

Day two-breakout session A

Chair: Rachel Richesson & Clem McDonald

A. Standardized vocabulary, terminology, CDE's, and diagnosis Questions and topics to cover:

Objectives for breakout session:

The primary objective of this session will be to clearly articulate (and rank?) the motivations, objectives, and requirements for data standards in the context of a multi-disease registry platform in rare disease context. This will then provide a basis of consensus with which to evaluate, endorse, and promote standards in rare disease registries. Once there is agreement on a statement of standards purposes, the discussion can move toward identifying the standards requirements and available options and gaps, and toward identifying a strategy for naming, evaluating, and promulgating standards. At the end of the 3 hour discussion, the group should have a list of outstanding needs (i.e., gaps in standards and missing tools & technologies), and a timeline for future collaboration & consensus-building to address those needs.

- 1.) Specify the *motivation* for standards for a rare disease registry (e.g., data sharing across registries, interoperability with EHR/PHR, question/system re-use across registries, “regulatory sufficiency”, etc.) I think the rare disease registry ‘community’ should make this explicit and consensual.
- 2.) Specify more specific *requirements* for standards (e.g., clinical definitions, demonstrated use, multi-lingual, organizational features, cost, access, etc.) – will look at standards criteria from caBIG, HITSP, HL7. This discussion topic can also include the *types of data collected* in rare disease registries (e.g., diagnosis info., family history, medical history, exposures, quality of life).
- 3.) Identify possible *approaches* to standards (e.g., Common Data Elements, structural features for disease specific items) in a multi-disease registry, and identify needed tools (e.g., metadata repository/question library, guidance on best practice for question development) for each.
 - a.) If time permits and the audience has the right people, then a standards strategy should be selected...
 - b.) Include provision in strategy for including HITSP and U.S. Federal Government standards as they evolve....
- 4.) Characterize different *types of standards* (e.g., form standards, question standards, terminology, etc.) and list the options that currently exist for each. This will enable a statement on ‘standards readiness’ for rare disease registries (e.g., all standards available, need standard for xxx, etc.)

5.) *Identify gaps* where standards do not exist, and tools that might be needed to access them or incorporate them in registries.

6.) Determine a process for moving forward, including agreements for future communication (i.e., meetings/activities), collaboration, governance (?), and consensus-building.



Uniting Rare Diseases



Breakout Session A **Standards Vocabulary Terminologies, CDEs and Diagnosis**

Outline of Summary

- Topics that were discussed during the session *(many & varied)*
- Points of concerns and questions *(themes & points of heavy discussion)*
 - Challenges
 - Goals for standards
- Suggestions and recommendations *(from panel experts and participants)*
 - Resources Needed
 - Recommendations

Topics Discussed

- How can we use existing standards? They are often difficult for people to use and understand
 - Is there a standard for xxx?
 - Does NIH have standard questions for my disease? Do others have questions? What are the standards?
 - How do I use HL7? What is the ‘map’ from EHR system xxx to my data?
- How do people find out where these standards exists/use?

Topics Discussed

- How do we handle multi-lingual?
- How can we use EHR / clinical data?
- How to confirm what diagnosis the person has? Data quality issues?
- Can we link registries to public health programs?
- Is there a master plan or clearinghouse resource?

Points of Concern & Questions

*(themes & points of
heavy discussion)*

- Challenges
- Goals for Standards

Challenges

- Lack of shared objectives
 - Multiple starting points
 - Various data points and sources
 - Lots of disease-specific data
- Standards don't necessarily include rare disease concepts
- Multiple, overlapping and complicated standards
- Confirmation/verification of data
 - how do you verify diagnosis?

Goals for Standards

- Facilitate use of EHR / clinical data for registries
- Bridge between multiple registries
- Link to biorepositories
- Provide standard questions for groups to share
 - e.g., by organ site
- Enable a generalized Contact registry (could be sharable resource)
- Link to public health programs

Resources Needed

- Repository of information and standards (user friendly)
 - Guidance on implementation
- Rare disease representation in various standards
- Interchange format standards
- Evidence base of questions and data elements that be used by registries
- Relevant resource guidance
 - LOINC, PROMIS, NeuroQual, others
- Lists of existing repositories of this information that is already put together

Resources Needed

- Need to have a standard set of questions, maybe phrased in different manners based on respondent
- Standard question structure is needed
- Specificity consensus (e.g., how detailed should questions be?)
- Consensus on standards “areas” (buckets)
- Consent/patient preference standards
- Build forms quickly: re-use questions
- Tools

Recommendations – What we need to do:

1. Standardize questions!!
2. Find commonalities in data collection across rare diseases to enable #1.
3. Provide guidance (to entire rare diseases research and advocacy community) on standards for structuring and representing questions.
4. Find a centralized “keeper” of the questions.

Recommendations – What we need to do:

5. Define a Minimal Common Registry Model (MCRM) – i.e., sharable questions that would enable a rare disease contact registry
6. Get active and piggyback on efforts for Electronic Health Record standardization (and watch HITSP's Clinical Research Use Case as well as others....)