Last Updated July 2022

Publications in Scientific Journals (Peer Reviewed)

1. Muenzer J, Vijayaraghavan S, Stein M, Kearney S, Wu Y, Alexanderian D. Long-term open-label phase I/II extension study of intrathecal idursulfase-IT in the treatment of neuronopathic mucopolysaccharidosis II. Genet Med. 2022 Jul;24(7):1437-1448.
2. Harmatz P, Muenzer J, Ezgü F, Dalén P, Huledal G, Lindqvist D, Gelius SS, Wikén M, Önnestam K, Bröijersén A. Mol Genet Metab. 2022 Jun 28;136(4):249-259. Chemically modified recombinant human sulfamidase (SOBI003) in mucopolysaccharidosis IIIA patients: Results from an open, non-controlled, multicenter study.
3. Sherwood DJ, Adams MC, Mazzella AJ, Abid A, Prasada S, Muenzer J, Johnson SM, Yeung M. Mucopolysaccharidosis Type I Diagnosed by Aortic and Mitral Valve Replacement. JACC Case Rep. 2021 Dec 15;3(18):1891-1894. PMID: 34984346
4. Muenzer J, Botha J, Harmatz P, Giugliani R, Kampmann C, Burton BK Evaluation of the long-term treatment effects of intravenous idursulfase in patients with mucopolysaccharidosis II (MPS II) using statistical modeling: data from the Hunter Outcome Survey (HOS). Orphanet J Rare Dis. 2021 Oct 30;16(1):456. doi: 10.1186/s13023-021-02052-4. PMID: 34717704
5. Elstein D, Giugliani R, Muenzer J, Schenk J, Schwartz IVD, Anagnostopoulou C. Impact of the COVID-19 pandemic on the standard of care for patients with lysosomal storage diseases: A survey of healthcare professionals in the Fabry, Gaucher, and Hunter Outcome Survey registries. Mol Genet Metab Rep. 2021 Sep;28:100788. doi: 10.1016/j.ymgmr.2021.100788. Epub 2021 Aug 4 PMID: 34367919
6. Wijburg FA, Whitley CB, Muenzer J, Gasperini S, Del Toro M, Muschol N, Cleary M, Sevin C, Shapiro E, Alexanderian D. A multicenter open-label extension study of intrathecal heparan-N-sulfatase in patients with Sanfilippo syndrome type A. Mol Genet Metab. 2021 Jul 7:S1096-7192(21)00743-5. PMID: 34247932
7. Mehta A, Ramaswami U, Muenzer J, Giugliani R, Ullrich K, Collin-Histed T, Panahloo Z, Wellhoefer H, Frader J. A charitable access program for patients with lysosomal storage disorders in underserved communities worldwide. Orphanet J Rare Dis. 2021 Jan 6;16(1):8. doi: 10.1186/s13023-020-01645-9. PMID: 33407729
8. Giugliani R, Muschol N, Keenan HA, Dant M, Muenzer J. Improvement in time to treatment, but not time to diagnosis, in patients with mucopolysaccharidosis type I. Arch Dis Child. 2020 Nov 2:archdischild-2020-319040. doi: 10.1136/archdischild-2020-319040. PMID: 33139350
9. van der Lee JH, Morton J, Adams HR, Clarke L, Eisengart JB, Escolar ML, Giugliani R, Harmatz P, Hogan M, Kearney S, Muenzer J, Muschol N, Rust S, Saville BR, Semrud-Clikeman M, Wang R, Shapiro E. Therapy development for the mucopolysaccharidoses: Updated consensus recommendations for neuropsychological endpoints. Mol Genet Metab. 2020 Sep-Oct;131(1-2):181-196. PMID: 32917509
10. Marianne S Muhlebach, Wei Sha, Beth MacIntosh, Thomas J Kelley, Joseph Muenzer. Metabonomics reveals altered metabolites related to inflammation and energy utilization at recovery of cystic fibrosis lung exacerbation. Metabol Open. 2019 3:100010 PMID: 32812947
11. Eisengart JB, King KE, Shapiro EG, Whitley CB, Muenzer J. The nature and impact of neurobehavioral symptoms in neuronopathic Hunter syndrome. Mol Genet Metab Rep. 2019 Dec 20;22:100549. PMID: 32055445
12. Viskochil D, Clarke LA, Bay L, Keenan H, Muenzer J, Guffon N. Growth patterns for untreated individuals with MPS I: Report from the international MPS I registry. Am J Med Genet A. 2019 Dec;179(12):2425-2432. PMID: 31639289
13. Clarke LA, Giugliani R, Guffon N, Jones SA, Keenan HA, Munoz-Rojas MV, Okuyama T, Viskochil D, Whitley CB, Wijburg FA, Muenzer J. Genotype-phenotype relationships in

mucopolysaccharidosis type I (MPS I): Insights from the International MPS I Registry. Clin Genet. 2019 Clin Genet. 2019 Oct;96(4):281-289. PMID: 31194252
14. Taylor JL, Clinard K, Powell CM, Rehder C, Young SP, Bali D, Beckloff SE, Gehtland LM, Kemper AR, Lee S, Millington D, Patel HS, Shone SM, Woodell C, Zimmerman SJ, Bailey DB Jr, Muenzer J. The North Carolina Experience with Mucopolysaccharidosis Type I Newborn Screening. J Pediatr. 2019 Aug;211:193-200. PMID: 31133280
15. Akyol MU, Alden TD, Amartino H, Ashworth J, Belani K, Berger KI, Borgo A, Braunlin E, Eto Y, Gold JI, Jester A, Jones SA, Karsli C, Mackenzie W, Marinho DR, McFadyen A, McGill J, Mitchell JJ, Muenzer J, Okuyama T, Orchard PJ, Stevens B, Thomas S, Walker R, Wynn R, Giugliani R, Harmatz P, Hendriksz C, Scarpa M; MPS Consensus Programme Steering Committee; MPS Consensus Programme Co-Chairs. Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. Orphanet J Rare Dis. 2019 Jun 13;14(1):137. doi: 10.1186/s13023-019-1074-9.
16. Akyol MU, Alden TD, Amartino H, Ashworth J, Belani K, Berger KI, Borgo A, Braunlin E, Eto Y, Gold JI, Jester A, Jones SA, Karsli C, Mackenzie W, Marinho DR, McFadyen A, McGill J, Mitchell JJ, Muenzer J, Okuyama T, Orchard PJ, Stevens B, Thomas S, Walker R, Wynn R, Giugliani R, Harmatz P, Hendriksz C, Scarpa M; MPS Consensus Programme Steering Committee; MPS Consensus Programme Co-Chairs. Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. Orphanet J Rare Dis. 2019 May 29;14(1):118.
17. Taylor JL, Clinard K, Powell CM, Rehder C, Young SP, Bali D, Beckloff SE, Gehtland LM, Kemper AR, Lee S, Millington D, Patel HS, Shone SM, Woodell C, Zimmerman SJ, Bailey DB Jr, Muenzer J. The North Carolina Experience with Mucopolysaccharidosis Type I Newborn Screening. J Pediatr. 2019 May 24.
18. Wijburg FA, Whitley CB, Muenzer J, Gasperini S, Del Toro M, Muschol N, Cleary M, Sevin C, Shapiro E, Bhargava P, Kerr D, Alexanderian D. Intrathecal heparan-N-sulfatase in patients with Sanfilippo syndrome type A: A phase IIb randomized trial. Mol Genet Metab. 2019 Feb;126(2):121-130
19. Fu H, Zaraspe K, Murakami N, Meadows AS, Pineda RJ, McCarty DM, Muenzer J. Targeting Root Cause by Systemic scAAV9-hIDS Gene Delivery: Functional Correction and Reversal of Severe MPS II in Mice. Mol Ther Methods Clin Dev. 2018 Sep 4;10:327-340.
20. Muenzer J, Giugliani R, Scarpa M, Tylki-Szymańska A, Jego V, Beck M. Clinical outcomes in idursulfase-treated patients with mucopolysaccharidosis type II: 3-year data from the hunter outcome survey (HOS). Orphanet J Rare Dis. 2017 Oct 3;12(1):161.
21. Viskochil D, Muenzer J, Guffon N, Garin C, Munoz-Rojas MV, Moy KA, Hutchinson DT. Carpal tunnel syndrome in mucopolysaccharidosis I: a registry-based cohort study. Dev Med Child Neurol. 2017 Dec;59(12):1269-1275.
22. Couser NL, Marchuk DS, Smith LD, Arreola A, Kaiser-Rogers KA, Muenzer J, Pandya A, Gucsavas-Calikoglu M, Powell CM. Co-occurring Down syndrome and SUCLA2-related mitochondrial depletion syndrome. Am J Med Genet A. Am J Med Genet A. 2017 Oct;173(10):2720-2724.
23. van der Lee JH, Morton J, Adams HR, Clarke L, Ebbink BJ, Escolar ML, Giugliani R, Harmatz P, Hogan M, Jones S, Kearney S, Muenzer J, Rust S, Semrud-Clikeman M, Wijburg FA, Yu ZF, Janzen D, Shapiro E. Cognitive endpoints for therapy development for neuronopathic mucopolysaccharidoses: Results of a consensus procedure. Mol Genet Metab. 2017 Jun;121(2):70-79. PMID: 28501294
24. Muenzer J, Jones SA, Tylki-Szymańska A, Harmatz P, Mendelsohn NJ, Guffon N, Giugliani R, Burton BK, Scarpa M, Beck M, Jangelind Y, Hernberg-Stahl E, Larsen MP, Pulles T, Whiteman DAH. Ten years of the Hunter Outcome Survey (HOS): insights, achievements, and lessons learned from a global patient registry. Orphanet J Rare Dis. 2017 May 2;12(1):82. PMID: 28464912

25. Clarke LA, Atherton AM, Burton BK, Day-Salvatore DL, Kaplan P, Leslie ND, Scott CR, Stockton DW, Thomas JA, Muenzer J. Mucopolysaccharidosis Type I Newborn Screening: Best Practices for Diagnosis and Management. J Pediatr. 2017 Mar;182:363-370.
26. Wooten WI 3rd, Muhlebach MS, Muenzer J, Loughlin CE, Vaughn BV. Progression of Polysomnographic Abnormalities in Mucolipidosis II (I-Cell Disease). J Clin Sleep Med. 2016 Dec 15;12(12):1695-1696.
27. Pupavac M, Watkins D, Petrella F, Fahiminiya S, Janer A, Cheung W, Gingras AC, Pastinen T, Muenzer J, Majewski J, Shoubridge EA, Rosenblatt DS. Inborn Error of Cobalamin Metabolism Associated with the Intracellular Accumulation of Transcobalamin-Bound Cobalamin and Mutations in ZNF143, Which Codes for a Transcriptional Activator. Hum Mutat. 2016 Sep;37(9):976-82.PMID: 27349184.
28. Couser NL, McClure J, Evans MW, Haines NR, Burden SK, Muenzer J. Homocysteinemia due to MTHFR deficiency in a young adult presenting with bilateral lens subluxations. Ophthalmic Genet. 2016 Apr 4:1-4. PMID: 27046515.
29. Fan Z, Kocis K, Valley R, Howard J Jr, Chopra M, Chen Y, An H, Lin W, Muenzer J, Powers W. High-pressure Transvenous Perfusion of the Upper Extremity in Human Muscular Dystrophy: A Safety Study with 0.9% Saline. Hum Gene Ther. 2015 Sep;26(9):614-21. doi: 10.1089/hum.2015.023. Epub 2015 Jul 30. PMID: 25953425.
30. Muenzer J, Hendriksz CJ, Fan Z, Vijayaraghavan S, Perry V, Santra S, Solanki GA, Mascelli MA, Pan L, Wang N, Sciarappa K, Barbier AJ. A phase I/II study of intrathecal idursulfase-IT in children with severe mucopolysaccharidosis II. Genet Med. 2016 Jan;18(1):73-81. doi: 10.1038/gim.2015.36. Epub 2015 Apr 2.
31. Wiklund I, Raluy-Callado M, Chen WH, Muenzer J, Fang J, Whiteman D. The Hunter Syndrome-Functional Outcomes for Clinical Understanding Scale (HS-FOCUS) Questionnaire: item reduction and further validation. Qual Life Res. 2014 Nov; 23(9):2457-62.
32. Beck M, Arn P, Giugliani R, Muenzer J, Okuyama T, Taylor J, Fallet S. The natural history of MPS I: global perspectives from the MPS I Registry. Genet Med. 2014 Oct;16(10):759-65.
33. Lampe C, Atherton A, Burton BK, Descartes M, Giugliani R, Horovitz DD, Kyosen SO, Magalhães TS, Martins AM, Mendelsohn NJ, Muenzer J, Smith LD. Enzyme Replacement Therapy in Mucopolysaccharidosis II Patients Under 1 Year of Age. JIMD Rep. 2014;14:99-113.
34. Muenzer J. Early initiation of enzyme replacement therapy for the mucopolysaccharidoses. Mol Genet Metab. 2014 Feb;111(2):63-72.
35. Chen Y, Fan Z, Ji S, Muenzer J, An H, Lin W. Patient-specific biomechanical modeling of ventricular enlargement in hydrocephalus from longitudinal magnetic resonance imaging. Med Image Comput Comput Assist Interv. 2013;16(Pt 3):291-8.
36. Wooten WI 3rd, Muenzer J, Vaughn BV, Muhlebach MS. Relationship of sleep to pulmonary function in mucopolysaccharidosis II. J Pediatr. 2013 Jun;162(6):1210-5.
37. Muhlebach MS, Shaffer CB, Georges L, Abode K, Muenzer J. Bronchoscopy and airway management in patients with mucopolysaccharidoses (MPS). Pediatr Pulmonol. 2013 Jun;48(6):601-7.
38. Muenzer J, Bodamer O, Burton B, Clarke L, Frenking GS, Giugliani R, Jones S, Rojas MV, Scarpa M, Beck M, Harmatz P. The role of enzyme replacement therapy in severe Hunter syndrome-an expert panel consensus. Eur J Pediatr. 2012 Jan;171(1):181-8. Epub 2011 Oct 29.
39. Muenzer J. Overview of the Mucopolysaccharidoses. Rheumatology (Oxford). 2011 Dec; 50 Suppl 5:v4-v12.
40. Fan Z, Kocis K, Valley R, Howard JF, Chopra M, An H, Lin W, Muenzer J, Powers W. Safety and Feasibility of High-pressure Transvenous Limb Perfusion With 0.9% Saline in Human Muscular Dystrophy. Mol Ther. 2012 Feb;20(2):456-61. doi: 10.1038/mt.2011.137. Epub 2011 Jul 19.
41. Muhlebach MS, Wooten W, Muenzer J. Respiratory manifestations in mucopolysaccharidoses. Paediatr Respir Rev. 2011 Jun;12(2):133-8. Epub 2010 Nov 26.

42. Bien Lai, Joseph Muenzer and Michael W. Roberts. Idiopathic Gingival Hyperplasia: A Case Report with a 17-Year Followup. Case Reports in DentistryVolume 2011 (2011), Article ID 986237, 5 pagesdoi:10.1155/2011/986237.
43. Muenzer J, Beck M, Giugliani R, Suzuki Y, Tylki-Szymanska A, Valayannopoulos V, Vellodi A, Wraith JE. Idursulfase treatment of Hunter syndrome in children younger than 6 years: results from the Hunter Outcome Survey. Genet Med. 2011 Feb;13(2):102-9.
44. Muenzer J, Beck M, Eng CM, Giugliani R, Harmatz P, Martin R, Ramaswami U, Vellodi A, Wraith JE, Cleary M, Gucsavas-Calikoglu M, Puga AC, Shinawi M, Ulbrich B, Vijayaraghavan S, Wendt S, Conway AM, Rossi A, Whiteman DA, Kimura A. Long-term, open-labeled extension study of idursulfase in the treatment of Hunter syndrome. Genet Med. 2011 Feb;13(2):95-101.
45. Fu H, DiRosario J, Kang L, Muenzer J, McCarty DM. Restoration of central nervous system alpha-N-acetylglucosaminidase activity and therapeutic benefits in mucopolysaccharidosis IIIB mice by a single intracisternal recombinant adeno-associated viral type 2 vector delivery. J Gene Med. 2010 Jul;12(7):624-33.
46. Fan Z, Styner M, Muenzer J, Poe M, Escolar M. Correlation of Automated Volumetric Analysis of Brain MR Imaging with Cognitive Impairment in a Natural History Study of Mucopolysaccharidosis II. Am J Neuroradiol. 2010 Aug;31(7):1319-23. Epub 2010 Mar 4
47. Beck M, Muenzer J, Scarpa M. Evaluation of disease severity in mucopolysaccharidoses. J Pediatr Rehabil Med. 2010;3(1):39-46.
48. Muenzer J, Beck M, Eng CM, Escolar ML, Giugliani R, Guffon NH, Harmatz P, Kamin W, Kampmann C, Koseoglu ST, Link B, Martin RA, Molter DW, Muñoz Rojas MV, Ogilvie JW, Parini R, Ramaswami U, Scarpa M, Schwartz IV, Wood RE, Wraith E. Multidisciplinary management of Hunter syndrome. Pediatrics. 2009 Dec;124(6):e1228-39.
49. McCarty DM, DiRosario J, Gulaid K, Muenzer J, Fu H. Mannitol-facilitated CNS entry of rAAV2 vector significantly delayed the neurological disease progression in MPS IIIB mice. Gene Ther. 2009 Nov;16(11):1340-52.
50. Surekha P and Muenzer J. The Diagnosis and Management of a Female With Mild Citrullinemia and Undetectable Argininosuccinate Synthetase Activity in Fibroblasts. Topics in Clinical Nutrition, 24:366:373.
51. Clarke LA, Wraith JE, Beck M, Kolodny EH, Pastores GM, Muenzer J, Rapoport DM, Berger KI, Sidman M, Kakkis ED, Cox GF. Long-term efficacy and safety of laronidase in the treatment of mucopolysaccharidosis I. Pediatrics. 2009 Jan;123(1):229-40.
52. Muenzer J, Wraith JE, Clarke LA; International Consensus Panel on Management and Treatment of Mucopolysaccharidosis I. Mucopolysaccharidosis I: management and treatment guidelines. Pediatrics. 2009 Jan;123(1):19-29.
53. Wraith JE, Beck M, Giugliani R, Clarke J, Martin R, Muenzer J; HOS Investigators. Initial report from the Hunter Outcome Survey. Genet Med. 2008;10(7):508-16.
54. Muenzer J, Martins AM. Hunter syndrome: to treat or not to treat. Acta Paediatr Suppl. 2008; 97(457):55-6
55. Martin R, Beck M, Eng C, Giugliani R, Harmatz P, Muñoz V, Muenzer J. Recognition and diagnosis of mucopolysaccharidosis II (Hunter syndrome). Pediatrics. 2008 Feb;121(2):377-86.
56. Whitley CB, Barranger JA, Eng CM, Davidson B, Grabowski GA, Muenzer J, Pastores GM, Patel SK, Shapiro EG, Steiner RD, Walkley SU, Wedehase BA, Wilcox WR. Lysosomal Disease Network's "WORLD Symposium 2008". Mol Genet Metab. 2008 Feb;93(2):S3-5.
57. Garcia AR, Pan J, Lamsa JC, Muenzer J. The characterization of a murine model of mucopolysaccharidosis II (Hunter syndrome). Inherit Metab Dis. 2007 30(6):924-34.
58. Fu H, Kang L, Jennings JS, Moy SS, Perez A, Dirosario J, McCarty DM, Muenzer J. Significantly increased lifespan and improved behavioral performances by rAAV gene delivery in adult mucopolysaccharidosis IIIB mice. Gene Ther. 2007 Jul;14(14):1065-77

59. Garcia AR, DaCosta JM, Pan J, Muenzer J, Lamsa JC. Preclinical dose ranging studies for enzyme replacement therapy with idursulfase in a knock-out mouse model of MPS II. Mol Genet Metab. 2007 Jun;91(2):183-90.
60. Pastores GM, Arn P, Beck M, Clarke JT, Guffon N, Kaplan P, Muenzer J, Norato DY, Shapiro E, Thomas J, Viskochil D, Wraith JE. The MPS I registry: design, methodology, and early findings of a global disease registry for monitoring patients with Mucopolysaccharidosis Type I. Mol Genet Metab. 2007 May;91(1):37-47.
61. Muenzer J, Gucsavas-Calikoglu M, McCandless SE, Schuetz TJ, Kimura A. A phase I/II clinical trial of enzyme replacement therapy in mucopolysaccharidosis II (Hunter syndrome). Mol Genet Metab. 2007 Mar;90(3):329-37.
62. Monte S. Willis, Alice A. Basinger, Zheng Fan, Surekha Pendyal, Joseph Muenzer, Catherine Hammett-Stabler. Hepatosplenomegaly in an 8 Month-Old Child. Laboratory Medicine. 2006 Nov, 37:665-695.
63. Muenzer J, Wraith JE, Beck M, Giugliani R, Harmatz P, Eng CM, Vellodi A, Martin R, Ramaswami U, Gucsavas-Calikoglu M, Vijayaraghavan S, Wendt S, Puga A, Ulbrich B, Shinawi M, Cleary M, Piper D, Conway AM, Kimura A. A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome). Genet Med. 2006 Aug; 8 (8):465-473.
64. Frazier DM, Millington DS, McCandless SE, Koeber DD, Weavil SD, Chaing SH, Muenzer J. The tandem mass spectrometry newborn screening experience in North Carolina: 1997-2005. J Inherit Metab Dis. 2006 Feb; 29:76-85.
65. Basinger AA, Booker JK, Frazier DM, Koeberl DD, Sullivan JA, Muenzer J. Glutaric acidemia type 1 in patients of Lumbee heritage from North Carolina. Mol Genet Metab. 2006 May;88:90-2.
66. Friso A, Tomanin R, Alba S, Gasparotto N, Puicher EP, Fusco M, Hortelano G, Muenzer J, Marin O, Zacchello F, Scarpa M. Reduction of GAG storage in MPS II mouse model following implantation of encapsulated recombinant myoblasts. J Gene Med. 2005 Nov; 7:1482-91.
67. Matheus MG, Castillo M, Smith JK, Armao D, Towle D, Muenzer J. Brain MRI findings in patients with mucopolysaccharidosis types I and II and mild clinical presentation. Neuroradiology. 2004 Aug; 46(8):666-72.
68. Muenzer J, Fisher A. Advances in the treatment of mucopolysaccharidosis type I. N Engl J Med. 2004 May 6; 350(19):1932-4.
69. Wraith JE, Clarke LA, Beck M, Kolodny EH, Pastores GM, Muenzer J, Rapoport DM, Berger KI, Swiedler SJ, Kakkis ED, Braakman T, Chadbourne E, Walton-Bowen K, Cox GF. Enzyme replacement therapy for mucopolysaccharidosis I: a randomized, double-blinded, placebo-controlled, multinational study of recombinant human alpha-L-iduronidase (laronidase). J Pediatr. 2004 May;144(5):581-8.
70. Muenzer, J. The mucopolysaccharidoses: a heterogeneous group of disorders with variable pediatric presentations. J Pediatr. 2004 May;144(5 Suppl):S27-34.
71. Fu H, Muenzer J, Samulski RJ, Breese G, Sifford J, Zeng X, McCarty DM. Self-complementary adeno-associated virus serotype 2 vector: global distribution and broad dispersion of AAV-mediated transgene expression in mouse brain. Mol Ther. 2003 Dec;8(6):911-7.
72. Koeberl DD, Millington DS, Smith WE, Weavil SD, Muenzer J, McCandless SE, Kishnani PS, McDonald MT, Chaing S, Boney A, Moore E, Frazier DM. Evaluation of 3-methylcrotonyl-CoA carboxylase deficiency detected by tandem mass spectrometry newborn screening. J Inherit Metab Dis. 2003;26(1):25-35.
73. Muenzer J, Lamsa JC, Garcia A, Dacosta J, Garcia J, Treco DA. Enzyme replacement therapy in mucopolysaccharidosis type II (Hunter syndrome): a preliminary report. Acta Paediatr Suppl. 2002; 91(439):98-9.
74. Mareska MC, Adams KK, Muenzer J, Braun TG, Howard JF: Late onset presentation of glutaric acidemia type II with myopathy. Journal of Clinical Neuromuscular Disease 2003, 4:124-8.

75. Fu, H, Samulski, RJ, McCown, TJ, Picornel, YJ, Fletcher, D and Muenzer, J. Neurological correction of lysosomal storage in a mucopolysaccharidosis IIIB mouse model by adeno-associated virus-mediated gene delivery. Molecular Therapy 2002, 5:42-9.
76. Kakkis ED. Muenzer J. Tiller GE. Waber L. Belmont J. Passage M. Izykowski B. Phillips J. Doroshow R. Walot I. Hoft R. Neufeld EF. Enzyme-replacement therapy in mucopolysaccharidosis I. New England Journal of Medicine. 344(3):182-8, 2001.
77. Andresen BS. Dobrowolski SF. O'Reilly L. Muenzer J. McCandless SE. Frazier DM. Udvari S. Bross P. Knudsen I. Banas R. Chace DH. Engel P. Naylor EW. Gregersen N. Medium-chain acyl-CoA dehydrogenase (MCAD) mutations identified by MS/MS-based prospective screening of newborns differ from those observed in patients with clinical symptoms: identification and characterization of a new, prevalent mutation that results in mild MCAD deficiency. American Journal of Human Genetics. 68(6):1408-18, 2001.
78. Weston, BW, Lin, J, Muenzer, J, et al. Glucose-6-phosphate mutation G188R confers an atypical glycogen storage disease type 1b phenotype. Pediatric Research 2000, 48: 329-34.
79. Reitnauer, PJ, Chaing, S and Muenzer, J. Why do critically ill newborns not get mandated screening? North Carolina Medical Journal 1999, 60: 256-58.
80. Van Hove, JL, Kishnani, P, Muenzer, et al. Benzoate therapy and carnitine deficiency in non-ketotic hypergycinemia. Am. J. Medical Genetics 1995, 59:444-53.
81. Marowitz, AJ, Chen, YT, Muenzer, J, Delbunono, EA, and Lucey, MR. A man with Type III glycogenosis associated with cirrhosis and portal hypertension. Gastroenterology 1993, 105: 1882-5.
82. Muenzer, J., Beekman, R.H., Profera, L.M., and Bove, E.L. Severe mitral insufficiency in mucopolysaccharidosis type III-B (Sanfilippo syndrome). Pediatr Cardiol 14:130-132, 1993.
83. Muenzer, J., Neufeld, E.F., Contanopolus, G.G. et al. Attempted enzyme replacement using human amnion membrane implantations in mucopolysaccharidosis. J Inher. Metab. Dis. 1992, 15:25-37.
84. Abrams, S.A., Sidbury, J.B., Muenzer, J., Esteban, N.V., Vieira, N.E., and Yergey, A.L. Stable isotopic measurement of endogenous fecal calcium excretion in children. J Pediatr Gastroenterol Nutr 1991, 12:469-473.
85. Robertson, PL., Buchanan, DN., and Muenzer, J. 5-Oxoprolinuria in an adolescent with chronic metabolic acidosis, mental retardation, and psychosis. J. Pediatr 1991, 118:92-5.
86. Buchanan, D.N., Muenzer, J., and Thoene, J.G. Positive ion thermospray liquid chromatography/mass spectrometry: Detection of organic acidurias. J Chromatogr/Biomedical Applications 1990, 534:1-11.
87. Bliziotes, M., Yergey, A., Nanes, M., Muenzer, J., Begley, M., Vieira, N., Kher, K., Brandi, M.L., and Marx, S. Absent intestinal response to 1,25 dihydroxyvitamin Documentation in vivo and in vitro and effective therapy with high dose intravenous calcium infusions. J Clin Endocrinol Metab 1988, 66:294-300.
88. Caruso, R.C., Kaiser-Kupfer, M.I., Muenzer, J., Ludwig, I.H., Zasloff, M.A., and Mercer, P.A. Electroretinographic Findings in the Mucopolysaccharidosis. Ophthalmology 1986, 93:1612-6.
89. Muenzer, J., Bildstein, C., Gleason, M., and Carlson, D.M. Properties of proline-rich proteins from parotid gland of isoproterenol-treated rats. J. Biol. Chem. 1979; 254: 5629-34.
90. Muenzer, J., Bildstein, C., Gleason, M., and Carlson, D.M. Purification of proline-rich proteins from parotid gland of isoproterenol-treated rats. J. Biol. Chem. 1979; 254: 5623-8.
91. Muenzer, J., Weinbach, E.C., and Wolfe, S.M. Oxygen consumption of human blood platelets. II. Effects of inhibitors. B.B.A. 1975; 376: 243-8.
92. Muenzer, J., Weinbach, E.C., and Wolfe, S.M. Oxygen consumption of human blood platelets. I. Effects of Thrombin. B.B.A. 1975; 376: 237-42.
93. Roth, J., Prout, T.E., Goldfine, I.D., Wolfe, S.M., Muenzer, J., Grauer, L.E., and Marcus, M.L. Sulfonylureas: Effects in Vivo and in Vitro. Ann. Intern. Med. 1971; 75: 607-21.

94. Gordon, R.S., Thompson, R.H., Muenzer, J., and Thrasher, D. Sweat lactate in man is derived from blood glucose. J. Appl.Physiol. 1971; 31: 713-6.