

Day two-breakout session C.1

Chairs: Jim Vaught & Chris Moskaluk

Note takers: Amy Farber & Josh Sommer

C.1 Biospecimens/Biorepositories

Biospecimen resources play a key role in translational research, and are especially critical in rare disease research. The lack of availability of specimens of appropriate quality and with appropriate annotation has been identified as a key obstacle in research.

Furthermore, efforts to create these resources and patient registries are currently scattered and do not have uniform protocols. In order to improve this situation several important steps need to be taken: Best practices including standard operating procedures must be adopted to assure consistent quality of data and specimens; access rules must be developed that allow for efficient and widespread availability of specimens and data; and databases are needed that will clearly identify the location of available specimens and data. These are general guidelines for improving the situation concerning identifying and acquiring specimens of consistent quality. For rare diseases the situation concerning biospecimens is made more difficult since these specimens are even more difficult to acquire in numbers sufficient for many studies. This Biorepositories/Biospecimens session will explore questions and issues related to locating and acquiring specimens and data from rare disease patients.

Some of the issues to be discussed include:

- ❖ Sample procurement and processing: need for standardized protocols
- ❖ Collecting adequate prospective clinical data: Linking samples to registries, use of patient identifiers in rare disease research
- ❖ Biobanks as part of the scientific infrastructure not a project by themselves (include in background)

- Locating biorepository resources for rare diseases: Should there be a specimen locator just for rare diseases?
- Should we include ICD codes when available?
- Should we track investigators using samples from the same specimen or from the same disease and, if so, how?
- There are about 7000 rare diseases; how should they be listed in the specimen locator? Should all be listed? Should we categorize them? What criteria shall we use to categorize them (e.g. anatomical and non-anatomical)?
- Many of the rare diseases are neurological and genetic diseases. The diagnosis of these diseases is based on bodily fluid (e.g. tissue, blood, urine, saliva) or imaging, and not on solid tissues. How can this information be of use to help identify biospecimens and research rare diseases?
- Centralizing biobanks for rare diseases: Issues of ownership, authorship and intellectual property

- **Command and Control:** Who decides what samples get released to whom and when?
- Should we promote TMA production to increase /sharing of valuable and rare specimens?

Additional questions from a participant: where/if to include these questions:

- If a national rare disease biobank and/or registry are created, how will subjects be recruited?
- Would recruitment be federated across disease specific organizations? How will registries and biobanks that DSOs have already created be connected to, or merged with, the national efforts? What role, if any, will DSO's have in stewardship and use of data and specimens in a national biobank? And if DSO's are given a role, which ones will be at the table?
- What are the obstacles for accessing biospecimens needed for clinical studies?
- How can this registry facilitate increased acquisition of biospecimens and improve sharing by different?

Uniting Rare Diseases



Breakout session C.1 Biospecimens/Biorepositories

Overarching Theme: Communication

- inventorying/cataloging existing resources
- vetting/education on biorepository best practices/SOPs
- communication of research results back to subjects
- partnering with pre-existing resources and programs to prevent duplication of effort and to leverage limited funds

Organizing/identifying biorepositories

- Retrospective efforts
 - create a catalog of biorepositories
 - create a general inventory of specimen types
 - identify ability of and extent to which the resource will share
 - formal collaborative basis, MTA, etc.
- Continue ORDR's partnerships with OBBR/NCI/NIDDK to integrate rare disease specimens into the Specimen Resource Locator

Organizing/identifying biorepositories

- Prospective efforts
 - Recruit rare disease biorepositories into a linked virtual repository
 - adherence to identical SOPs
 - adoption of common data standards to allow seamless data sharing
 - adoption of agreements/rules for specimen and data sharing

Organizing/identifying biorepositories

- Prospective efforts (cont.)
 - Where funds are available, ORDR should consider some centralized biobanking resources
 - specimen procurement service – technical staff travel to subject
 - procurement of tissue from operations/organ donation/autopsy
 - organized blood/biofluid collection from familial cohorts
 - Potential to partner with pre-existing resources (NDRI)
 - centralized biobank that operates with evidenced-based best practices and manages some aspects of informed consent

Procedures and protocols

- Recommend that ORDR identify and vet SOPs for specific biospecimen types
 - best not to duplicate efforts
 - partner with OBRR, ISBER and other biorepository resources in creating & identifying evidenced-based protocols

Procedures and protocols

- Biorepository best practices should include:
 - Collection of longitudinal data
 - Consent form that indicates specimens may be shared with outside investigators in widest possible use of the specimen
 - Consent form that allows recontact of subjects for additional data/specimens

Communication of research results

- No consensus was reached over the degree to which subjects should be informed of research results obtained from donated biospecimens.
- Issues include
 - rights of subjects to be informed of research in which they are involved
 - the preliminary nature of the data that is not appropriate to base clinical decision making upon
 - ethics of enticement to join a clinical trial based on personal research results

Communication of research results

- Recommend that ORDR sponsor additional workshops and focus groups to investigate these issues.

Thinking outside the box: renewable biorepository resources

- Create some renewing biospecimen resources in a centralized facility
- EBV immortalized lymphoblastoid cell lines
- creation of cell lines from tumors, non-neoplastic diseased tissue and normal tissues
- propagate xenografts of rare tumors