Rare Diseases Clinical Research Network (RDCRN) Publications

Updated: June 5, 2018

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

Abstracts Presented at Conferences
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.


Contact Registry for the Inherited Neuropathies Consortium. Poster presented at Peripheral Nerve Society Annual Meeting; Jul. 8-12, 2017; Sitges, Spain.

22. 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

Conference Proceedings

Journal Articles


RDCRN Descriptive Publications

Conference Proceedings

Journal Articles
3. Hampton T. Rare disease research gets boost. JAMA. Jun 28 2006;295(24):2836-2838. PMID: 16804140


## Advancing Research & Treatment for Frontotemporal Lobar Degeneration

### Journal Articles


Autonomic Rare Diseases Clinical Research Consortium

**Book Chapters**


Journal Articles


10. Raj SR, Black BK, Biaggioni I, Paranjape SY, Ramirez M, Dupont WD, Robertson D. Propranolol decreases tachycardia and improves symptoms in the postural tachycardia syndrome: less is more. *Circulation*. Sep 1 2009;120(9):725-734. PMID: 9687359, PMCID: PMC2758650


68. Coffin ST, Black BK, Biaggioni I, Paranjape SY, Orozco C, Black PW, Dupont WD, Robertson D, Raj SR. Desmopressin acutely decreases tachycardia and improves symptoms in the postural tachycardia syndrome. *Heart Rhythm.* May 3 2012. PMID: 22561596, PMCID: PMC3419341


**Brain Vascular Malformation Consortium**

**Book Chapters**


Abstracted Presented at Conferences
1. Khan Y, Hart B, Morrison L. CCM in Hispanic children in New Mexico Paper presented at: 2nd Conference on Clinical Research for Rare Diseases; September 21, 2010; Washington, DC.


Conference Proceedings


Journal Articles


43. Lo WD, Kumar R. Arterial Ischemic Stroke in Children and Young Adults. *Continuum (Minneapolis, Minn).* 2017;23(1, Cerebrovascular Disease):158-180. PMID: 28157749


Brittle Bone Disorders Consortium

Abstracts


Journal Articles


Clinical Research in ALS & Related Disorders for Therapeutic Development

Journal Articles


**Consortium of Eosinophilic Gastrointestinal Disease Researchers**

**Abstracts**


Book Chapters

Journal Articles

2. Cianferoni A, Spergel JM, Muir A. Recent advances in the pathological understanding of eosinophilic esophagitis. Expert review of gastroenterology & hepatology. 2015;9(12):1501-1510. PMID: 26470602, PMCID: PMC4943572


53. Philpott H, Dellon E. Histologic improvement after 6 weeks of dietary elimination for eosinophilic esophagitis may be insufficient to determine efficacy. Asia Pacific allergy. 2018;8(2):e20. PMID: 29732296, PMCID: PMC5931927


Developmental Synaptopathies Consortium

Journal Articles


**Dystonia Coalition**

**Abstracts Presented at Conferences**


4. Rosen A, Jinnah HA. An innovative infrastructure for common consortium challenges. Poster presented at: Conference on Clinical Research for Rare Diseases; September 21, 2010; Bethesda, MD.


17. Prudente C, Pardo C, Jinnah H. Neuropathology of primary cervical dystonia. Paper presented at: Division Students Advisory Council (DSAC) Symposium2012; Atlanta, GA.


**Journal Articles**


**Genetic Disorders of Mucociliary Clearance**

**Book Chapters**

Abstracts Presented at Conferences


Journal Articles


Inherited Neuropathies Consortium

Book Chapters


Abstracts Presented at Conferences


Conference Proceedings

Journal Articles


44. Sinclair CD, Morrow JM, Miranda MA, Davagnanam I, Cowley PC, Mehta H, Hanna MG, Koltzenburg M, Yousry TA, Reilly MM, Thornton JS. Skeletal muscle MRI magnetisation transfer


97. Lewis RA, McDermott MP, Herrmann DN, et al. High-Dosage Ascorbic Acid Treatment in Charcot-Marie-Tooth Disease Type 1A: Results of a Randomized, Double-Masked, Controlled Trial. JAMA neurology. Aug 1 2013;70(8):981-987. PMID: 23797954, PMCID: PMC3752369


134. Hershey LA, Perlmutter JS. Smoking and Parkinson disease: where there is smoke there may not be fire. *Neurology.* Oct 14 2014;83(16):1392-1393. PMID: 25217061


188. Panosyan FB, Mountain JM, Reilly MM, Shy ME, Herrmann DN. Rydel-Seiffer fork revisited: Beyond a simple case of black and white. *Neurology.* 2016;87(7):738-740. PMID: 27412138, PMCID: PMC4999169


Lysosomal Disease Network

Book Chapters


Abstracts Presented at Conferences


10. Schiffman R. The natural history of mucolipidosis type IV. Paper presented at: Lysosomal Disease Network's WORLD Symposium; February 8-10, 2012; San Diego, CA.


12. Polgreen LE, Miller BS. Preliminary data on the growth impact and safety of human growth hormone treatment in children with Hurler and Hunter syndromes. Poster presented at: Lysosomal Disease Network's WORLD Symposium; February 8-10, 2012; San Diego, CA.


17. Utz JR CT, Sorgen PJ, Whitley CB. Metabolomic study of CSF and serum markers in infantile and juvenile gangliosidosis diseases. Lysosomal Disease Network’s 10th annual WORLD Symposium; February 11-13, 2014; San Diego, CA.

18. Utz JR SP, Crutcher T, Whitley CB. Screening for CSF and serum biomarkers in infantile and juvenile gangliosidosis diseases. Lysosomal Disease Network’s 10th annual WORLD Symposium; February 11-13, 2014; San Diego, CA.

19. Utz JR WC. Eradication of inhibitors to enzyme replacement therapy in Hunter syndrome patient using non-cytotoxic, non-immunosuppressing regimen. Lysosomal Disease Network’s 10th annual WORLD Symposium; February 11-13, 2014; San Diego, CA.

20. JR U. Non-cytotoxic, non-immunosuppressing approach to tolerance induction during ERT treatment. FDA Workshop on Immune Responses to Enzyme Replacement Therapies: Role of Immune Tolerance Induction; June 9, 2014; Silver Spring, MD.

21. Utz JR CT, Schneider J, Whitley CB. Biomarkers of inflammation in CSF and serum of infantile and juvenile gangliosidoses. American College of Medical Genetics and Genomics Annual Meeting; March 24-28, 2015; Salt Lake City, UT.

22. Schneider J RK, Whitley CB, Utz JR. Prevalence of hypothyroidism in adult patients with Pompe disease. Lysosomal Disease Network’s 11th annual WORLD Symposium; February 10-12, 2015; Orlando, FL.

Conference Proceedings


**Journal Articles**


47. Adams HR, Mink JW. Neurobehavioral features and natural history of juvenile neuronal ceroid lipofuscinosis (Batten disease). *J. Child Neurol.* Sep 2013;28(9):1128-1136. PMID: 24014508, PMCID: PMC3976549


60. Prater SN, Banugaria SG, Morgan C, Sung CC, Rosenberg AS, Kishnani PS. Letter to the Editors: Concerning "CRIM-negative Pompe disease patients with satisfactory clinical outcomes on
enzyme replacement therapy" by Al Khallaf et al. *J. Inherit. Metab. Dis.* Jan 2014;37(1):141-143. PMID: 23887636, PMCID: PMC4353589


112. Shapiro EG, Whitley CB, Eisengart JB. Beneath the floor: re-analysis of neurodevelopmental outcomes in untreated Hurler syndrome. Orphanet J Rare Dis. 2018;13(1):76. PMID: 29751845, PMCID: PMC5948735

**Nephrotic Syndrome Rare Disease Clinical Research Network**

**Abstracts Presented at Conferences**


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.


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.


Journal Articles


**North American Mitochondrial Diseases Consortium**

**Journal Articles**


Porphyrias Consortium

Book Chapters


Abstracts Presented at Conferences

1. Anderson K. CYP1A2*1F and GSTM1 Alleles Are Associated with Susceptibility to Porphyria Cutanea Tarda. Poster presented at: International Porphyrins & Porphyrias Meeting; April 2011; Cardiff, Wales.


7. Desnick RJ. The Porphyrias: Cardinal Signs and Diagnosis/Treatment. Paper presented at: American College of Medical Genetics Annual Clinical Genetics Meeting; March 31, 2012; Charlotte, NC.


Conference Proceedings


Journal Articles


Special Projects


Primary Immune Deficiency Treatment Consortium

Journal Articles


89. Dietz AC, Duncan CN, Alter BP, et al. The Second Pediatric Blood and Marrow Transplant Consortium International Consensus Conference on Late Effects after Pediatric Hematopoietic Cell Transplantation: Defining the Unique Late Effects of Children Undergoing Hematopoietic Cell Transplantation for Immune Deficiencies, Inherited Marrow Failure Disorders, and
Rare Kidney Stone Consortium

Book Chapters

Abstracts Presented at Conferences


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.


27. Lieske J. Hypertension and Urolithiasis. Paper presented at: 12th International Symposium on Urolithiasis; May, 2012; Ouro Preto, Brazil.


33. Lieske J. Primary hyperoxaluria: not such a rare disease? Paper presented at: Research on Calculous Kinetics Society Session at the American Urological Association Meeting; May, May 2012; Atlanta, GA.


42. Milliner D. Crystal nephropathies and chameleons. Paper presented at: 8th Uruguayan Congress of Nephrology; September, 2012; Montevideo, Uruguay.


44. Edvardsson VO, Hardarson S, Palsson R. Renal histopathological findings in patients with 2,8-dihydroxyadeninuria. Paper presented at: European Society For Pediatric Nephrology; September, 2012; Krakow, Poland.

45. Edvardsson V, Agustsdottir I, Palsson R. Childhood presentation and renal outcome of APRT deficiency (2,8-Dihydroxyadeninuria) in Iceland. Paper presented at: European Society For Pediatric Nephrology, 45th Annual Meeting; September, 2012; Krakow, Poland.

47. Edvardsson V. Clinical features and long-term renal outcome of Islandic patients with APRT deficiency and 2,8-dihydroxyadeninuria. Paper presented at: 11th Congress of the European Society of Internal Medicine; October, 2012; Madrid, Spain.


Journal Articles


64. Lieske JC, Turner ST, Edeh SN, Ware EB, Kardia SL, Smith JA. Heritability of dietary traits that contribute to nephrolithiasis in a cohort of adult sibships. *Journal of nephrology*. Feb 2016;29(1):45-51. PMID: 25963767, PMCID: PMC4643420


**Rare Lung Diseases Consortium**

**Book Chapters**


**Journal Articles**


27. Young LR, Trapnell BC, Mandl KD, Swarr DT, Wambach JA, Blaisdell CJ. Accelerating Scientific Advancement for Pediatric Rare Lung Disease Research. Report from a National Institutes of


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

Book Chapters


Abstracts Presented at Conferences


Journal Articles


**Sterol and Isoprenoid Diseases Consortium**

**Book Chapters**


**Journal Articles**


Urea Cycle Disorders Consortium

Abstracts Presented at Conferences


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.


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.


Meeting of the Society for Inherited Metabolic Disorders; February 27 - March 2, 2011; Asilomar, CA.


22. Ludwig W, Ellenbogan A, Gropman A. MD Fiber Tracts in the Corpus Callosum Correlate with Scores on Behavioral Tasks and Glutamine Levels in Patients with OTCD, initial study (selected for travel award). Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; March 31 - April 2, 2012; Charlotte, NC.

23. Mokhtarani M, Diaz G, Rhead W, et al. Elevated Phenylacetic Acid (PAA) Levels Appear Linked to Neurological Adverse Events in Healthy Adults But Not in Urea Cycle Disorder (UCD)
Patients. Paper presented at: Society for Inherited Metabolic Disorders Annual Conference; March 31 - April 2, 2012; Charlotte, NC.


Conference Proceedings


Journal Articles


36. Gropman AL, Gertz B, Shattuck K, Kahn IL, Seltzer R, Krivitsky L, Van Meter J. Diffusion tensor imaging detects areas of abnormal white matter microstructure in patients with partial...


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75. Pacheco-Colon I, Friske S, VanMeter J, Gropman AL. Advances in urea cycle neuroimaging: Proceedings from the 4th International Symposium on urea cycle disorders, Barcelona, Spain,


**Vasculitis Clinical Research Consortium**

**Book Chapters**


**Abstracts Presented at Conferences**


Journal Articles


35. Herlyn K, Hellmich B, Seo P, Merkel PA. Patient-reported outcome assessment in vasculitis may provide important data and a unique perspective. *Arthritis Care Res (Hoboken).* Nov 2010;62(11):1639-1645. PMID: 20556814, PMCID: PMC3123033


64. Espy C, Morelle W, Kavian N, et al. Sialylation levels of anti-proteinase 3 antibodies are associated with the activity of granulomatosis with polyangiitis (Wegener's). *Arthritis Rheum.* Jul 2011;63(7):2105-2115. PMID: 21437874


**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**

**Book Chapters**


Abstracts Presented at Conferences


hybridization (CGH): Large segmental duplicons flank the breakpoints. Poster presentation Paper presented at: The American Society of Human Genetics meeting; October 9-13, 2006; New Orleans, LA.


31. Cassidy SB, McCandless SE, Driscoll DJ, Schwartz S. Do some people with severe Prader-Willi syndrome have two microdeletion syndromes? Paper presented at: David Smith Meeting; August, 2008; Greenwood, SC.


35. Butler MG, Kibiryeva N, Fischer W, Bittel DC. High resolution array comparative genomic hybridization (aCGH) in individuals with Prader-Willi syndrome. Paper presented at: KUMC Faculty Research Day and Poster Session; November 6, 2008; Kansas City, KS.


Journal Articles


72. Meng L, Person RE, Beaudet AL. Ube3a-ATS is an atypical RNA polymerase II transcript that represses the paternal expression of Ube3a. *Hum. Mol. Genet.* Jul 1 2012;21(13):3001-3012. PMID: 22493002, PMCID: PMC3465693


Special Projects


Bone Marrow Failure Consortium

Abstracts Presented at Conferences


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.


Journal Articles


**Cholestatic Liver Disease Consortium**

**Abstracts Presented at Conferences**


Conference Proceedings


Journal Articles


**Chronic Graft Versus Host Disease Consortium**

**Conference Proceedings**


Journal Articles


32. Muller JA, Zirafi O, Roan NR, Lee SJ, Munch J. Evaluation of EPI-X4 as a urinary peptide biomarker for diagnosis and prognosis of late acute GvHD. *Bone Marrow Transplant.* 2016;51(8):1137-1139. PMID: 27042833, PMCID: PMC4972659
Clinical Research Consortium for Spinocerebellar Ataxias

**Book Chapters**


**Abstracts Presented at Conferences**


**Journal Articles**


**Consortium for Clinical Investigation of Neurologic Channelopathies**

**Abstracts Presented at Conferences**


Journal Articles


**Rare Genetic Steroid Disorders**

**Journal Articles**


**Rare Thrombotic Diseases Consortium**

**Journal Articles**


Salivary Gland Carcinomas Consortium

Journal Articles


20. Li N, Xu L, Zhao H, El-Naggar AK, Sturgis EM. A comparison of the demographics, clinical features, and survival of patients with adenoid cystic carcinoma of major and minor salivary glands versus less common sites within the Surveillance, Epidemiology, and End Results registry. *Cancer.* Aug 15 2012;118(16):3945-3953. PMID: 22179977, PMCID: PMC3412946


39. Sandulache VC, Ow TJ, Daram SP, et al. Residual nodal disease in patients with advanced-stage oropharyngeal squamous cell carcinoma treated with definitive radiation therapy and


