Rare Diseases Clinical Research Network (RDCRN) Publications

Updated: August 1, 2016

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
Chronic Graft Versus Host Disease 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 Hannah Fitterman at Hannah.Fitterman@epi.usf.edu
**RDCRN Publications, Previous Grant Cycle Consortia**

- **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**
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.


Conference Proceedings


Journal Articles


**RDCRN Descriptive Publications**

**Journal Articles**


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**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**


**Brittle Bone Disorders Consortium**

**Journal Articles**


**Chronic Graft Versus Host Disease Consortium**

**Journal Articles**


Clinical Research in ALS & Related Disorders for Therapeutic Development

Journal Articles


**Consortium of Eosinophilic Gastrointestinal Disease Researchers**

**Journal Articles**


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


96. 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


132. 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


**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.


Conference Proceedings


Journal Articles


73. 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


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.


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


Conference Proceedings


Journal Articles


Special Projects


Primary Immune Deficiency Treatment Consortium

Journal Articles


**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 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


**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


Vasculitis Clinical Research Consortium

Book Chapters


Abstracts Presented at Conferences


Journal Articles


16. Mahr AD, Neogi T, Lavalley MP, Davis JC, Hoffman GS, McCune WJ, Specks U, Spiera RF, St Clair EW, Stone JH, Merkel PA. Assessment of the item selection and weighting in the Birmingham...


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


**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


**Cholestatic Liver Disease Consortium**

**Abstracts Presented at Conferences**


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


**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


