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


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


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


5. Petersen TA, Morrison LA, Schrader RM, Hart BL. Familial versus sporadic cavernous malformations: differences in developmental venous anomaly association and lesion


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**


29. Collins MH, Martin LJ, Alexander ES, et al. Newly developed and validated eosinophilic esophagitis histology scoring system and evidence that it outperforms peak eosinophil count


75. 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


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-Masked, 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


260. 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**


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.


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.


of Clinical Chemistry & International Congress of Porphyrins and Porphyrias; May 16-18, 2013; Lucerne, Switzerland.


Conference Proceedings


2. Hwang SI, Lee YY, Park JO, Norton HJ, Clemens E, Schrum LW, Bonkovsky HL. The measurement of hepcidin from human urine and serum to study effects of a single dose of oral iron by an
optimized LC-MS/MS method. [Abstract #1204 presented at Annual Meeting of the American Association for the Study of Liver Diseases]. *Hepatology.* 2011;54(S1):931A.


**Journal Articles**


Special Projects


**Primary Immune Deficiency Treatment Consortium**

**Journal Articles**


6. Patel NC, Chinen J, Rosenblatt HM, et al. Outcomes of patients with severe combined immunodeficiency treated with hematopoietic stem cell transplantation with and without...


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


**Rare Kidney Stone Consortium**

**Book Chapters**


Abstracts Presented at Conferences


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.

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.

patients: a retrospective analysis. American Society of Nephrology (ASN) Kidney Week. San
Diego, CA2012.

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


Week. San Diego, CA2012.

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

calcification in primary hyperoxaluria. American Society of Nephrology (ASN) Kidney Week. San
Diego2012.

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.

26. Goldfarb D. Cystinuria. Paper presented at: 12th International Symposium on Urolithiasis; May,
2012; Ouro Preto, Brazil.

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

2,8-dihydroxyadenine with liquid chromatography - electrospray tandem mass spectrometry
May, 2012; Paris, France.

29. Cohen J, Goldfarb DS, Grasso M. Management of cystinuric patients in a dedicated stone clinic
decreases stone events. Paper presented at: Annual Meeting of the American Urological
Association; May, 2012; Atlanta, GA.


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


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**


5. Trotta BM, Stolin AV, Williams MB, Gay SB, Brody AS, Altes TA. Characterization of the relation between CT technical parameters and accuracy of quantification of lung attenuation on


**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.


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.


Clinical features in 102 patients with Angelman syndrome. Paper presented at: American College of Medical Genetics 2009 Annual Clinical Meeting; March 27, 2009; Tampa, Florida.


Journal Articles


41. Sahoo T, Bacino CA, German JR, Shaw CA, Bird LM, Kimonis V, Anselm I, Waisbren S, Beaudet AL, Peters SU. Identification of novel deletions of 15q11q13 in Angelman syndrome by array-


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**


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


secondary outcomes show improvement in symptoms and signs of myotonia. Paper presented at: American Academy of Neurology; April 26, 2012; New Orleans, LA.

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


