

Day one –presentation- session II
Jeff Thomas

Abstract for the Meeting “Advancing Rare Disease Research: The Intersection of Patient Registries, Biospecimen Repositories and Clinical Data”

“The Use of Patient Registries to Increase Procurement of Rare Disease Biospecimens”

Jeffrey Thomas

Director of Donor Services, National Disease Research Interchange

It is critically important to introduce patients to the concept of tissue donation, organize donor consent and plan coordination of tissue recovery as far as possible in advance of donation events. Donation events are complex and frequently occur at very short notice and it is essential to have a plan in place, especially to facilitate recoveries from surgical centers and *post mortem*. Donors are an instrumental driving force in ensuring that surgeons and pathologists understand the importance to patients of a successful tissue recovery for research. Trained personnel are required to discuss and document all components of the donation process. Informed consent, donor data and key contacts must be obtained, and a detailed plan developed to ensure the recovery, processing, preservation and shipment of high quality biospecimens.

Given the large number and diversity of rare diseases, and the fact that potential donors are inevitably distributed across the entire USA, it is essential that an established national tissue recovery network, such as NDRI's, is in place to support the uniform recovery of tissues. A productive tissue collection center network includes organ procurement organizations, eye banks, tissue banks, surgical centers, hospitals and contracted professional recovery specialists. Access to such a broad network allows for a wide range of donation opportunities, even at short notice. An important element of an effective donor registry is the ability to recover serial donations from living donors, together with extensive medical history data that reflects disease progression. These donors can also be consented to provide blood and other tissue samples from which DNA and cell lines can be generated and banked. An example of this type of program is a special project with the Von Hippel-Lindau (VHL) Family Alliance to process and store buffy coats, plasma and DNA from VHL patients. Another unique NDRI initiative funded by NHLBI is the Lymphangioliomyomatosis (LAM) Tissue Procurement Program, which exemplifies the value of patient registries in the procurement of biospecimens during transplant and *post mortem*, especially for rare diseases like LAM with only 250,000 patients worldwide. Through this program, 1,510 tissues have been recovered from 47 donors, processed and provided to 27 scientists studying LAM. Although primarily a lung disease, the scope of the program has allowed the recovery of many other relevant tissues, including kidney, blood, bone marrow, bronchus, brain, lymph nodes, and pleural fluid. In collaboration with the Cystic Fibrosis Foundation, NDRI has been responsible for the recovery of over 80 diseased lungs from transplant recipients with cystic fibrosis. Cells derived from explanted lungs have been used to develop two new CF drugs which are currently in clinical trials.

NDRI has created and implemented a National Rare Disease Voluntary Health Organization (VHO) Partnership, which incorporates the building of donor registries to facilitate the donation of rare disease tissues from surgical procedures, transplants and *post mortem*. Twenty-two VHOs have officially joined the NDRI Rare Disease VHO Partnership and NDRI works with 40 to 50 VHOs in an ongoing way. The NDRI rare disease donor registry has grown to include almost 300 individuals representing 47 rare diseases.

Bullets

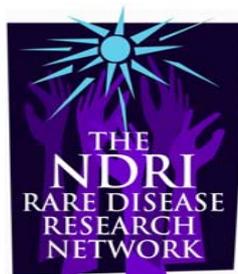
- Donor registries provide the foundation for the procurement of rare disease biospecimens
- Patient advocacy is key to the expansion of successful rare disease tissue recovery programs
- Donor registries are effective when paired with a national tissue recovery network



*Uniting Rare Diseases
Advancing Rare Disease Research:
The Intersection of Patient Registries, Biospecimen Repositories
and Clinical Data
Session II
Biospecimens/Biorepositories*

***The Use of Patient Registries to Increase
Procurement of Rare Disease Biospecimens***

*Jeffrey Thomas
NDRI Director of Donor Services*

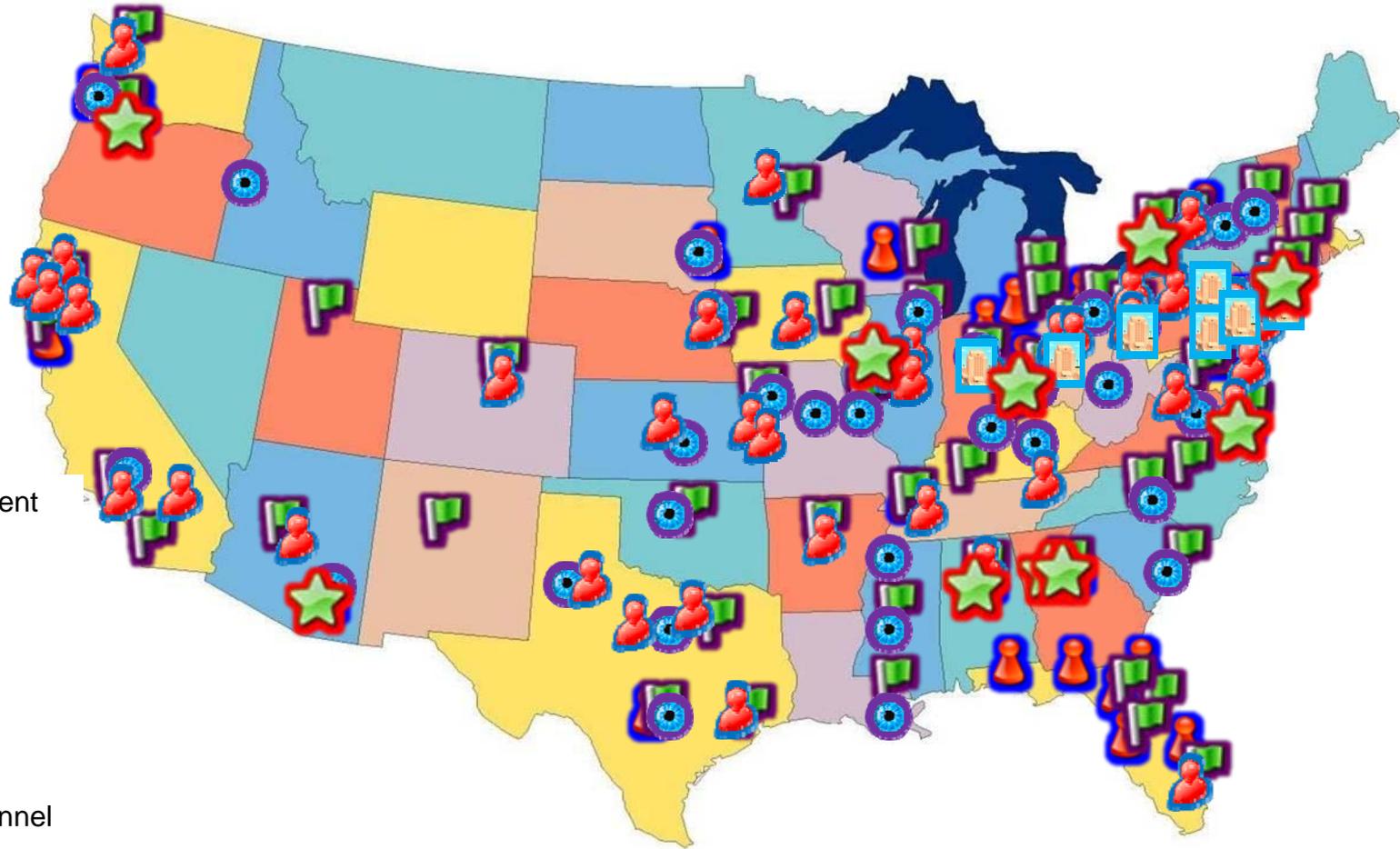


Patient/Donor Registries

Tools to Identify and Support Biospecimen Collection

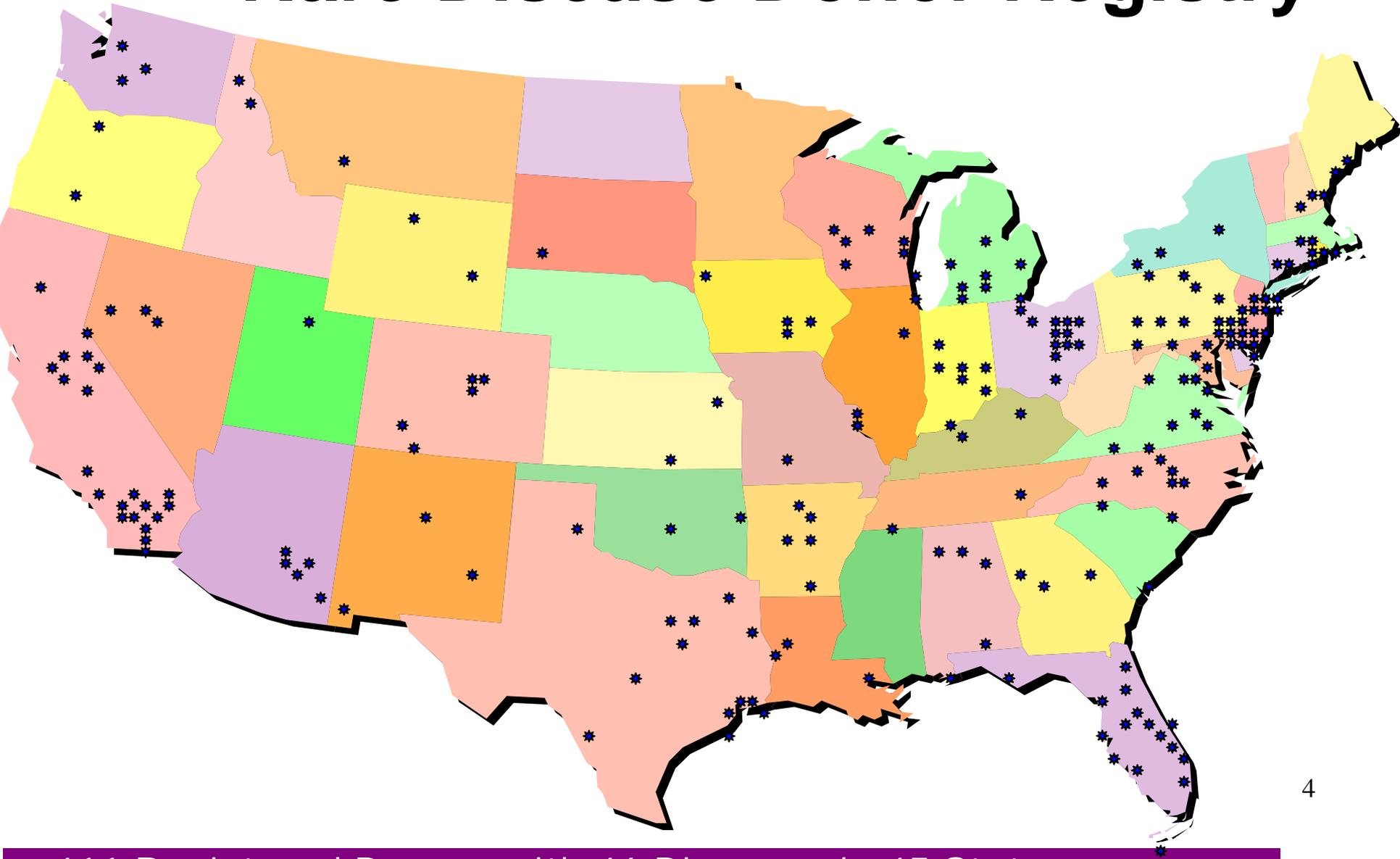
- Essential components of successful tissue procurement programs
- Multiple opportunities for tissue donations (e.g. surgical/transplant, *post mortem*)
- Facilitates multiple recoveries from the same donor
- Comprehensive donor medical history data available
- Paired with national procurement network

National Procurement Network



- 58 Organ Procurement Organizations
- 35 Eye Banks
- 25 Tissue Banks
- 12 Hospital Centers
- 14 Remote Sites
- 41 Recovery Personnel

Rare Disease Donor Registry



- 411 Registered Donors with 46 Diseases in 45 States
- 2,925 Tissues Recovered from 174 Donors with 33 Diseases

Key Elements of Donor Registries

- Timely identification and registration of patients prior to donation
- Validation of informed consent
- Planning with patients & family members
- Organization of procurement team
- Notification strategy for donation event
- Utilization of standardized procurement and processing protocols
- 24/7 availability requirement

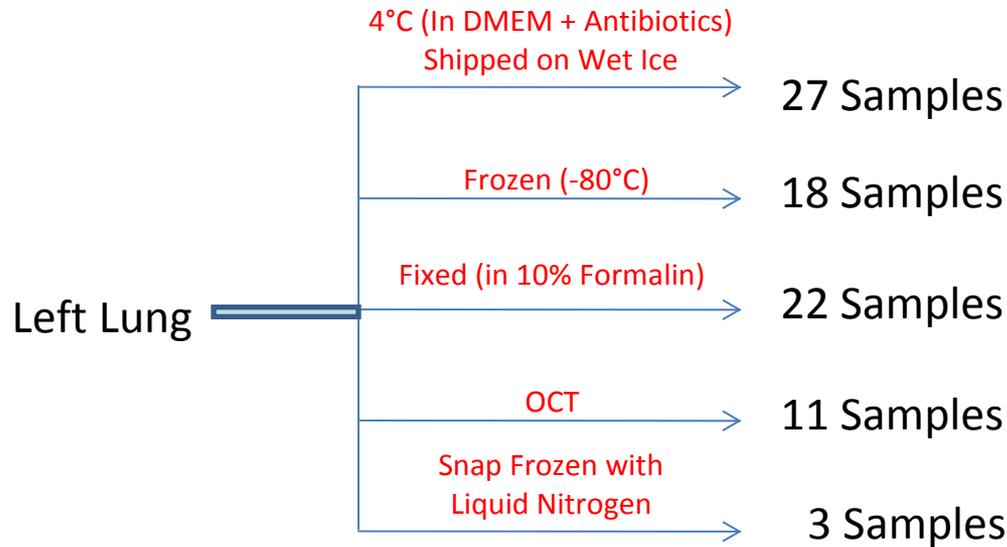
Advantages Resulting from Pre-registration of Donors

- Medical history data and informed consent documented in a secure database
- 24/7 electronic access to key documents by authorized personnel
- Defined and pre-staged tissue procurement logistics
- Control of pre-analytical variables
- Procurements matched with researcher's needs or tissues banked in online inventory

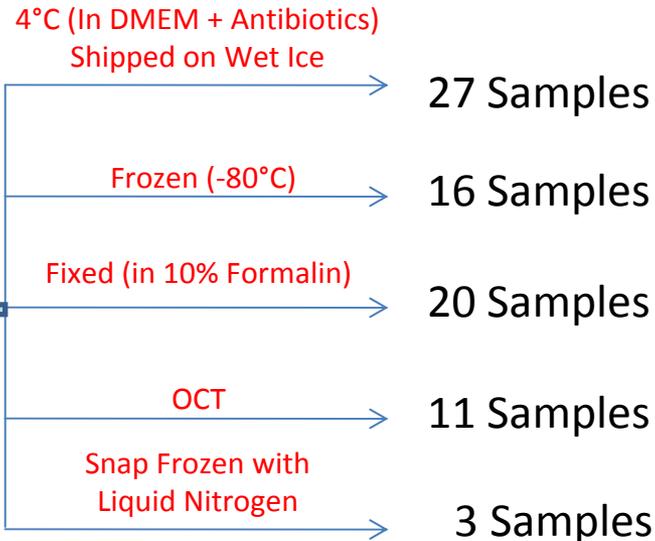
An Example of the LAM Registry Increasing the Recovery of Biospecimens

- LAM Donor Registry
 - 105 Registrants
 - 88 Consented donors
 - 1530 Biospecimens recovered from 47 donors
 - 1513 Biospecimens Placed with Researchers
- LAM Researcher Registry
 - 25 Researchers in 2009, from 6 in 2007
 - Recovery and processing protocols in place

With registered donors, predefined recovery logistics and matched procurement protocols, 2 LAM lungs can serve 25 researchers with 158 samples.



81 Total Samples for Research



77 Total Samples for Research



In the absence of pre-arranged recovery logistics two LAM lungs can serve 9 researchers with 42 samples.

