Rare Diseases Clinical Research Network (RDCRN) Publications

Updated: April 1, 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

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.


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.


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


Journal Articles


**RDCRN Descriptive Publications**

**Conference Proceedings**


**Journal Articles**


**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


134. Wada N, Singer W, Gehrking TL, Sletten DM, Schmelzer JD, Low PA. Comparison of baroreflex sensitivity with a fall and rise in blood pressure induced by the Valsalva manoeuvre. *Clinical


**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


73. 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**


30. Shy ME. Inherited peripheral neuropathies. *Continuum (Minneapolis, Minn.).* Apr 2011;17(2 Neurogenetics):294-315. PMID: 22810821


32. Scherer SS. CMT2A: the name doesn't tell the whole story. *Neurology.* May 17 2011;76(20):1686-1687. PMID: 21508332


101. 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-Blinded, Controlled Trial. JAMA neurology. Aug 1 2013;70(8):981-987. PMID: 23797954, PMCID: PMC3752369


138. 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


256. Feifei T, Beecham G.W., Rebelo A, et al. Genome-Wide Association Study Identifies SIPA1L2 As A Genetic Modifier of Charcot Marie Tooth Disease Type 1A. Submitted

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**


72. Pena LD, Proia AD, Kishnani PS. Postmortem Findings and Clinical Correlates in Individuals with Infantile-Onset Pompe Disease. JIMD reports. 2015;23:45-54. PMID: 25763511, PMCID: PMC4484900


123. 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**


52. Schuetz C, Neven B, Dvorak CC, et al. SCID patients with ARTEMIS vs RAG deficiencies following HCT: increased risk of late toxicity in ARTEMIS-deficient SCID. *Blood.* Jan 9 2014;123(2):281-289. PMID: 24144642, PMCID: PMC3953035


95. Heimall J, Buckley RH, Puck J, et al. Recommendations for Screening and Management of Late Effects in Patients with Severe Combined Immunodeficiency after Allogenic Hematopoietic Cell Transplantation: A Consensus Statement from the Second Pediatric Blood and Marrow...
Transplant Consortium International Conference on Late Effects after Pediatric HCT. Biol Blood Marrow Transplant. 2017;23(8):1229-1240. PMID: 28479164, PMCID: PMC6015789


**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.


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**Journal Articles**


61. Thorsteinsdottir M, Thorsteinsdottir UA, Eiriksson FF, et al. Quantitative UPLC-MS/MS assay of urinary 2,8-dihydroxyadenine for diagnosis and management of adenine


**Rare Lung Diseases Consortium**

**Book Chapters**


**Journal Articles**


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.


(GPB). Paper presented at: American College of Medical Genetics Conference; March 27-31, 2012; Charlotte, NC.


22. Ludwig W, Ellenbogan A, Gropman A. MDFiber 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.


Conference Proceedings


Journal Articles


83. Caldovic L, Abdikarim I, Narain S, Tuchman M, Morizono H. Genotype-Phenotype Correlations in Ornithine Transcarbamylase Deficiency: A Mutation Update. *Journal of genetics and


**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


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 (aCHG) in individuals with Prader-Willi syndrome. Paper presented at: KUMC Faculty Research Day and Poster Session; November 6, 2008; Kansas City, KS.


Angelman syndrome with genotype-phenotype correlations. Paper presented at: American College of Medical Genetics Annual Clinical Meeting; March 27, 2009; Tampa, Florida.


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**


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.


Journal Articles


15. Jasek M, Gondek LP, Bejanyan N, Tiu R, Huh J, Theil KS, O'Keefe C, McDevitt MA, Maciejewski JP. TP53 mutations in myeloid malignancies are either homozygous or hemizygous due to copy

197


**Cholestatic Liver Disease Consortium**

**Abstracts Presented at Conferences**


karyotyping. Paper presented at: American Society of Hematology; December 8-11, 2007; Atlanta, GA.


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**


**Journal Articles**


**Chronic Graft Versus Host Disease Consortium**

**Conference Proceedings**


**Journal Articles**


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


symptoms and signs of myotonia in nondystrophic myotonia: a randomized controlled trial. JAMA. Oct 3 2012;308(13):1357-1365. PMID: 23032552, PMCID: PMC3564227


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


