

**CTSA Consortium
Child Health
Oversight Committee
(CC-CHOC)**

Rare Disease Workgroup

May 1st, 2009

Bob Steiner, Nancy Green, Jennifer Puck

CC-CHOC Rare Disease Work Group

- **Genesis:**
 - Discussion between Jennifer Puck & Bob Steiner at 1st CTSA POC meeting
 - CC-CHOC members voted this a high priority initiative
- **Activities:**
 - Define goals, mission
 - Teleconference with NIH CTSA Program staff
 - Teleconference with external rare disease experts 1/2008
 - Define Workgroup mission/vision based on input from rare disease experts and CC-CHOC members
- **Potential for Collaboration**
 - NIH Rare Disease Clinical Research Network
 - NIH Office of Rare Diseases
 - FDA Office of Orphan Products Development
 - NHGRI, NORD, Genetic Alliance, NBSTRN/ACMG, etc.

CC-CHOC RDWG Mission Statement

The Workgroup aims to serve as a platform to facilitate the systematic study of patients with rare diseases. Multicenter collaborations are essential to achieve sufficient cohorts and to harness research resources to make significant findings. The workgroup will outline pressing rare disease research issues and identify hurdles and opportunities to overcome them via expertise and resources from across the CTSA Consortium as well as from federal and other entities.

CC-CHOC Rare Disease Workgroup

- Membership
 - Volunteers from CTSA sites (CC-CHOC members and others)
- Objectives
 - Identify gaps in systematic study of rare diseases, and how CTSA program might help
- Challenges/Issues
 - Inadequate patient numbers at individual sites
 - Many rare diseases not covered in existing networks
 - Infrastructure needed to organize multi-center efforts
 - Limited number of both current and future investigators
 - Need for multi-center training mechanisms

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- Challenges/Issues (continued)
 - Incompatible mechanisms for data collection, encoding, storage, AE reporting
 - Biorepositories exist, but little coordinated information about rare disease specimen collections (when we began)
 - How to involve industry partners
- Goals
 - Platform for communication, collaborative research, education and training
 - Platform for providing resources to RD researchers, trainees

CC-CHOC Rare Disease Workgroup

The top 5 reasons to join our group:

- Your calendar has an opening the afternoon of the 4th Monday every month allowing you to join our teleconferences
- You're tired of common disease research and looking for a new challenge
- You've heard about a proposed 5 yr doubling of NIH funding for rare disease research
- You don't have enough committee work
- If you don't join at least one of these workgroups, you may be nominated for CC-CHOC Chair