Rare Diseases Clinical Research Network (RDCRN) Publications

Updated: June 5, 2019

RDCRN Data Management and Coordinating Center Publications

DMCC Publications

RDCRN Descriptive Publications

RDCRN Publications

RDCRN Publications, Current Grant Cycle Consortia

Advancing Research & Treatment for Frontotemporal Lobar Degeneration
Autonomic Rare Disease Clinical Research Consortium
Brain Vascular Malformations Consortium
Brittle Bone Disorders Consortium
Clinical Research in ALS & Related Disorders for Therapeutic Development
Consortium of Eosinophilic Gastrointestinal Disease Researchers
Developmental Synaptopathies Consortium
Dystonia Coalition
Genetic Disorders of Mucociliary Clearance
Inherited Neuropathies Consortium
Lysosomal Disease Network
Nephrotic Syndrome Rare Disease Clinical Research Network
North American Mitochondrial Diseases Consortium
Porphyrias Consortium
Primary Immune Deficiency Treatment Consortium
Rare Kidney Stone Consortium
Rare Lung Diseases Consortium
Rett Syndrome, MECP2 Duplications, and Rett-related Disorders Consortium
Sterol and Isoprenoid Diseases Consortium
Urea Cycle Disorders Consortium
Vasculitis Clinical Research Consortium

Resources are listed by consortium and publication type and sorted by date. JAMA Style.
Send any updates to Cameron Hainline at Cameron.Hainline@epi.usf.edu

RDCRN Publications, Previous Grant Cycle Consortia

Angelman, Rett, and Prader-Willi Syndromes Consortium
Bone Marrow Failure Consortium
Cholestatic Liver Disease Consortium
Chronic Graft Versus Host Disease Consortium
Clinical Research Consortium for Spinocerebellar Ataxias
Consortium for Clinical Investigation of Neurologic Channelopathies
Rare Genetic Steroid Disorders
Rare Thrombotic Diseases
Salivary Gland Carcinomas Consortium

RDCRN Data Management and Coordinating Center Publications

Book Chapters

1. Krischer JP. Clinical trials for rare lung diseases. In: McCormack FX, Panos RJ, Trapnell BC, eds. *Molecular Basis of Pulmonary Disease*. 1st ed: Springer; 2010.
2. Richesson R, Vehik K. Patient Registries: Utility, Validity and Inference. In: (ed) IPMGS, ed. *Rare Diseases Epidemiology, 1st edn*: Springer, The Netherlands; 2010.
3. Richesson R, Andrews J. Clinical Research Informatics: Springer-Verlag:, London; 2010.

Abstracts Presented at Conferences & Conference Proceedings

1. Krischer J. Data collection and analysis from multiple research sites: The Rare Diseases Clinical Research Network. Paper presented at: International Conference on Rare Diseases & Orphan Drugs; February 16, 2005; Stockholm, Sweden.
2. Richesson R, Young K, Malloy J, Lloyd J, Krischer J. A contact registry for persons with rare diseases: a tool for recruiting and retaining participants in a clinical research network. Paper presented at: Inventory and Evaluation of Clinical Research Networks; May 31, 2006; Washington, DC.
3. Malloy J, Richesson R, Krischer J. The Adverse Event Management System for the Rare Disease Clinical Research Network. Paper presented at: Inventory and Evaluation of Clinical Research Networks; May 31, 2006; Washington, D.C.
4. Richesson R. Identifying and evaluating health data standards. Paper presented at: American Public Health Association; November 4-8, 2006; Boston, MA.
5. Richesson R, Young K, Malloy J, Guillette H, Krischer J. A contact registry for persons with rare diseases: a tool for recruiting and retaining. Paper presented at: Clinical Trials Expo, Annual Symposium of the American Medical Informatics Association; November 13, 2006; Washington, DC.
6. Krischer PJ. Clinical trial design issues and options for study of rare diseases. Presented at: RDCRN Conference on Clinical Research for Rare Diseases. 2007. Washington, DC.
7. Lee HS. Data presentation and study action item discussion. Urea Cycle Disease Consortium Meeting. Spring and Fall 2007, 2008.
8. Richesson RL, Young K, Lloyd J, Adams T, Guillette H, Malloy J, Krischer JP. An automated communication system in a Contact Registry for persons with rare diseases: tools for retaining potential clinical research participants. *AMIA. Annu. Symp. Proc.* 2007:1094. PMID: 18694191
9. Richesson R. Best practices for collaborative development of terminology and standards. Paper presented at: First Annual NHLBI Community Forum on Data Standards and Terminology; May 29-30, 2007; Bethesda, MD.
10. Richesson R, Smith S, Malloy J, Krischer J. Achieving standardized medication data in clinical research studies: two approaches and applications for implementing RxNorm. Paper presented at: AMIA (American Medical Informatics Association), Spring Congress; May 22-24, 2007; Orlando, FL.

11. Niland J, Dorr D, El Saadawi G, Embi P, Richesson R, Sim I, Martin R. Knowledge representation of eligibility criteria in clinical trials. Paper presented at: American Medical Informatics Association Annual Symposium; November, 2007; Chicago, IL.

12. Richesson R, Lee H, Cuthbertson D, Lloyd J, Young K, Krischer J. An automated contact registry for persons with rare diseases: scalable tools for identifying and communicating with clinical research participants. Paper presented at: International Conference on Rare Diseases and Orphan Drugs (ICORD); May 20-22, 2008; Washington, DC.

13. Jeffrey Krischer. The Rare Diseases Clinical Research Network's (RDCRN) Data Management and Coordination Center, J.P. Krischer, University of South Florida, Tampa, FL. Molecular Genetics and Metabolism. Volume 99, Issue 2, 2010, Page S24, ISSN 1096-7192, https://doi.org/10.1016/j.ymgme.2009.10.094. (http://www.sciencedirect.com/science/article/pii/S1096719209003783)

14. Krischer PJ. Clinical trial design issues and options for study of rare diseases. Presented at: RDCRN Conference on Clinical Research for Rare Diseases. 2010. Washington, DC.

15. Lee HS, Krischer JP, Young L and McCormack FX. (2011) Categorization of VEGF-D in response to sirolimus therapy or prognosis of disease progression: MILES Trial. Poster presentation at the LAM Foundation/ International Research Conference (First Prize in poster section), Cincinnati, OH. (http://www.thelamfoundation.org/images/pdfs/scientificprogram2011.pdf)

16. Lee HS, McCormack FX, Young L and Krischer JP. (2011) Regression approach to examine sirolimus effect on the correlation between FEV1 and VEGF-D: MILES Trial. Platform presentation at the International Society for Clinical Biostatistics (ISCB), Ottawa, CA. (http://www.iscb2011.info/00-welcome_e.shtml)

17. Oster-Granite ML, Parisi MA, Abbeduto L, Berlin DS, Bodine C, Bynum D, Capone G, Collier E, Hall D, Kaeser L, Kaufmann P, Krischer J, Livingston M, McCabe LL, Pace J, Pfenninger K, Rasmussen SA, Reeves RH, Rubinstein Y, Sherman S, Terry SF, Whitten MS, Williams S, McCabe ER, Maddox YT. Down syndrome: National conference on patient registries, research databases, and biobanks. *Mol Genet Metab.* 2011;104(1):13-22. PMID: 21835664, PMCID: PMC3171614

18. Clowse M, Richesson R, Pieper C, Merkel PA, Consortium VCR. Pregnancy in Men and Women with Vasculitis. Paper presented at: American College of Rheumatology Annual Scientific Meeting; November 5-9, 2011; Chicago, IL.

19. Clowse M, Richesson R, Pieper C, Merkel PA, Consortium VCR. Infertility Among Patients with Vasculitis. Paper presented at: American College of Rheumatology Annual Meeting; November 5-9, 2011; Chicago, IL.

20. Ashizawa T, Perlman S, Gomez C, Wilmot G, Schmahmann J, Ying S, Zesiewicz T, Paulson H, Shakkottai VG, Bushara K, Mazzoni P, Kuo S, Pulst S, Figueroa K, Xia G, Krischer J, Cuthbertson D, Holbert AR, Ferguson J, Galpern W, Subramony SH. Clinical Characteristics of Spinocerebellar Ataxias 1, 2, 3 and 6. Paper presented at: American Academy of Neurology 64th Annual Meeting; April 21-28, 2012; New Orleans, LA.

21. Diethelm-Okita B, Utz J, Eichler FS, Ziegler RS, Leduc RL, Whitley CB. A Natural History of Hexosaminidase Deficiency. Paper presented at: Molecular Genetics and Metabolism. 2012.

22. Leigh M, Shapiro AJ, Pittman JE, Davis SD, Lee H, Krischer J, Ferkol TW, Atkinson JJ, Sagel SD, Rosenfeld M, Dell SD, Milla C, Olivier KN, Knowles M. Definition of clinical criteria for diagnosis

of primary ciliary dyskinesia. Paper presented at: American Journal of Respiratory and Critical Care Medicine. 2012.

23. Richesson R, Shereff D, Sutphen R, Guillette H, Paulus K, Merkel PA, Clowse M, Harris J, Cuthbertson D, Leduc R, Krischer JP. Patient Registries to Support Research in Rare Diseases – Experience from the Rare Diseases Clinical Research Network. Paper presented at: Lysosomal Disease Network, 8th Annual World Symposium 2012; San Diego, CA.

24. Richesson R, Shereff D, Sutphen R, et al. Patient Registries to Support Research in Rare Diseases – Experience from the Rare Diseases Clinical Research Network. *Mol Genet Metab.* 2012;105(2):S54.

25. Guillette H, Shereff D, Andrews JE, Spisla CM, Albarracin N, Konicek D, Richesson RL. PRISM: A shared resource for global data standards in patient registries. Poster presented at the 2012 International Conference on Rare Diseases & Orphan Drugs. February 2012. Tokyo, Japan.

26. Graves T, Cha Y, Hahn A, Barohn RJ, Amato A, Griggs R, Bundy B, Jen JC, Baloh R, Hanna M. Episodic ataxia type 1: Characterization of the disease and its effect on quality of life. Paper presented at: American Academy of Neurology; April 21-28, 2012; New Orleans.

27. Graves T, Fialho D, Smith S, Cha Y, Amato A, Griggs R, Bundy B, Jen JC, Baloh R, Hanna M. EEG abnormalities in the episodic ataxias. Paper presented at: American Academy of Neurology; April 21-28, 2012; New Orleans, LA.

28. Richesson R, Shereff D, Lloyd J, Young K, Guillette H, Paulus K, Harris J, Cuthbertson D, Krischer J, Rare Diseases Clinical Research Network. RDCRN Contact Registry to Support Research in Rare Diseases. Poster presented at: Yale Center for Clinical Investigation. 5th Annual Clinical Research Management Workshop; June 4-5, 2012; New Haven, CT.

29. Grayson PC, Amudala NA, McAlear C, LeDuc RL, Shereff D, Richesson R, Fraenkel L, Merkel PA. Illness perceptions among patients with different forms of vasculitis. Paper presented at: American College of Rheumatology/Association of Rheumatology Health Professionals (ACR/ARHP) Annual Meeting; November 10-14, 2012; Washington, DC.

30. Grayson PC, Amudala NA, McAlear C, LeDuc RL, Shereff D, Richesson R, Fraenkel L, Merkel PA. Assessing fatigue in systemic vasculitis from the patient's perspective. Paper presented at: American College of Rheumatology/Association of Rheumatology Health Professionals (ACR/ARHP) Annual Meeting; November 10-14, 2012; Washington, DC.

31. Grayson PC, Amudala NA, McAlear C, LeDuc RL, Shereff D, Richesson R, Fraenkel L, Merkel PA. Causal beliefs of disease among patients with systemic vasculitis. Paper presented at: American College of Rheumatology/Association of Rheumatology Health Professionals (ACR/ARHP) Annual Meeting; November 10-14, 2012; Washington, DC.

32. Krischer PJ. Clinical trial design issues and options for study of rare diseases. Presented at: RDCRN Conference on Clinical Research for Rare Diseases. 2012. Washington, DC.

33. Hall CA, Bacon CJ, Shy ME, Inherited Neuropathies Consortium, Rare Diseases Clinical Research Network Data Management and Coordinating Center. The Rare Diseases Clinical Research Network Contact Registry for the Inherited Neuropathies Consortium. *Charcot-Marie-Tooth Association, 5th International CMT Consortium Meeting*. Antwerp, Belgium2013.

34. Leduc R, Hall C, Shereff D, Lloyd J, Young K, Guillette H, Harris J, Gandolfo L, Cuthbertson D, Krischer J. An Overview of the Rare Diseases Clinical Research Network. Paper presented at: Lysosomal Disease Network, 9th Annual WORLD Symposium; Feb. 12-15, 2013; Orlando, FL.

35. Lee HS. (2013, 2016). Poster presentation and rountable discussion. LAM Meeting.

36. Richesson R. Strategies to harmonize data collection: a perspective from the RDCRN Data Management & Coordinating Center. Paper presented at: Pediatric Academic Societies Annual Meeting; May 4-7, 2013; Washington, DC.

37. Lee HS. (2014). Protocol development and rountable discussion. LAM Biomarker Innovation Summit

38. Holbert A. Clinical Research Compliance in the Field: What really works? Paper presented at: Society of Research Administrators International Meeting; October 21. 2014; San Diego, CA.

39. Tamura RN. (2015). A sequential multiple assignment randomized Phase 2 trial for rare diseases. ASA Biopharamceutical Section Statistics Workshop. Washington DC

40. Krischer PJ. Clinical trial design issues and options for study of rare diseases. Presented at: RDCRN Conference on Clinical Research for Rare Diseases. 2016. Washington, DC.

41. Lee HS, (2016) A Phase II Clinical Trial of an Aromatase Inhibitor for Postmenopausal Women with Lymphangioleiomyomatosis. Poster presentation at the LAM Foundation/International Research Conference, Cincinnati, OH.

42. Tamura RN. (2016). A journey through clinical trials in the past 25 years. 75th Anniversary Conference of Department of Statistics. North Carolina State University. Raleigh, NC

43. Tamura RN. (2016). A small n sequential multiple assignment randomized trial design for use in rare disease research. Joint Statistical Meetings. Chicago, IL

44. Chen YC, Tamura RN. (2017). Innovative designs for rare disease studies. Duke-FDA Rare Disease Think Tank. Washington DC. and DIA Clinical Research Community Webcast.

45. Hainline C, Rizzo D, Shy ME, Inherited Neuropathies Consortium, Rare Diseases Clinical Research Network Data Management and Coordinating Center. Enhancements to the RDCRN Contact Registry for the Inherited Neuropathies Consortium. Poster presented at Peripheral Nerve Society Annual Meeting; Jul. 8-12, 2017; Sitges, Spain.

46. Tamura RN, Kidwell KM, Wei B, Braun TM. (2017). The use of small n sequential multiple assignment randomized trials (SnSMART) in rare disease research. Joint Statistical Meeting. Baltimore, MD.

47. Williams M, Krischer J. Enhancements to the Rare Diseases Clinical Research Network Contact Registry, Poster presented at: Lysosomal Disease Network, 14th Annual WORLD Symposium; Feb. 5-9, 2018; San Diego, CA. https://doi.org/10.1016/j.ymgme.2017.12.410

48. Tamura RN. SMART clinical trial design for large and small samples (Proposal submitted for Workshop in 2019). Society of Clinical Trials. New Orleans, LA

Journal Articles

1. Merkel PA, Seo P, Aries P, Neogi T, Villa-Forte A, Boers M, Cuthbertson D, Felson DT, Hellmich B, Hoffman GS, Jayne DR, Kallenberg CG, Krischer J, Mahr A, Matteson EL, Specks U, Luqmani R, Stone J. Current status of outcome measures in vasculitis: focus on Wegener's granulomatosis

and microscopic polyangiitis. Report from OMERACT 7. *J. Rheumatol.* Dec 2005;32(12):2488-2495. PMID: 16331794

2. Richesson R, Young K, Guillette H, Tuttle M, Abbondondolo M, Krischer J. Standard terminology on demand: facilitating distributed and real-time use of SNOMED CT during the clinical research process. *AMIA. Annu. Symp. Proc.* 2006:1076. PMID: 17238695, PMCID: PMC1839627

3. Richesson RL, Andrews JE, Krischer JP. Use of SNOMED CT to represent clinical research data: a semantic characterization of data items on case report forms in vasculitis research. *J. Am. Med. Inform. Assoc.* Sep-Oct 2006;13(5):536-546. PMID: 16799121, PMCID: PMC1561787

4. Richesson R, Syed A, Guillette H, Tuttle MS, Krischer J. A web-based SNOMED CT browser: distributed and real-time use of SNOMED CT during the clinical research process. *Stud. Health Technol. Inform.* 2007;129(Pt 1):631-635. PMID: 17911793

5. Andrews JE, Richesson RL, Krischer J. Variation of SNOMED CT coding of clinical research concepts among coding experts. *J. Am. Med. Inform. Assoc.* Jul-Aug 2007;14(4):497-506. PMID: 17460128, PMCID: PMC2244907

6. Richesson RL, Krischer J. Data standards in clinical research: gaps, overlaps, challenges and future directions. *J. Am. Med. Inform. Assoc.* Nov-Dec 2007;14(6):687-696. PMID: 17712081, PMCID: PMC2213488

7. Richesson RL, Malloy JF, Paulus K, Cuthbertson D, Krischer JP. An automated standardized system for managing adverse events in clinical research networks. *Drug Saf.* 2008;31(10):807-822. PMID: 18759506

8. Moyers S, Richesson R, Krischer J. Trans-Atlantic data harmonization in the classification of medicines and dietary supplements: a challenge for epidemiologic study and clinical research. *Int J Med Inform.* Jan 2008;77(1):58-67. PMID: 17289429, PMCID: PMC2259273

9. Tuchman M, Lee B, Lichter-Konecki U, Summar ML, Yudkoff M, Cederbaum SD, Kerr DS, Diaz GA, Seashore MR, Lee HS, McCarter RJ, Krischer JP, Batshaw ML. Cross-sectional multicenter study of patients with urea cycle disorders in the United States. *Mol. Genet. Metab.* Aug 2008;94(4):397-402. PMID: 18562231, PMCID: PMC2640937

10. Richesson RL, Fung KW, Krischer JP. Heterogeneous but "standard" coding systems for adverse events: Issues in achieving interoperability between apples and oranges. *Contemp. Clin. Trials.* Sep 2008;29(5):635-645. PMID: 18406213, PMCID: PMC2575842

11. Andrews JE, Patrick TB, Richesson RL, Brown H, Krischer JP. Comparing heterogeneous SNOMED CT coding of clinical research concepts by examining normalized expressions. *J. Biomed. Inform.* Dec 2008;41(6):1062-1069. PMID: 18328789, PMCID: PMC2605270

12. Richesson RL, Lee HS, Cuthbertson D, Lloyd J, Young K, Krischer JP. An automated communication system in a contact registry for persons with rare diseases: scalable tools for identifying and recruiting clinical research participants. *Contemp. Clin. Trials.* Jan 2009;30(1):55-62. PMID: 18804556, PMCID: PMC2640948

13. Griggs RC, Batshaw M, Dunkle M, Gopal-Srivastava R, Kaye E, Krischer J, Nguyen T, Paulus K, Merkel PA. Clinical research for rare disease: opportunities, challenges, and solutions. *Mol. Genet. Metab.* Jan 2009;96(1):20-26. PMID: 19013090 , PMCID: PMC3134795

14. Richesson R, Shereff D, Andrews J. [RD] PRISM Library: Patient Registry Item Specifications and Metadata for Rare Diseases. *Journal of library metadata.* 2010;10(2-3):119-135. PMID: 21057650, PMCID: PMC2967796

15. Seminara J, Tuchman M, Krivitzky L, Krischer J, Lee HS, Lemons C, Baumgartner M, Cederbaum S, Diaz GA, Feigenbaum A, Gallagher RC, Harding CO, Kerr DS, Lanpher B, Lee B, Lichter-Konecki U, McCandless SE, Merritt JL, Oster-Granite ML, Seashore MR, Stricker T, Summar M, Waisbren S, Yudkoff M, Batshaw ML. Establishing a consortium for the study of rare diseases: The Urea Cycle Disorders Consortium. *Mol. Genet. Metab.* 2010;100 Suppl 1:S97-105. PMID: 20188616, PMCID: PMC2858794

16. Glaze DG, Percy AK, Skinner S, Motil KJ, Neul JL, Barrish JO, Lane JB, Geerts SP, Annese F, Graham J, McNair L, Lee HS. Epilepsy and the natural history of Rett syndrome. *Neurology.* Mar 16 2010;74(11):909-912. PMID: 20231667 , PMCID: PMC2836870

17. Percy AK, Lee HS, Neul JL, Lane JB, Skinner SA, Geerts SP, Annese F, Graham J, McNair L, Motil KJ, Barrish JO, Glaze DG. Profiling scoliosis in Rett syndrome. *Pediatr. Res.* Apr 2010;67(4):435-439. PMID: 20032810, PMCID: PMC2852102

18. Weng C, Tu SW, Sim I, Richesson R. Formal representation of eligibility criteria: a literature review. *J. Biomed. Inform.* Jun 2010;43(3):451-467. PMID: 20034594, PMCID: PMC2878905

19. Richesson RL, Smith SB, Malloy J, Krischer JP. Achieving standardized medication data in clinical research studies: two approaches and applications for implementing RxNorm. *J. Med. Syst.* Aug 2010;34(4):651-657. PMID: 20703919, PMCID: PMC2977947

20. Gentile JK, Tan WH, Horowitz LT, Bacino CA, Skinner SA, Barbieri-Welge R, Bauer-Carlin A, Beaudet AL, Bichell TJ, Lee HS, Sahoo T, Waisbren SE, Bird LM, Peters SU. A neurodevelopmental survey of Angelman syndrome with genotype-phenotype correlations. *J. Dev. Behav. Pediatr.* Sep 2010;31(7):592-601. PMID: 20729760, PMCID: PMC2997715

21. Percy AK, Neul JL, Glaze DG, Motil KJ, Skinner SA, Khwaja O, Lee HS, Lane JB, Barrish JO, Annese F, McNair L, Graham J, Barnes K. Rett syndrome diagnostic criteria: lessons from the Natural History Study. *Ann. Neurol.* Dec 2010;68(6):951-955. PMID: 21104896, PMCID: PMC3021984

22. Statland JM, Wang Y, Richesson R, Bundy B, Herbelin L, Gomes J, Trivedi J, Venance S, Amato A, Hanna M, Griggs R, Barohn RJ, The CINCH Consortium. An interactive voice response diary for patients with non-dystrophic myotonia. *Muscle Nerve.* 2011;44(1):30-35. PMID: 21674518, PMCID: PMC3233757

23. McCormack FX, Inoue Y, Moss J, et al. Efficacy and safety of sirolimus in lymphangioleiomyomatosis. *N. Engl. J. Med*. Apr 28 2011;364(17):1595-1606. PMID: 21410393, PMCID: PMC3118601

24. Richesson RL, Nadkarni P. Data standards for clinical research data collection forms: current status and challenges. *J. Am. Med. Inform. Assoc.* May 1 2011;18(3):341-346. PMID: 21486890 , PMCID: PMC3078665

25. Lane JB, Lee HS, Smith LW, et al. Clinical severity and quality of life in children and adolescents with Rett syndrome. *Neurology.* Nov 15 2011;77(20):1812-1818. PMID: 22013176, PMCID: PMC3233210

26. Lee HS, Paik MC, Rundek T, Sacco RL, Dong C, Krischer JP. Heritability Estimation using Regression Models for Correlation. *J Biom Biostat.* Nov 15 2011;2(119). PMID: 22457844, PMCID: PMC3312250

27. Monach PA, Tomasson G, Specks U, Stone JH, Cuthbertson D, Krischer J, Ding L, Fervenza FC, Fessler BJ, Hoffman GS, Ikle D, Kallenberg CG, Langford CA, Mueller M, Seo P, St Clair EW, Spiera R, Tchao N, Ytterberg SR, Gu YZ, Snyder RD, Merkel PA. Circulating markers of vascular injury and angiogenesis in antineutrophil cytoplasmic antibody-associated vasculitis. *Arthritis Rheum.* Dec 2011;63(12):3988-3997. PMID: 21953143, PMCID: PMC3227746

28. Kelley JM, Monach PA, Ji C, Zhou Y, Wu J, Tanaka S, Mahr AD, Johnson S, McAlear C, Cuthbertson D, Carette S, Davis JC, Jr., Dellaripa PF, Hoffman GS, Khalidi N, Langford CA, Seo P, St Clair EW, Specks U, Stone JH, Spiera RF, Ytterberg SR, Merkel PA, Edberg JC, Kimberly RP. IgA and IgG antineutrophil cytoplasmic antibody engagement of Fc receptor genetic variants influences granulomatosis with polyangiitis. *Proc. Natl. Acad. Sci. U. S. A.* Dec 20 2011;108(51):20736-20741. PMID: 22147912, PMCID: PMC3251158

29. Monach PA, Kumpers P, Lukasz A, Tomasson G, Specks U, Stone JH, Cuthbertson D, Krischer J, Carette S, Ding L, Hoffman GS, Ikle D, Kallenberg CG, Khalidi NA, Langford CA, Seo P, St Clair EW, Spiera R, Tchao N, Ytterberg SR, Haubitz M, Merkel PA. Circulating angiopoietin-2 as a biomarker in ANCA-associated vasculitis. *PLoS ONE.* 2012;7(1):e30197. PMID: 22279570, PMCID: PMC3261176

30. Shurin S, Krischer J, Groft SC. Clinical trials In BMT: ensuring that rare diseases and rarer therapies are well done. *Biol. Blood Marrow Transplant.* Jan 2012;18(1 Suppl):S8-11. PMID: 22226117

31. Tomasson G, Boers M, Walsh M, LaValley M, Cuthbertson D, Carette S, Davis JC, Hoffman GS, Khalidi NA, Langford CA, McAlear CA, McCune WJ, Monach PA, Seo P, Specks U, Spiera R, St Clair EW, Stone JH, Ytterberg SR, Merkel PA. Assessment of health-related quality of life as an outcome measure in granulomatosis with polyangiitis (Wegener's). *Arthritis Care Res (Hoboken).* Feb 2012;64(2):273-279. PMID: 21954229, PMCID: PMC3250569

32. Grayson PC, Tomasson G, Cuthbertson D, Carette S, Hoffman GS, Khalidi NA, Langford CA, McAlear CA, Monach PA, Seo P, Warrington KJ, Ytterberg SR, Merkel PA. Association of vascular physical examination findings and arteriographic lesions in large vessel vasculitis. *J. Rheumatol.* Feb 2012;39(2):303-309. PMID: 22174204, PMCID: PMC3729730

33. Nyland SB, Krissinger DJ, Clemente MJ, Irby RB, Baab KT, Jarbadan NR, Sokol L, Schaefer E, Liao J, Cuthbertson D, Epling-Burnette P, Paquette R, List AF, Maciejewski JP, Loughran TP, Jr. Seroreactivity to LGL leukemia-specific epitopes in aplastic anemia, myelodysplastic syndrome and paroxysmal nocturnal hemoglobinuria: Results of a bone marrow failure consortium study. *Leuk. Res.* Mar 1 2012. PMID: 22386729, PMCID: PMC3312981

34. Richesson RL, Sutphen R, Shereff D, Krischer JP. The Rare Diseases Clinical Research Network Contact Registry update: features and functionality. *Contemp. Clin. Trials.* Jul 2012;33(4):647-656. PMID: 22405970, PMCID: PMC3652679

35. Grayson PC, Maksimowicz-McKinnon K, Clark TM, Tomasson G, Cuthbertson D, Carette S, Khalidi NA, Langford CA, Monach PA, Seo P, Warrington KJ, Ytterberg SR, Hoffman GS, Merkel PA. Distribution of arterial lesions in Takayasu's arteritis and giant cell arteritis. *Ann. Rheum. Dis.* Aug 2012;71(8):1329-1334. PMID: 22328740, PMCID: PMC3729734

36. Motil KJ, Caeg E, Barrish JO, Geerts S, Lane JB, Percy AK, Annese F, McNair L, Skinner SA, Lee HS, Neul JL, Glaze DG. Gastrointestinal and nutritional problems occur frequently throughout life in girls and women with rett syndrome. *J. Pediatr. Gastroenterol. Nutr.* Sep 2012;55(3):292-298. PMID: 22331013, PMCID: PMC3393805

37. Couluris M, Kinder BW, Xu P, Gross-King M, Krischer J, Panos RJ. Treatment of idiopathic pulmonary fibrosis with losartan: a pilot project. *Lung.* Oct 2012;190(5):523-527. PMID: 22810758, PMCID: PMC4620709

38. Statland JM, Bundy BN, Wang Y, Trivedi JR, Raja Rayan D, Herbelin L, Donlan M, McLin R, Eichinger KJ, Findlater K, Dewar L, Pandya S, Martens WB, Venance SL, Matthews E, Amato AA, Hanna MG, Griggs RC, Barohn RJ. A quantitative measure of handgrip myotonia in non-dystrophic myotonia. *Muscle Nerve.* Oct 2012;46(4):482-489. PMID: 22987687, PMCID: PMC3564214

39. Statland JM, Bundy BN, Wang Y, Rayan DR, Trivedi JR, Sansone VA, Salajegheh MK, Venance SL, Ciafaloni E, Matthews E, Meola G, Herbelin L, Griggs RC, Barohn RJ, Hanna MG. Mexiletine for symptoms and signs of myotonia in nondystrophic myotonia: a randomized controlled trial. *JAMA.* Oct 3 2012;308(13):1357-1365. PMID: 23032552, PMCID: PMC3564227

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Conference Proceedings

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Advancing Research & Treatment for Frontotemporal Lobar Degeneration

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Brain Vascular Malformation Consortium

Book Chapters

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Journal Articles

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Brittle Bone Disorders Consortium

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Clinical Research in ALS & Related Disorders for Therapeutic Development

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Consortium of Eosinophilic Gastrointestinal Disease Researchers

Abstracts

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Developmental Synaptopathies Consortium

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Dystonia Coalition

Abstracts Presented at Conferences

1. LeDoux MS, Xiao J, Rudzinska M, Bastian RW, Van Gerpen JA, Wszolek ZK. Expansion of the Genotypic and Phenotypic Spectra of THAP1 Mutations. Poster presented at: 14th International Congress of Parkinson's Disease and Movement Disorders; June 13-17, 2010; Buenos Aires, Argentina.

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6. LeDoux MS. Expansion of the Genotypic and Phenotypic Spectra of THAP1 Mutations. *15th International Congress of Parkinson's Disease and Movement Disorders*. Toronto, Canada2011.

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12. Prudente C, Pardo C, Jinnah H. Neuropathology of idiopathic cervical dystonia. Paper presented at: 5th International Dystonia Symposium; October 20-22, 2011; Barcelona, Spain.
13. Self M, Corriveau R, Rosen A, et al. Establishment of a biorepository for primary dystonias: Collaborative efforts of the NINDS human genetic resource center and the Dystonia Coalition. Paper presented at: 5th International Dystonia Symposium; October 20-22, 2011; Barcelona, Spain.
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Genetic Disorders of Mucociliary Clearance

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Abstracts Presented at Conferences

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2. Hainline C, Rizzo D, Shy ME, Inherited Neuropathies Consortium, Rare Diseases Clinical Research Network Data Management and Coordinating Center. Enhancements to the RDCRN Contact Registry for the Inherited Neuropathies Consortium. Poster presented at Peripheral Nerve Society Annual Meeting; Jul. 8-12, 2017; Sitges, Spain.

Conference Proceedings

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Lysosomal Disease Network

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Nephrotic Syndrome Rare Disease Clinical Research Network

Abstracts Presented at Conferences

1. Barisoni L, Jennette JC, Hodgin JB, Herzenberg A, Kopp J, Conway C, Hewett S, Nast C. The NEPTUNE pathology scoring system: a novel virtual microscopy protocol for evaluation of nephrotic syndrome. Paper presented at: American Society of Nephrology (ASN) Renal Week; November 8-13, 2011; Philadelphia.

2. Kretzler M, Holzman LB, Ojo A, Gadegbeku C, Gipson D. NEPTUNE: the Nephrotic Syndrome Study Network in the Rare Diseases Clinical Research Network. Paper presented at: American Society of Nephrology (ASN) Renal Week; November 8-13, 2011; Philadelphia.

3. Barisoni L. Overview of the pathology of minimal change disease and focal segmental glomerulosclerosis. Paper presented at: ISN (International Society of Nephrology) Update Course in Nephrology; December 9-12, 2011; Dubai, UAE.

4. Kretzler M. NEPTUNE. Paper presented at: 9th International Podocyte Conference; April 22-25, 2012; Miami, FL.

5. Kretzler M. Translational research in nephrotic syndrome across disciplines and continents. Paper presented at: 2012 International Society of Nephrology (ISN) Continuous Medical Education (CME) on "Forefronts in Glomerular Disease" August 23-25, 2012; Nanjing, China.

6. Barisoni L, Nast C, Jennette J, et al. The NEPTUNE Digital Pathology Protocol for Evaluation of Nephrotic Syndrome. Paper presented at: American Society of Nephrology (ASN) Kidney Week; October 30 - November 4, 2012; San Diego, CA.

7. Kretzler M, Holzman L, Gadegbeku C, Gipson D, Consortium TNSSN. NEPTUNE: The Nephrotic Syndrome Study Network in the Rare Diseases Clinical Research Network. Paper presented at: American Society of Nephrology (ASN) Kidney Week; October 30 - November 4, 2012; San Diego, CA.

8. Wickman L, Afshinnia F, Yang Y, et al. Profiling Human Urinary mRNA. Paper presented at: American Society of Nephrology (ASN) Kidney Week; October 30 - November 4, 2012; San Diego, CA.

9. Gipson D. Pediatric Nephrotic Syndrome. Paper presented at: International Pediatric Nephrology Kidney Fellows Conference; January 21-24, 2013; Wyndham Rio Mar, Puerto Rico.

10. Kretzler M. Biobanking for rare renal disease, forefronts in nephrology. Paper presented at: Eurenomics General Assembly Meeting; April 3-6, 2013; Heidelberg, Germany.

11. Barisoni L. A new role of renal pathology in systems biology era of nephrotic syndrome. Paper presented at: The Second International Renal Meeting and Mayo Clinic Day Meeting; April 30 - May 3, 2013; Cagliari, Italy.

12. Kretzler M. International collaborations in rare disease. Paper presented at: Rare Diseases Clinical Research Network (RDCRN) Steering Committee Meeting; May 1, 2013; Rockville, MD.

13. Trachtman H, Sethna C, Gibson K, et al. Focal Segmental Glomerulosclerosis (FSGS) in Pediatrics: Comparison R of Patients enrolled in an Observational Versus Interventional Trial. Paper presented at: Pediatric Academic Societies (PAS) Annual Meeting; May 4-7, 2013; Washington, DC.

14. Gibson K, Greenbaum L, Trachtman H, et al. NEPTUNE Cohort Study: Pediatric Perspectives. Paper presented at: International Pediatric Nephrology Association Congress; August 30 - September, 2013; Shanghai, China.

15. Gipson D. Patient Reported Outcomes in Nephrotic Syndrome. Paper presented at: Rare Disease Clinical Research Network (RDCRN) Steering Committee Meeting; October 2, 2013; Rockville, MD.

16. Barisoni L. Molecular Diagnosis and Classification of FSGS. Paper presented at: American Society of Nephrology Kidney Week; November 5-10, 2013; Atlanta, GA.

17. Barisoni L, Nast C. The NEPTUNE Digital Pathology Morphologic Profiling Of Nephrotic Syndrome. Paper presented at: American Society of Nephrology Kidney Week; November 5-10, 2013; Atlanta, GA.

18. Gibson K, Troost J, Trachtman H, et al. Baseline Immunosuppression Exposure in Neptune Study Cohort. Paper presented at: American Society of Nephrology Kidney Week; November 5-10, 2013; Atlanta, GA.

19. Hogan M, Reich H, Nelson P, et al. Random Urine Protein to Creatinine Ratio (UPCR) Is a Relatively Poor Predictor of 24 hr Protein Excretion in Patients with Glomerular Disease. Paper presented at: American Society of Nephrology Kidney Week; November 5-10, 2013; Atlanta, GA.

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23. Sampson M, Tan A, Gadebeku C, Kretzler M, Hang H. Genetic Epidemiology and Other Genetic Studies of Common Kidney Diseases. Paper presented at: American Society of Nephrology Kidney Week; November 5-10, 2013; Atlanta, GA.

24. Trachtman H, Lafayette R. Pathobiology: Clinical/Diagnostic Renal Pathology and Lab Medicine – II. Paper presented at: American Society of Nephrology Kidney Week; November 5-10, 2013; Atlanta, GA.

25. Wickman L, Afshinnia F, Wang S, et al. Urine Podocyte mRNAs, Proteinuria, and Progression in Human Glomerular Diseases. Paper presented at: American Society of Nephrology Kidney Week; November 5-10, 2013; Atlanta, GA.

26. Tanzer M, Smith K, Light C, Shereff D, Kretzler M, Gipson D. Assessment of the Educational Experience for Patients with a New Chronic Illness. Paper presented at: Pediatric Academic Society 2014 Annual Meeting; May 3-6, 2014; Vancouver, Canada.

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28. Martini S, Nair V, Eichinger F, Reich N, Kretzler M. Transcriptome-based cluster analysis identifies TNF-subgroup in Focal and Segmental Glomerulosclerosis (#4799). Paper presented at: American Society of Nephrology Kidney Week; November 11-16, 2014; Philadelphia, PA.

29. Reich H, Nair V, Ju W, et al. Biomarkers of disease pathobiology in patients with glomerulonephritis in the NEPTUNE cohort. Paper presented at: American Society of Nephrology Kidney Week; November 11-16, 2014; Philadelphia, PA.

30. Sampson M, Mariani L, Martini S, et al. Integrative genomics identifies novel Associations of APOL1 risk genotype in African-American NEPTUNE subjects. Paper presented at: American Society of Nephrology Kidney Week; November 11-16, 2014; Philadelphia, PA.

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32. Mariani L, Zee J, Wang T, et al. Non-linear progression of kidney disease in Nephrotic Syndrome. Paper presented at: American Society of Nephrology Kidney Week; November 2-8, 2015; San Diego, CA.

33. Nair V, Zhu H, Mariani L, et al. Urinary EGF predicts composite endpoints in three independent chronic kidney disease cohorts. Paper presented at: American Society of Nephrology Kidney Week; November 2-8, 2015; San Diego, CA.

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Journal Articles

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North American Mitochondrial Diseases Consortium

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Porphyrias Consortium

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Conference Proceedings

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Primary Immune Deficiency Treatment Consortium

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Rare Kidney Stone Consortium

Book Chapters

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Abstracts Presented at Conferences

1. Edvardsson V, Gudbjartsson D, Indridason O, et al. Sequence Variants at the UMOD Gene Associate with Risk of Kidney Stone Disease. American Society of Nephrology Kidney Week. San Diego, CA2009.
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5. Monico C, Olson J, Bergstralh E, Heilman R, Milliner D. Effect of betaine on urine oxalate in primary hyperoxaluria, type I. American Society of Nephrology (ASN) Kidney Week. Philadelphia, PA2011.
6. Lieske J. How do kidney stones start? Paper presented at: Second International Istanbul Urolithiasis Days; February, 2011; Istanbul, Turkey.
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8. Milliner D. Hyperoxaluria: an update. Paper presented at: Pediatric Nephrology Seminar XXXVIII; March, 2011; Miami Beach, Florida.
9. Lewis M, Sahota A, Ward MD, Goldfarb DS, Holmes E, Want EJ. Characterization of small molecule aggregate components in cystine stones as a screen for candidate stone inhibitors. Paper presented at: American Society for Mass Spectrometry; June, 2011; Denver, CO.
10. Milliner D. Effects of betaine as molecular chaperone in primary hyperoxaluria, type I. Paper presented at: Research on Calculus Kinetics Society; September, 2011; Boston, Massachusetts.
11. Langman C, Vural G, Brooks E. Proteomics in genetic kidney stone disease. Paper presented at: 44th Annual Scientific Meeting of European Society of Paediatric Nephrology (ESPN); September 14-17, 2011; Dubrovnik, Croatia.
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13. Sahota A, Yang M, Shikhel S, Kathuria N, Lewis M, Ward MD, Goldfarb DS, Tischfield JA. A new pharmacologic approach for cystinuria. Paper presented at: American Society for Human Genetics; October, 2011; Montreal, Canada.
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15. Sigurdsson B, Thorsteinsdottir M, Eiriksson FF, Edvardsson VO, Palsson R. Rapid method for determination of urinary 2,8-dihydroxyadenine concentration with by UPLC-MS/MS. Paper presented at: Annual Meeting of the European Bioanalysis Forum; November, 2011; Barcelona, Spain.

16. Mattoo A, Modersitzki F, Asplin J, Cohen BH, Grasso M, Goldfarb DS. Clinical validation of a novel assay used for monitoring treatment of patients with cystinuria. Paper presented at: Annual meeting of the American Society of Nephrology; November, 2011; Philadelphia, PA.

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18. Edvardsson V. Clinical features and long-term renal outcome in Icelandic patients with APRT deficiency and 2,8-dihydroxyadeninuria. *The 11th Congress of the European Society of Internal Medicine*. Madrid, Spain2012.

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21. Lieske J. Primary Hyperoxaluria: not such a rare disease? *Research on Calculous Kinetics Society Session at the American Urological Association Meeting*. Atlanta, GA2012.

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23. Runolfsdottir H, Palsson R, Agustsdottir I, Edvardsson V. Clinical Features and Long-Term Renal Outcome in Icelandic Patients with APRT Deficiency and 2,8-Dihydroxyadeninuria. American Society of Nephrology (ASN) Kidney Week. San Diego, CA2012.

24. Edvardsson V. Rare causes of kidney stones and kidney failure: focus on APRT deficiency and 2, 8-dihydroxyadeninuria. Paper presented at: Symposium on Rare Disease, Annual Meeting of the Icelandic Medical Association; January, 2012; Reykjavic, Iceland.

25. Lieske J. Stones from the inside out: what every urologist sees but few nephrologists know about. Paper presented at: Hong Kong Society of Nephrology, Renal Commissioned Training; January, 2012; Hong Kong, People's Republic of China.

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Rare Lung Diseases Consortium

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Rett Syndrome, MECP2 Duplication, and Rett-related Disorders Consortium

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Sterol and Isoprenoid Diseases Consortium

Book Chapters

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Urea Cycle Disorders Consortium

Abstracts Presented at Conferences

1. Lichter-Konecki U, Cabrera-Luque J, Moses L, Gallo V. Identifying astrocyte functions altered during hyperammonemic encephalopathy. Paper presented at: Society for Neuroscience Annual Meeting; November 12-16, 2005; Washington, DC.

2. Tuchman M. Collaborative Investigations of Urea Cycle Disorders: The Importance of Research Networks in the Study of Rare Diseases. Paper presented at: American Society of Human Genetics Annual Meeting; October 23-27, 2007; San Diego, CA.

3. Gropman A. 13C MRS study of ornithine transcarbamylase deficiency (OTCD). Paper presented at: Society for Inherited Metabolic Disorders Meeting; March, 2008; Asilomar, CA.
4. Kahn I, Seltzer R, Van Meter J, Gropman A. Diffusion tensor imaging detects areas of abnormal white matter microstructure in patients with partial ornithine transcarbamylase deficiency (OTCD). Paper presented at: Society for Inherited Metabolic Disorders Meeting; March, 2008; Asilomar, CA.
5. Tuchman M. Setting up Multi-Institutional Network Research in Rare Diseases: The Urea Cycle Consortium. Paper presented at: Society for Inherited Metabolic Disorders Meeting; March, 2008; Asilomar, CA
6. Lee B. Urea cycle disorders best practices and new developments: clinical presentation, laboratory diagnosis, and chronic management. Paper presented at: American College of Medical Genetics Annual Clinical Genetics Meeting; March 12-16, 2008; Phoenix, AZ.
7. Lichter-Konecki U. A Rare Disease Clinical Research Consortium for the collaborative investigation of UCDs Paper presented at: Pediatric Academic Societies Annual Meeting; May, 2009; Baltimore, MD.
8. Batshaw M. The Urea Cycle Disorders Consortium. Paper presented at: National Urea Cycle Disorders Foundation; July, 2009; Pasadena, California.
9. Batshaw M. The Urea Cycle Disorders Consortium. Paper presented at: Satellite Symposium to the 11th International Congress on Inborn Errors of Metabolism; August, 2009; La Jolla, California.
10. Batshaw M. Setting up multi-institutional research network in rare disease: The Urea Cycle Disorders Consortium. Paper presented at: Institute of Medicine Committee on Accelerating Rare Diseases Research and Orphan Product Development; November 23, 2009.
11. Gropman A, Shattuck K, Prust M, et al. Increased dorsolateral prefrontal cortex activation in OTCD during working memory. Paper presented at: National Urea Cycle Disorders Foundation Annual Conference; July 9-11, 2010; Cambridge, MA.
12. Krivitzky L, Waisbren S. Neuropsychological Functioning in Rare Diseases; Research Challenges and Potential Solutions. Paper presented at: 39th Annual Meeting of the International Neuropsychological Society; February 2-5, 2011; Boston, MA.
13. Breedan A, Prust M, Krivitzky L, Gropman A. Cognitive Tests Sensitive to the Neurological Impairment in Ornithine Transcarbamylase Deficiency (OTCD). Paper presented at: 34th Annual Meeting of the Society for Inherited Metabolic Disorders; February 27 - March 2, 2011; Asilomar, CA.
14. Lichter U. Update on the urea cycle disorders registry. Paper presented at: 34th Annual Meeting of the Society for Inherited Metabolic Disorders; February 27 - March 2, 2011; Asilomar, CA.
15. Shattuck K, Prust M, Seltzer R, et al. Increased Dorsolateral Prefrontal Cortex Activation in Ornithine Transcarbamylase Deficiency (OTCD) During Working memory: An fMRI study. Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; February 27 - March 2, 2011; Asilomar, CA.

16. Shattuck K, Prust M, Seltzer R, Hailu A, vanMeter J, Gropman A. Altered Neural Activation in ornithine Transcarbamylase Deficiency during working memory. Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; February 27 - March 2, 2011; Asilomar, CA.

17. Batshaw M. The Urea Cycle Disorders Consortium (UCDC). Paper presented at: 4th Annual Rare Disease Day February 28, 2011; Bethesda, MD.

18. Gropman A, Shattuck K, Prust M, et al. Increased dorsolateral prefrontal cortex activation in OTCD during working memory. Paper presented at: 11th International Congress of the European Society for Magnetic Resonance in Neuropediatrics; March 24-26, 2011; Amsterdam, The Netherlands.

19. Batshaw M. Data Quality Challenges of Multisite Clinical Trials Panel. Paper presented at: Quality Data from Pediatric Clinical Trials Meeting; October 21, 2011; Bethesda, MD.

20. Diaz G, Krivitzky L, Mokhtarani M, et al. Ammonia (NH3) Control and Improved Neurocognitive Outcome Among Urea Cycle Disorder (UCD) Patients Treated with Glycerol Phenylbutyrate (GPB). Paper presented at: American College of Medical Genetics Conference; March 27-31, 2012; Charlotte, NC.

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27. Lee B, Mokhtarani M, Diaz G, et al. Optimizing ammonia (NH3) control in urea cycle disorder (UCD) patients: short and long-term implications. Paper presented at: Garrod Association Symposium; May 31 - June 1, 2013; Sherbrooke, Canada.
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29. Lee B, Mokhtarani M, Diaz G, et al. Optimizing ammonia (NH3) control in urea cycle disorder (UCD) patients: short and long-term implications. Paper presented at: 12th International Congress of Inborn Errors of Metabolism; September 3-6, 2013; Barcelona, Spain.

Conference Proceedings

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2. Bhavsar S, Khalidi N, Carette S, et al. Venothromboembolism in Large Vessel Vasculitis. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting; October 2014, 2014; Boston, MA.
3. Carmona F, Mackie S, Martin J, et al. An Immunochip Study Confirms a Strong Contribution of HLA Class I and II Genes in the Susceptibility to Giant Cell Arteritis. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting2014; Boston, MA.
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7. Sy A, Dehghan N, Khalidi N, et al. Vasculitis and Inflammatory Bowel Diseases: A Study of 32 Patients with Both Conditions and Systematic Review of the Literature. Paper presented at: American College of Rheumatology (ACR/ARHP) Annual Meeting2014; Boston, MA.

Journal Articles

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12. Gropman AL, Seltzer RR, Yudkoff M, Sawyer A, VanMeter J, Fricke ST. 1H MRS allows brain phenotype differentiation in sisters with late onset ornithine transcarbamylase deficiency (OTCD) and discordant clinical presentations. *Mol. Genet. Metab.* May 2008;94(1):52-60. PMID: 18262815, PMCID: PMC2486377
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Vasculitis Clinical Research Consortium

Book Chapters

1. Mahr A, Merkel PA. Vasculitis. In: Creager MA, ed. *Atlas of vascular disease*. 3rd ed. Philadelphia, PA: Current Medicine Group/Springer; 2008.

2. Rodriguez-Pla A. Polyarteritis nodosa and other necrotizing systemic vasculitis. In: Gómez-Reino ea, ed. *Handbook of the Spanish Society of Rheumatology*. 5th ed: Editorial Médica Panaméricana, S.A.; 2008.

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Abstracts Presented at Conferences

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RDCRN Publications, Previous Partners

Angelman, Rett, and Prader-Willi Syndromes Consortium

Bone Marrow Failure Consortium

Cholestatic Liver Disease Consortium

Clinical Research Consortium for Spinocerebellar Ataxias

Consortium for Clinical Investigation of Neurologic Channelopathies

Rare Genetic Steroid Disorders

Rare Thrombotic Diseases

Salivary Gland Carcinomas Consortium

Angelman, Rett, and Prader-Willi Syndromes Consortium

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Abstracts Presented at Conferences

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3. Beaudet A, Bacino C, Bird L, Kimonis V, Peters S, Bichell T, Barbieri-Welge R, Shinawi L. Results of a trial of folic acid and betaine in Angelman syndrome and future directions. Paper presented at: Biennial Angelman Syndrome Foundation Conference; June 28, 2005; Anaheim, CA.
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Special Projects

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2. Butler MG, Welch J, Riske M, Vogel R, Troxell R, Rope A. Prader-Willi Syndrome. *Medical Home Portal* 2009; http://www.medicalhomeportal.org/diagnoses-and-conditions/prader-willi-syndrome/description.

Bone Marrow Failure Consortium

Abstracts Presented at Conferences

1. Epling-Burnette P, Xhen X, Bai F, Lubomir S, Ku E, Painter J, JianXiang Z, Edwards T, Julie D, Lynn M, Loughran T, S W. Aberrant NK receptor signaling associated with large granular lymphoycte and primary pulmonary hypertension. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.

2. Sekeres M, List A, Cuthbertson D, Paquette R, Loughran T, Maciejewski J. Preliminary Results from a Phase I study of Revlimid (Lenalidomide) in combination with Vidaza (Azacitidine) in patients with advanced myelodysplastic syndromes (MDS). Paper presented at: American Society of Hematology; December 8-11, 2007; Altanta, GA.

3. Epling-Burnette P, Sokol L, Moscinski L, Elkabani M, Bai F, Blaskovich M, Zou J, Painter J, Sebti S, Loughran T. First clinical report on Tipifarnib for the treatment of T-large granular lymphocyte (LGL) leukemia. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.

4. Wei S, Rocha K, Williams A, Chen X, Burnette P, Djeu J, Liu Q, Byrd J, Sokol L, Lawrence N, Pireddu R, Dewald G, Maciejewski J, List A. Gene dosage of the cell cycle regulatory phosphatases Cdc25C and PP2A determines sensitivity to lenalidomide in del(5q) MDS. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.

5. Bai F, Zou J, Wei S, Painter J, Blaskovich M, Sebti S, Loughran T, Epling-Burnette P. Th2 polarization induced by the farnesyltransferase inhibitor Tipifarnib (Zarnestra, R115777) through suppression of t-bet. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.

6. Painter J, Costellanos A, Bai F, Wei S, Loughran T, Epling-Burnette P. Constitutive activation of ras and mitogen-activated protein kinase (MAPK/ERK) in LGL leukemia unrelated to activating

mutations in N and KRas genes. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta.

7. Nyland S, Cuthbertson D, Loughran T. Elevated fas ligand and BA21 serorecognition as disease correlates in bone marrow failure disorders: taking advantage of the LGL leukemia model. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.
8. Gondek L, Tiu R, Wlodarski M, O'Keefe C, McDevitt M, Maciejewski J. Enhancement of cytogenetic diagnosis of myeloid disorders through application of SNP-array-based karyotyping. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.
9. O'Keefe C, Gondek L, Woodarski M, Karp J, McDevitt M, Maciejewski J. Can genomic copy number variants be a part of complex genetic traits predisposing to marrow failure? Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta.
10. Gondek L, Dunbar A, O'Keefe C, McDevitt M, Batista D, Theil K, Maciejewski J. SNP-A karyotyping facilitates improved mapping of deletions and uniparental disomy within the long arm of Chromosome 5 in myeloid disorders. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.
11. Serio B, Ramsingh G, Tiu R, Risitano A, Sekeres M, Maciejewski J. Immunogenetic analysis reveals the association of INF (+874 A/T) hypersecretor genotype in AA and a low frequency of KIR-2DL3/C1 mismatch in responders to immunosuppression. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.

Journal Articles

1. Nearman ZP, Wlodarski M, Jankowska AM, Howe E, Narvaez Y, Ball E, Maciejewski JP. Immunogenetic factors determining the evolution of T-cell large granular lymphocyte leukaemia and associated cytopenias. *Br. J. Haematol.* Jan 2007;136(2):237-248. PMID: 17156396
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6. Sekeres MA, Schoonen WM, Kantarjian H, List A, Fryzek J, Paquette R, Maciejewski JP. Characteristics of US patients with myelodysplastic syndromes: results of six cross-sectional physician surveys. *J. Natl. Cancer Inst.* Nov 5 2008;100(21):1542-1551. PMID: 18957672, PMCID: PMC2579314

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9. Sekeres MA, Maciejewski JP, Giagounidis AA, Wride K, Knight R, Raza A, List AF. Relationship of treatment-related cytopenias and response to lenalidomide in patients with lower-risk myelodysplastic syndromes. *J. Clin. Oncol.* Dec 20 2008;26(36):5943-5949. PMID: 19018091, PMCID: PMC2645116

10. Sekeres MA, Elson P, Kalaycio ME, Advani AS, Copelan EA, Faderl S, Kantarjian HM, Estey E. Time from diagnosis to treatment initiation predicts survival in younger, but not older, acute myeloid leukemia patients. *Blood.* Jan 1 2009;113(1):28-36. PMID: 18827183, PMCID: PMC2614639

11. Uchida K, Nakata K, Suzuki T, Luisetti M, Watanabe M, Koch DE, Stevens CA, Beck DC, Denson LA, Carey BC, Keicho N, Krischer JP, Yamada Y, Trapnell BC. Granulocyte/macrophage-colony-stimulating factor autoantibodies and myeloid cell immune functions in healthy subjects. *Blood.* Mar 12 2009;113(11):2547-2556. PMID: 19282464, PMCID: PMC2656275

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16. Uchida K, Carey B, Suzuki T, Nakata K, Trapnell B. Response: Granulocyte/macrophage colony-stimulating factor autoantibodies and myeloid cell immune functions in healthy persons. *Blood.* Jan 14 2010;115(2):431-433. PMID: 20075172, PMCID: PMC2808163

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Cholestatic Liver Disease Consortium

Abstracts Presented at Conferences

1. Shneider B, Brown M, Sokol R, Whitington P, Schwarz K, Squires R, Bucuvalas J, Haber B, Sheperd R, Rosenthal P, Robuck P, BARC. A multi-center analysis of outcome at 24 months of age in children with biliary atresia in the United States. Paper presented at: Annual Meeting of the American Association for the Study of Liver Diseases; November, 2004; Boston, MA.
2. Haber B, Brown M, Shneider B, Sokol R, Whitington P, Derusso P, Squires R, Bezerra J, Sheperd R, Rosenthal P, Robuck P, BARC. Patient demographics and clinical practices in a US multi-center study of biliary atresia. Paper presented at: Annual Meeting of the American Association for the Study of Liver Diseases; November 2004; Boston, MA.
3. DeRusso P, Ye W, Haber B, Shneider B, Sokol R, Whitington P, Squires R, Bezerra J, Shepherd R, Rosenthal P, Hoofnagle J. Early growth failure after portoenterostomy is associated with liver transplantation or death in infants with biliary atresia. Paper presented at: 56th Annual Meeting of the American Association for the Study of Liver Diseases; November, 2005; San Francisco, CA.
4. Shneider B, Norton K, Superina R, Erlichman J, Magee J, Bucuvalas J, Whitington P, Rosenthal P, Squires R, Benson J, Karpen S, Shepherd R, Sokol R. Diagnostic imaging in neonatal cholestasis: a multi-center prospective analysis. Paper presented at: AASLD Annual Meeting; October, 2006; Boston, MA.
5. Russo P, Boitnott J, Bove K, Brown M, Finegold M, J H, Jaffe R, Kim G, Magee J, Magid M, Melin-Aldana H, Sokol R, White F. A multi-institutional study of interobserver agreement on the histologic diagnosis of biliary obstruction in liver biopsies of cholestatic infants less than six months of age. Paper presented at: AASLD Annual Meeting; October, 2006; Boston, MA.
6. Shneider B, Shepherd R, Magee J, Bucuvalas J, Haber B, Karpen S, Rosenthal P, Schwarz K, Suchy F, Whitington P, Sokol R. Discriminating features of biliary atresia-a prospective multi-centered analysis. Paper presented at: AASLD Annual Meeting2007; Boston, MA.
7. Miethke A, Matte U, Liu C, Balistreri W, Ryckman F, Bezerra J. Gene mutations and clinical outcome after biliary diversion surgery for intractable pruritus in children with intrahepatic cholestasis. Paper presented at: NASPGHAN annual meeting; October, 2007; Salt Lake City, UT.
8. Gondek L, Tiu R, Wlodarski M, O'Keefe C, McDevitt M, Maciejewski J. Enhancement of cytogenetic diagnosis of myeloid disorders through application of SNP-array-based karyotyping. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.
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10. Haber B, Fredericks E, Magee J, Bezerra JA, Karpen S, Kerkar N, Rosenthal P, Schwarz K, Shepherd R, Shneider B, Whitington P, Robuck P, Sokol R, BARC. Predictors of neurodevelopmental outcome in non-transplanted children with biliary atresia at one year of age. Paper presented at: AASLD Annual Meeting; November 2008.
11. Schwarz K, Shepherd R, Magee J, Rosenthal P, Mack C, Raghumathan T, Bezerra J, Haber B, Karpen S, Suchy F, Whitington P, Robuck P, Sokol R, BARC. Clinical and demographic features of three major biliary atresia phenotypes in the BARC study. Paper presented at: AASLD Annual Meeting; November 2008.
12. Heubi J, Setchell K, Rosenthal P, Shah S, Buckley D, Jha P, Zhang W, Potter C, Suskind D, Bull L. Oral glycocholic acid treatment of patients with bile acid amidation defects improves growth and fat-soluble vitamin absorption; 2009, 2009.
13. Sokol R, Hines J. The Cholestatic Liver Disease Consortium (CLiC): a multi-institutional collaboration to better understand and treat liver disease in children. Paper presented at: NIH conference "Advancing Rare Diseases Research through Networks and Collaboration."; July 16, 2009; Bethesda, MD.
14. Hahn C, Sokol R, Hines J. The patient advocacy group committee of the Cholestatic Liver Disease Consortium: giving families, parents and patients a partnership with researchers. Paper presented at: NIH conference "Advancing Rare Diseases Research through Networks and Collaboration." July 16, 2009; Bethesda, MD.
15. Bass LM, Xie H, Yu S, Malladi P, Bento-Soares M, Whitington P. Expression in progressive familial intrahepatic cholestasis. Paper presented at: AASLD Liver Meeting; Oct. 30-Nov. 1, 2009; Boston, MA.
16. Sundaram S, Fredericks E, Kamath B, Haber B, Raghumathan T, Magee J, Bezerra J, Karpen S, Kerkar N, Rosethal P, Schwarz K, Shepherd R, Schneider B, Whitington P, Robuck P, Sokol R, BARC. Cross sectional assessment of quality of life in biliary aresia patients ages 2-25 years. Paper presented at: AASLD Annual Meeting; November, 2009; Boston, MA.
17. Sokol R, Magee J, Hahn C, Robuck P. The new Childhood Liver Disease Research and Education Network (ChiLDREN): a new cooperative effort between NIDDK, academic centers and patient advocacy groups. Paper presented at: NASPGHAN Annual Meeting; November 2009; National Harbor, MD.
18. Shneider B, Abel R, Raghunathan T, Magee J, Bezerra J, Haber B, Karpen S, Rosenthal P, Schwarz K, Shepherd R, Suchy F, Whitington P, Robuck P, Sokol R, BARC. A prospective multi-centered investigation of vitamin supplementation in infants with biliary atresia: interim analysis from the Biliary Atresia Research Consortium (BARC). Paper presented at: AASLD Annual Meeting; November 2009; Boston, MA.
19. Shneider B, Abel R, Haber B, Karpen S, Magee J, Romero R, Schwartz K, Bass L, Kerkar N, Miethke A, Rosenthal P, Turmelle Y, Sokol R. Multi-center analysis of portal hypertension in 163 children with biliary atresia. Paper presented at: Annual Meeting of the American Association for the Study of Liver Disease; November 2010; Boston, MA.

Conference Proceedings

1. Bass LM, Malladi P, Whitington P. FXR response in cholestatic liver disease. AASLD Abstracts 676–894. *Hepatology.* 2008;48(S1):609A-706A.

Journal Articles

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5. Shneider BL, Brown MB, Haber B, Whitington PF, Schwarz K, Squires R, Bezerra J, Shepherd R, Rosenthal P, Hoofnagle JH, Sokol RJ. A multicenter study of the outcome of biliary atresia in the United States, 1997 to 2000. *J. Pediatr.* Apr 2006;148(4):467-474. PMID: 16647406
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7. McDaniell R, Warthen DM, Sanchez-Lara PA, Pai A, Krantz ID, Piccoli DA, Spinner NB. NOTCH2 mutations cause Alagille syndrome, a heterogeneous disorder of the notch signaling pathway. *Am. J. Hum. Genet.* Jul 2006;79(1):169-173. PMID: 16773578, PMCID: PMC1474136
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9. Setchell KD, Heubi JE. Defects in bile acid biosynthesis--diagnosis and treatment. *J. Pediatr. Gastroenterol. Nutr.* Jul 2006;43 Suppl 1:S17-22. PMID: 16819396
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11. He M, Rutledge SL, Kelly DR, et al. A new genetic disorder in mitochondrial fatty acid beta-oxidation: ACAD9 deficiency. Am. J. Hum. Genet. Jul 2007;81(1):87-103. PMID: 17564966, PMCID: PMC1950923
12. Heubi JE, Setchell KD, Bove KE. Inborn errors of bile acid metabolism. *Semin. Liver Dis.* Aug 2007;27(3):282-294. PMID: 17682975
13. Lee WS, Sokol RJ. Liver disease in mitochondrial disorders. *Semin. Liver Dis.* Aug 2007;27(3):259-273. PMID: 7682973, PMCID: PMC3888320
14. Sokol RJ, Shepherd RW, Superina R, Bezerra JA, Robuck P, Hoofnagle JH. Screening and outcomes in biliary atresia: summary of a National Institutes of Health workshop. *Hepatology.* Aug 2007;46(2):566-581. PMID: 7661405, PMCID: PMC3888317
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Chronic Graft Versus Host Disease Consortium

Conference Proceedings

1. Khera N, Xiaoyu C, Duong H, et al. Prospective Longitudinal Study of Late Acute Graft Versus Host Disease after Hematopoietic Cell Transplantation: A Report from Chronic GVHD Consortium. Paper presented at: BMT Tandem Meetings2015; San Diego, CA.

2. Palmer J, Chai X, Martin P, et al. Physician-Reported CR+PR at 6 Months Predicts Subsequent Survival in Patients with Chronic GVHD. Paper presented at: BMT Tandem Meetings2015; San Diego, CA.

3. Wood W, Lee S, Chai X, et al. Survival without Progressive Impairment As a Novel Endpoint in Chronic Graft-Versus-Host Disease. Paper presented at: BMT Tandem Meetings2015; San Diego, CA.

4. Yu J, Storer B, Daguindau E, et al. A Biomarker Panel for Chronic Graft-Versus-Host Disease. Paper presented at: BMT Tandem Meetings2015; San Diego, CA.

Journal Articles

1. Chronic Graft Versus Host Disease Consortium. Rationale and design of the chronic GVHD cohort study: improving outcomes assessment in chronic GVHD. *Biol. Blood Marrow Transplant.* Aug 2011;17(8):1114-1120. PMID: 21664473, PMCID: PMC4016312

2. Pidala J, Kurland BF, Chai X, Vogelsang G, Weisdorf DJ, Pavletic S, Cutler C, Majhail N, Lee SJ. Sensitivity of changes in chronic graft-versus-host disease activity to changes in patient-reported quality of life: results from the Chronic Graft-versus-Host Disease Consortium. *Haematologica.* Oct 2011;96(10):1528-1535. PMID: 21685473, PMCID: PMC3186315

3. Mitchell SA, Jacobsohn D, Thormann Powers KE, et al. A multicenter pilot evaluation of the National Institutes of Health chronic graft-versus-host disease (cGVHD) therapeutic response measures: feasibility, interrater reliability, and minimum detectable change. *Biol. Blood Marrow Transplant.* Nov 2011;17(11):1619-1629. PMID: 21536143, PMCID: PMC3158826

4. Pidala J, Vogelsang G, Martin P, Chai X, Storer B, Pavletic S, Weisdorf DJ, Jagasia M, Cutler C, Palmer J, Jacobsohn D, Arai S, Lee SJ. Overlap subtype of chronic graft-versus-host disease is associated with an adverse prognosis, functional impairment, and inferior patient-reported

outcomes: a Chronic Graft-versus-Host Disease Consortium study. *Haematologica.* Mar 2012;97(3):451-458. PMID: 22058206, PMCID: PMC3291602

5. Pidala J, Chai X, Martin P, et al. Hand grip strength and 2-minute walk test in chronic graft-versus-host disease assessment: analysis from the Chronic GVHD Consortium. *Biol. Blood Marrow Transplant.* Jun 2013;19(6):967-972. PMID: 23542686, PMCID: PMC3966477
6. Jacobsohn DA, Kurland BF, Pidala J, et al. Correlation between NIH composite skin score, patient-reported skin score, and outcome: results from the Chronic GVHD Consortium. *Blood.* Sep 27 2012;120(13):2545-2552; quiz 2774. PMID: 22773386, PMCID: PMC3460679
7. Inamoto Y, Martin PJ, Chai X, et al. Clinical benefit of response in chronic graft-versus-host disease. *Biol. Blood Marrow Transplant.* Oct 2012;18(10):1517-1524. PMID: 22683612, PMCID: PMC3443259
8. Inamoto Y, Storer BE, Lee SJ, et al. Failure-free survival after second-line systemic treatment of chronic graft-versus-host disease. *Blood.* Mar 21 2013;121(12):2340-2346. PMID: 23321253, PMCID: PMC3606068
9. Arora M, Pidala J, Cutler CS, et al. Impact of prior acute GVHD on chronic GVHD outcomes: a chronic graft versus host disease consortium study. *Leukemia.* Apr 2013;27(5):1196-1201. PMID: 23047477, PMCID: PMC3942496
10. Pidala J, Chai X, Kurland BF, et al. Analysis of gastrointestinal and hepatic chronic grant-versus-host disease manifestations on major outcomes: a chronic grant-versus-host disease consortium study. *Biol. Blood Marrow Transplant.* May 2013;19(5):784-791. PMID: 23395601, PMCID: PMC3896215
11. Inamoto Y, Storer BE, Petersdorf EW, et al. Incidence, risk factors, and outcomes of sclerosis in patients with chronic graft-versus-host disease. *Blood.* Jun 20 2013;121(25):5098-5103. PMID: 23547053, PMCID: PMC3689252
12. Pidala J, Sarwal M, Roedder S, Lee SJ. Biologic markers of chronic GVHD. *Bone Marrow Transplant.* Jul 22 2013. PMID: 23872737, PMCID: PMC3976639
13. Inamoto Y, Martin PJ, Storer BE, et al. Association of severity of organ involvement with mortality and recurrent malignancy in patients with chronic graft-versus-host disease. *Haematologica.* 2014;99(10):1618-1623. PMID: 24997150, PMCID: PMC4181259
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16. Palmer J, Williams K, Inamoto Y, et al. Pulmonary symptoms measured by the national institutes of health lung score predict overall survival, nonrelapse mortality, and patient-reported outcomes in chronic graft-versus-host disease. *Biol. Blood Marrow Transplant.* Mar 2014;20(3):337-344. PMID: 24315845, PMCID: PMC3973401
17. Allen JL, Tata PV, Fore MS, et al. Increased BCR responsiveness in B cells from patients with chronic GVHD. *Blood.* Mar 27 2014;123(13):2108-2115. PMID: 24532806, PMCID: PMC3968393

18. Inamoto Y, Jagasia M, Wood WA, et al. Investigator feedback about the 2005 NIH diagnostic and scoring criteria for chronic GVHD. *Bone Marrow Transplant.* Apr 2014;49(4):532-538. PMID: 24464142, PMCID: PMC3975688

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Clinical Research Consortium for Spinocerebellar Ataxias

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Consortium for Clinical Investigation of Neurologic Channelopathies

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