

**CTSA Consortium Child Health Oversight Committee (CC-CHOC)
CC-CHOC Rare Diseases Workgroup Teleconference
July 27, 2009
4:30 PM – 5:30 PM ET**

Co-Chairs: Bob Steiner and Nancy Green

Conference Number: 1-866-285-7778

Participant Code: 7334287

Agenda

I. Advancing Rare Disease Research meeting NIH July 17, 2009

Much synergy between CTSA, CC-CHOC RDWG and RDCRN and other groups. How do we formalize a relationship? What should we do together?

II. Ideas for Face-to-Face Meeting in conjunction with (PAS) Meetings May '09

PAS meeting and CC-CHOC: role of RDWG? Decide what we should do and communicate this to Bill Hay. Should we try to have our own research presentations or educational symposium? Please send us concrete examples of use of CTSA's in Rare Disease Research.

III. Brief update on 2010 ORD/NCRR RD Meeting: ORD, NCRR, and the RDCRN are in the early stages of planning a meeting that will engage the many communities in rare diseases research (NIH ICs; various rare diseases networks; resources, like CTSA's; patient advocacy groups; industry; investigators; clinicians; and others) to extend and develop frameworks for collaboration and synergies in rare diseases research. RDWG played a role in development of the R13 conference grant written by Peter Merkel, submitted to fund that conference.

IV. Continue to Determine Work Group priorities for rare disease research (RDR):

- Develop a virtual repository of relevant RDR resources: Work in progress. RD toolkit/reference library. These could be existing successful documents for a variety of needs: generic consent and related human subject research (e.g. Cornell documents and UAB portal-linked documents, Harvard/Boston Children's documents), go-to resources for database searches such as CRISP, GeneTests, ClinicalTrials.gov, call for participants for open clinical studies and registries, other items?
- Identify ourselves (or a sub-group) as being available for scientific consultation for RDR – as a “social network” of expert RD investigators. Potentially work with NIH RDCRN. Website. Especially to help junior investigators interested in RDR.
- Additional ideas?

V. Ideas for possible products/abstracts/publications/grants from the group: White paper on barriers or bottlenecks that are slowing down translational research with rare diseases, others. Address bias by grant, manuscript reviewers against rare disease research (low impact, etc.)

VI. Old business: Biorepositories: Rand Corporation may be working on inventorying biorepositories. Contact person Elyes Iseman? Rashmi or Jennifer may know. CETT program also has database of repositories

List of RD researchers: NDRI has one. ORDR may have one or could help us? FDA orhapn products division? CRISP? Clincial trials.gov? We should survey our own sites to identify additional.

Toolkit: OHSU developed a rare disease website. This could be worthwhile to do at other sites. Could our toolkit appear on ORDR website? How can rare disease researchers benefit from using resources of CTSA's?

Request update on Challenge Grants related to rare disease, ask Rashmi.

Training. Should we be ready to direct trainees to their local RD expert if they contact us, match up mentors? Have to develop our toolkit more. Maybe develop list of potential RDR mentors.

Question: Should we invite members to our WG who from non-CTSA sites but who want to participate? We think it's a good idea

VII. New business

VIII. Action items for next meeting