

Day two-session D Informatics and technology

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Session Overview

Families, clinicians, industry, advocacy organizations and counselor requirements must all be considered when developing and prioritizing technical functionality for national rare disease registries. This session will discuss major user needs for each of these stakeholder organizations.

Understanding user needs and goals in advance of any technology decision-making is critical. This is not a 'techie' session, but a discussion of stakeholder requirements that will guide technology, on-going support models and funding models.

Goals

- Identify primary stakeholders and goals/objectives of each
- Identify major functionality to address the goals/objectives of each stakeholder group

Questions & Topics for Discussion

- Identify primary stakeholders & functionality/objectives required for each
- Facilitating multiple sponsoring organizations, existing registries in a single disease area
- Facilitating patient contact requests
- Data aggregation versus data capture systems
- Duplicate registrations, defining a unique patient identifier
- Multi-lingual, international requirements
- Should we allow derivative works from the de-identified patient data
- How do we implement these registries in an affordable, scalable, repeatable manner
- Providing technical support, upgrades, advanced functionality and customizations
- Additional questions/discussions as time permits

Uniting Rare Diseases



Breakout session D Technology and Informatics

Discussion Topics

- Identify primary stakeholders & functionality / objectives for each
 - Patients, Researchers, Foundations/Gov't, Clinicians/Curators, Industry
- What are the minimum data elements required
- Facilitating multiple sponsoring organizations in a given disease area
- How to migrate/integrate existing patient registry data
- How to handle duplicate registrations, single person, multiple disorders
- How to connect patient registries with biorepositories
- How do we implement registries in an affordable, scalable, repeatable manner
- Mutli-lingual requirements

Points of Concerns & Questions

- How to interface/work with EHR's, specifically EPIC?
- If provide opensource software tools users can download & modify, who would provide support?
- There's a big difference between patient self-report vs clinician entered vs Electronic Health Record extraction - not a 1 size fits all solution
- Hosted solutions are attractive, but who would fund / support / develop on-going?
- How to drive down the costs of implementation and reduce the number of custom build registries
- How to aggregate/consolidate data from existing registries

Suggestions and Recommendations

- Opensource software / hosted solutions
 - Allow users to access registry software via a hosted solution for smaller organizations that don't have IT support
 - Provide download of tools and technologies with configurable 'modules' of functionality
- Center / cooperative method of curation & validation
 - Need the ability to curate / validate the data if patient self-report, but data entry and curation is expensive.
 - Consider support of a loosely federated global curators to achieve economies of scale
- Central wiki / website for resources, best practices
 - Implement a wiki or website where registry developers can share best practices, learn from other's experiences, implementation guides, registry vendors, downloadable tools
 - Provide a listing of what registries currently exist with links, # of registered users, technologies used
- Any technology solution must be multi-lingual
 - Store all recorded data in english for researchers to use, but data entry / capture must allow toggling between various languages

Suggestions and Recommendations

- Develop a 'contact registry' ala ResearchMatch
 - Lightweight minimum data set approach
 - Consumer driven, name, contact info, diagnosis, do you want to be contacted for study opportunities
 - Unique patient ID could be generated from this system, other registries could check this system through an open interface to see if there is already a patient identifier to reduce duplicate registrations
- Need for standardized way to share data
 - Registry to registry (eg Immunodeficiency disease vs bone marrow registry)
 - Electronic Health Record to registry
 - Sharing with public databases - provide data to CA-BIG?
- Common repository of questions, answers & data elements
 - How to agree on questions, commonality
 - Multiple sources of potential data elements, reconcile and provide a comprehensive, federated view
 - Provide listing of resources/links to existing standards