

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

**Rare Disease Workgroup- Update**

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# Rare Disease Workgroup: Update

## Co-chairs: Bob Steiner (OSU) and Nancy Green (Columbia)

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- **Genesis:**

- Discussion at 1<sup>st</sup> CTSA POC meeting
- CC-CHOC members voted this a high priority initiative

- **Potential for Collaboration**

- NIH Rare Disease Clinical Research Network
- NIH Office of Rare Diseases
- FDA Office of Orphan Products Development
- NHGRI, NORD, NBSTRN/ACMG, Genetic Alliance, others

# Rare Disease Workgroup

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- **Membership**

- Volunteers from CTSA sites (CC-CHOC members and others),
- Federal partners (staff from NCRR, NICHD, ORD, others)

- **Goals**

- Platform for communication, collaborative research, education and training
- Platform for providing resources to RD researchers and trainees

- **Challenges/Issues**

- Inadequate patient numbers at individual sites
- Many rare diseases not covered in existing networks
- Infrastructure needed to organize multi-center efforts
- Biorepositories exist, but little coordinated information
- Limited number of both current and future investigators
- Need for multi-center training mechanisms
- Coordinating functions with the multiple other RD groups

# Rare Disease Workgroup

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## Activities to date:

- Define goals, mission based on input from rare disease experts and CC-CHOC members
- Develop relationships to other child and adult rare disease groups, both research and advocacy/patient
- Presentation at 2009 PAS/SPR meeting, with on-going plans for activities at next year's meeting
- Project on Distributed Biobanks for Rare Diseases (DBRD)
- Peter Merkel (BU) leading an R13 application through NINDS for a training day in rare disease research (funding decision pending)
- Nascent virtual library of relevant documents and resources (e.g. protocols, consent forms, other)

# Rare Disease Workgroup

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- Great potential among CC-CHOC, other CTSA groups and expertise, as well as partnerships with other research and patient groups/activities
- Off to a great start!
- Inviting new members from existing and new CTSAs.
- Due for change of leadership – inviting nominations to chair or co-chair this group. Please nominate yourself or confirm willingness of your chosen nominee prior to submitting name, contact info and a 2-sentence statement of interests to Farrell Bowen, [bowen\\_farrell@bah.com](mailto:bowen_farrell@bah.com) by Sept. 8, 2009.