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BRIEF REPORT

Landscape Analysis of Registries and Biobanks: A Tool for Disease Advocacy Organizations to Enhance Translational Research Systems

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Disease advocacy organizations play an increased role in the development and management of registries and biorepositories. Genetic Alliance developed criteria to assist organizations in selecting vendors and conducted a technical assessment survey of potential vendors. More than half offered customizable solutions, and biorepositories surveyed offered a variety of genomic services. Nearly three-quarters collect deidentified data, and few use controlled vocabulary, limiting the ability to recontact participants and share data. While this does not represent the complete registry and biorepository landscape, it is a starting point to assist organizations in assessing appropriate solutions.

Introduction

Disease advocacy organizations (DAO) play a significant role in accelerating translation of basic science into tests and treatments.[1,2] Registries and biorepositories/biobanks are an important tool in this endeavor.[3] Many DAOs are engaged in the management of registries and biorepositories and frequently rely on outside vendors to provide these services. Best practices have been developed by a variety of organizations, including the RAND Corporation,[4] the International Society for Biological and Environmental Repositories (ISBER),[5] and NCI OBBR,[6] but these resources may not take into account the specific needs of advocacy organizations. We developed criteria that may be useful to assess registry and repository vendors and conducted a technical assessment to help DAOs identify appropriate, customizable, extensible, and scalable solutions. For this exercise, registries were defined as those that collect only clinical data, while biorepositories were defined as those that collect biological samples with or without robust, associated clinical information.

Materials and Methods

A vendor survey was developed in partnership with Health Advances, a health care consulting agency. The online survey, comprised of 108 questions, was fielded from May 11, 2009–June 10, 2009. Fifty-one registries and biorepositories were emailed, and a general invitation was sent to the ISBER listserv, a professional organization for biobanking. A scoring system was developed where points were awarded for criteria deemed important for DAO-initiated registries and biorepositories, and responses were evaluated for registry capacity, biorepository capacity, scalability, quality of information, and ability to adapt to future legal standards (Table 1).

Results

Twenty-nine organizations responded to the survey. Of these, 11 were responses to the invitation, and the remaining responses were volunteers from the post to the ISBER listserv. Of the 29 vendors, 13 were for-profit or commercial operations, 10 non-profit organizations, and 6 were academic institutions. Of the 22 organizations with complete responses, 4 reported only registry capabilities, 14 reported only biorepository capabilities, and 4 reported both registry and biorepository capabilities.

Scalability and data/sample collection

Many vendors provide scalable systems for data and samples, including customizable solutions for specific diseases or research projects (55%), customized data collection for specific disease needs (73%), and biorepositories that can grow to accommodate multiple diseases (78%). Vendors also have the capacity to store demographic (73%) and longitudinal data (55%). However, only 31% use any controlled language or messaging system, with SNOMED-CT and HL7 being the most common. Collection of data and samples is frequently deidentified (73%) and sometimes anonymous (36%). In some instances, donors can be relinked to samples (41%) or recontacted (23%).

Genetic Alliance, Washington, DC.

Table 1. Criteria and Scoring Used for Vendor Assessment

Criteria [maximum score]	*Data Element [score]*
Type [4 points]	Registry* [3] Biobank** [1]
Scalability [3 points]	Biobank can grow to accommodate multiple diseases** [1] Collection can be customized for specific disease and/or research project needs [1] Can function in disease specific clinics [1]
Data [5 points]	Stores demographic data [1] Stores longitudinal data [1] Data can be customized for disease specific needs [1] Can import EMR/PHR data* [1] Can export EMR/PHR data [1]
Data Standards [7 points]	HL7 [1] LOINC [1] ICD9/ICD10 [2] SNOMED-CT [3]
Data Linkage [4 points]	Stores deidentified data [1] Stores anonymized data [1] Data can be relinked [1] Participants can be re-contacted [1]
Samples [5 points]	Stores physical samples** [1] Extracts DNA from whole blood** [1] Grows cell lines** [1] Sections tissue** [1] Performs pathology services** [1]
Quality Control (QC) [5 points]	Automatic QC [1] Human QC [1] Researchers can search sample database** [1] Audit trails capture all access [1] Solution is HIPAA compliant [1]
Institutional Review Board (IRB) [1 point]	Functions under one IRB [1] Functions under multiple IRBs [1]

Total scores: registry = 26, biorepository = 30, combined registry and biorepository = 34.
*Registry only; **Biobank/biorepository only.

Sample processing

Of the biorepository vendors surveyed, 83% store physical samples. Many offer a variety of services, including extracting DNA from whole blood (56%), growing cell lines (44%), sectioning tissue (61%), and performing pathology services (56%).

Quality control and compliance

Many vendors use human quality control procedures (73%), but 18% have automatic quality control procedures in place, and 61% have audit trails to capture all access. Vendors function under multiple IRBs (50%) or a single IRB (50%), but the majority (86%) are HIPAA compliant.

Scoring

Vendors scoring $\geq$50% of selected criteria include the following:

- For registry functionality: Emerge.MD, Genetic Alliance BioBank, Irody, Inc., Sidney Kimmel Cancer Center, UBC-James Hogg iCAPTURE Centre, WellCentive;
- For biorepository functionality: Biobanque de Picardie, Cooperative Human Tissue Network Eastern Division, Fred Hutchinson Cancer Research Center, Genetic Alliance BioBank, M.D. Anderson Cancer Center, Methodist Research Institute, Sidney Kimmel Cancer Center, UBC-James Hogg iCAPTURE Centre, University of New Mexico School of Medicine;
- For both registry and biorepository functionality: Genetic Alliance BioBank, Sidney Kimmel Cancer Center, UBC-James Hogg iCAPTURE Centre.

Discussion

DAOs seek effective solutions for their registry and biobanking needs. Most would prefer an integrated solution, however <15% vendors surveyed have both registry and biorepository solutions, and not all biorepositories offer comprehensive services. In general, DAOs require systems that begin with baseline information collected, and then customize the registry intake with disease-specific data elements with the capacity to collect longitudinal data. While approximately one half of vendors offered customizable solutions and the collection of longitudinal data, only one third used controlled vocabulary and messaging. It is probable, given the HITECH Act[7] that many will convert to these systems. Of those that used controlled vocabulary, a variety of languages are being used. Standardization and use of controlled language and messaging is essential for data sharing across projects and diseases.

Many DAOs build registries and biorepositories to characterize the disease, determine potential studies, and build cohorts for emerging clinical trials. The ability to link identifiers to data and samples as well as to recontact donors is critical for these activities. Not all vendors provided the necessary software and systems to allow samples and data to continue to be linked to donors. Few provided solutions to make recontact and long-term follow-up a streamlined process. Further, this survey focused on comprehensive biorepository services, as DAO-established biorepositories will likely collect multiple sample types and require a variety of sample processing solutions depending on their collection. Quality control procedures and regulatory compliance are important to any data or sample collection endeavor, and most vendors offered these.

Limitations

This assessment focused on potential solutions for DAOs, and the complete registry/biorepository landscape was not captured. Survey participation was voluntary, participants self-selected and self-reported, and only those who received the survey could respond. Because DAO registry and biorepository needs may differ from that of other stakeholders, this assessment does not address the needs of all customers, and perhaps does not provide the necessary parameters for all DAOs.

Summary

DAOs are important champions for research, and tools, such as criteria for registry and repository vendor selection, must be developed to assist these organizations as they navigate the scientific landscape. Biomedical research and the technologies that support it are accelerating rapidly. Any system that is to provide a robust infrastructure must not only keep pace, but also envision the future beyond its own structure. It is probable that simple solutions that are customizable and cost effective need to be developed so that DAOs can spend more resources on the tasks of recruiting participants, characterizing the condition, and enrolling studies. Further, multiple solutions, with the ability to network them through interoperable platforms may provide better overall tools for advancing genetic disease research.

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