Rare Diseases Clinical Research Network (RDCRN) Publications

Updated: January 8, 2019

RDCRN Data Management and Coordinating Center Publications

DMCC Publications

RDCRN Descriptive Publications

RDCRN Publications

RDCRN Publications, Current Grant Cycle Consoritia

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**

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

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

4. Thompon P. On the trail of rare disease: investigators and advocates hunt for collaboration and 

Merkel PA. Clinical research for rare disease: opportunities, challenges, and solutions. Mol. 

6. 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 

7. Rubinstein YR, Groft SC, Bartek R, Brown K, Christensen RA, Collier E, Farber A, Farmer J, 
Ferguson JH, Forrest CB, Lockhart NC, McCurdy KR, Moore H, Pollen GB, Richesson R, Miller VR, 
Hull S, Vaught J. Creating a global rare disease patient registry linked to a rare diseases 
2010;31(5):394-404. PMID: 20609392, PMCID: PMC2930109

8. Richesson RL, Sutphen R, Shereff D, Krischer JP. The Rare Diseases Clinical Research Network 
656. PMID: 22405970, PMCID: PMC3652679

9. Groft SC, Gopal-Srivastava R. A model for collaborative clinical research in rare diseases: 
experience from the Rare Disease Clinical Research Network program. J Clin 
Invest. 2013;3(11):1015–21

2014;311(17):1729-1730. PMID: 24794360

Network's Organization and Approach to Observational Research and Health Outcomes 
PMC4124127

and clinical investigators in the rare diseases clinical research network. Orphanet J. Rare Dis. 
2016;11(1):66. PMID: 27194034, PMCID: PMC4870759

Consortium of Eosinophilic Gastrointestinal Disease Researchers (CEGIR). Translational science 
of rare diseases. 2017;2(3-4):141-155. PMID: 29333363, PMCID: PMC5757645
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


134. Wada N, Singer W, Gehring 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**


64. 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.


at: The Voice Foundation Annual Symposium: Care of the Professional Voice; June 5, 2011; Philadelphia, PA.


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**


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

Journal Articles


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


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.


**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


118. 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


#904 presented at the Annual Meeting of the American Association for the Study of Liver Diseases. *Hepatology*. 2011;54(S1):785A.

## Journal Articles


13. Boynton TO, Gerdes S, Craven SH, Neidle EL, Phillips JD, Dailey HA. Discovery of a gene involved in a third bacterial protoporphyrinogen oxidase activity through comparative genomic analysis


**Special Projects**


**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; Reykjavik, 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 Icelandic 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 Disease Consortium**

**Book Chapters**


**Journal Articles**


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**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.


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


52. Fike CD, Sidoryk-Wegrzynowicz M, Aschner M, Summar M, Prince LS, Cunningham G, Kaplowitz M, Zhang Y, Aschner JL. Prolonged hypoxia augments L-citrulline transport by system A in the


**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.


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.

Driscoll D. Prader-Willi syndrome, genetics of obesity section. Paper presented at: American College of Medical Genetics Meeting; March 25-29, 2009; Tampa, FL.


Journal Articles


49. Sahoo T, del Gaudio D, German JR, Shinawi M, Peters SU, Person RE, Garnica A, Cheung SW, Beaudet AL. Prader-Willi phenotype caused by paternal deficiency for the HBII-85 C/D box


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**


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; 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.


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.


Journal Articles


**Cholestatic Liver Disease Consortium**

**Abstracts Presented at Conferences**


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.


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


**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


