

CTSA Consortium  
Child Health  
Oversight Committee  
(CC-CHOC)

Rare Disease Workgroup

May 1<sup>st</sup>, 2009

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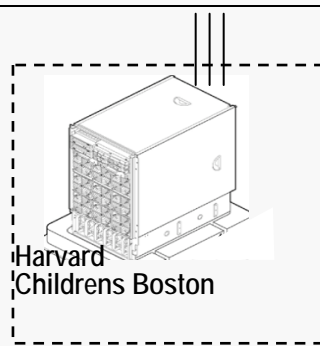
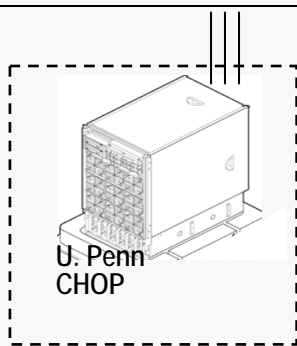
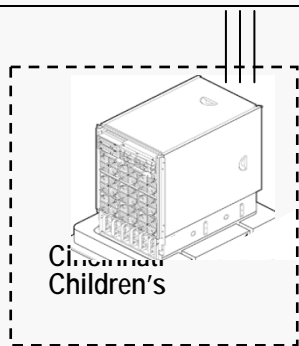
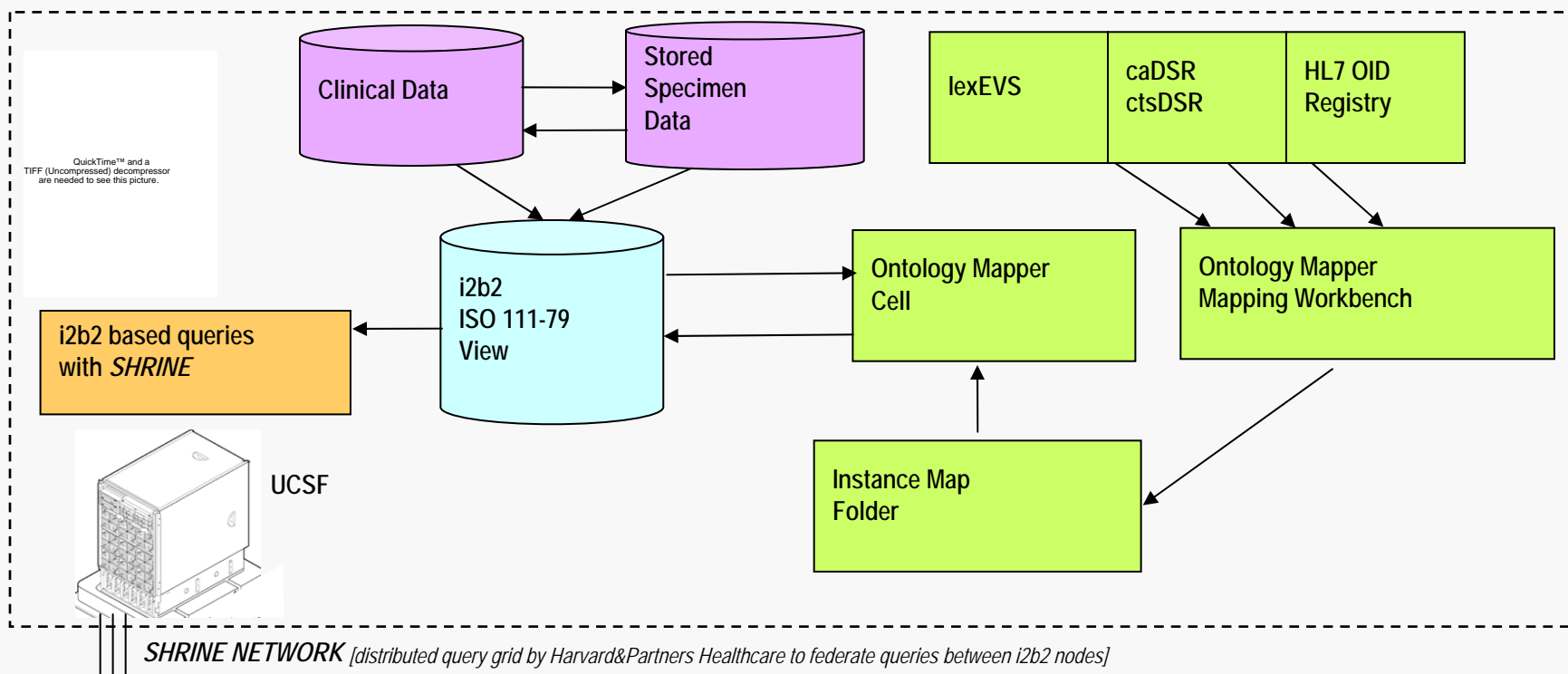
## CC-CHOC RDWG: Accomplishments

- Planned Educational/Training Symposium
  - RDWG members worked with NIH RDCRN investigators on conference grant application, development of agenda for combined 2010 RDCRN/ORD/CTSA Rare Disease Symposium
- Collaboration with NICHD-funded Newborn Screening Translational Research Network (NBSTRN)
  - RDWG members serve on planning/steering committees
- RDCRN applications included CTSA infrastructure as critical strength/resources
- Successful Grant Application: Administrative Supplement for Rare Disease Biobanking

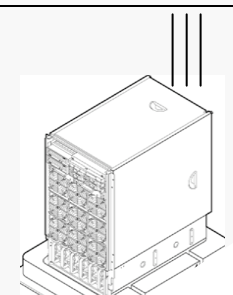
# CC-CHOC RDWG: Distributed Biobanks for Rare Diseases, RDWG CTSA Admin Supplement, 2008-9

- **Governance**
  - Biobank best practices, SOPs for sample handling and tracking
  - Legal, ownership issues
  - Human subjects issues (archived, prospective collections)
- **Rare Disease Use Cases**
  - SCID, Down syndrome; science determines what is collected
- **Informatics**
  - Collaborate with CTSA-wide Informatics Program
  - 2-step process
    - Initial de-identified query to find appropriate samples
    - Collaborations between sample holders and investigators

# DBRD Informatics, SCID Project



Additional Future Sites,  
USIDNET, CIBMTR



# CC-CHOC RDWG Accomplishments: CTSA RDWG and NBSTRN: Strong Partnership

- CC-CHOC RDWG and NBSTRN

RDWG and ACMG submitted responses to NICHD request for capability statements for NBSTRN published Dec. 7, 2007

Newborn Screening Translational Research Network Coordinating Center.

<http://www2.fbo.gov/spg/HHS/NIH/NICHD/Reference%2DNumber%2DNICHD%2DMRDD%2DNewborn/SynopsisR.html>

RDWG and ACMG Goals very similar

ACMG and CTSA CC-CHOC RDWG strongly prefer to collaborate to achieve common goals

Great opportunities for synergy/collaboration

We can achieve more working together

# CC-CHOC: SGC & KFC Mapping

- Proposed connections to Strategic Goal Committees (SGC) and Key Function Committees (KFC)
  - SGC1 : National Clinical and Translational Research Capability – Reference Library/Toolkit: sharing IRB/HSP documents
  - SGC 2: Training/Career Development of Clinical Translational Scientists – R13 conference grant application for rare disease research day targeted at junior investigators/trainees

## CC-CHOC: SGC & KFC Mapping (2)

- Proposed connections to Strategic Goal Committees (SGC) and Key Function Committees (KFC)
  - Clinical Research Management Key Function Committee - building networks of rare disease researchers
  - Education and Career Development Key Function Committee – 2010 Rare Disease conference

## CC-CHOC: SGC & KFC Mapping (3)

- Proposed connections to Strategic Goal Committees (SGC) and Key Function Committees (KFC)
- Biobanking Project maps to:
  - SGC3: Consortium-Wide Collaborations, part b, Data sharing network
  - Informatics Key Function Committee

## CC-CHOC RDWG: Current Initiatives

- Reference library/toolkit
  - Example: Rare Disease IRB protocols, consent forms
- Central Pre-Review IRB submissions
- Administrative Supplement for Rare Disease Biobanking

# CC-CHOC RDWG Goals

- Continue to identify gaps in systematic study of rare diseases, and how CTSA's might help
- Identify existing rare disease research networks and opportunities to expand
- When possible, develop infrastructure to organize multi-center efforts in rare disease research
- Explore mechanisms for training future generation of rare disease investigators
- Work towards common mechanisms for data collection, encoding, biospecimen storage, AE reporting for rare diseases (e.g. NBSTRN & CaBIG, CTSA administrative supplement)

## CC-CHOC RDWG Goals (2)

- Complete planning for 2010 Rare Disease Educational/Training Symposium and jointly hold the conference
- Continue to build the reference library/toolkit
- Build mechanism for central pre-review of planned IRB submissions