

Day one-presentation-session III  
Andy Faucett

**Data and test result validation: Reporting research data and clinical test results from the researcher and patient perspective**

**Andy Faucett**

Registries must consider how to collect and insure the accuracy and currency of laboratory test results.

- Current laboratory regulations
- Quality of laboratory testing
- Research and current clinical trials based on genotype
  - Documenting patient populations and clinical endpoints by genotype
  - Pre-research and engaging researchers
  - Clinical trials and requirements
- Capturing accurate test results in a registry
  - Importance of confirmation of diagnosis
  - Complexity of gene and/or disease
  - Impact of technology and knowledge advancements
- Options for a curation plan
  - Now or later – available funds versus benefit
  - Collection of copies of test reports
- Importance of Collaboration
  - CETT Program model



## *Uniting Rare Diseases*

# *Advancing Rare Disease Research: The Intersection of Patient Registries, Biospecimen Repositories and Clinical Data*

## *Session III*

### *Clinical Research, Patient Care and Disease Management*

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*Data and Test Result Validation: Reporting Research Data and Clinical  
Test Results to Patients (Research & Patient Perspective)*



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*Amplified by expertise. Guided by humanity.*

# Accurate Diagnosis is Critical

- ▶ Power of Registry will depend on reliability of information
  - Clinical Diagnosis
  - Test Result Diagnosis

# Information Accuracy

- ▶ Important for Registry community to understand information may need to be confirmed
- ▶ Clinical based diagnosis
  - Consensus among clinicians
  - Collection of clinical records to support
- ▶ Test Result based diagnosis
  - Validation of results
  - Standardization (current nomenclature)

# Sharing Test Results

- ▶ Clinical results versus Research results
- ▶ Most groups start with Research results, need to move to clinical lab results
- ▶ The important Rules
  - CLIA regulation
    - Most research labs not CLIA
  - Common misconceptions
    - \$ exception
    - Research exception
    - IRB exception

# Using Research Results or Clinical

- ▶ Lab error rate higher in research
- ▶ Timeline in research versus clinical
  - Initial research testing
  - Move to clinical lab
    - Collaboration, Education & Test Translation Program for Rare Genetic Diseases (CETT)
- ▶ Information on Report
  - When to retest “negatives”

# Collection of Results in Registry

- ▶ Clinician
  - Confirm nomenclature
- ▶ Patient
  - Right to copy of report
    - Information about how

# Cost / Benefit – Now or Later

- ▶ Insure accuracy of information at capture
  - Clinician entry
  - Curation by expert
- ▶ Collect information / Curate later
  - Decide what to collect
  - Teach how to get report copies

# Additional Reasons

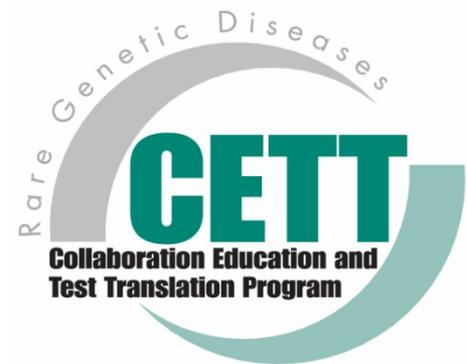
- ▶ Learn unexpected Genotype / Phenotype associations
- ▶ Develop clinical endpoints for clinical trials
- ▶ New treatments based on genotype
  - Type of mutation
    - May make your condition “attractive”
- ▶ FDA
  - May be required for clinical trials

# Curation in Action

- ▶ DuchenneConnect
  - General congruence of information – “gestalt”
  - Genotype
  - Cardiac reports
  - Lung function reports

# Benefits of Collaboration

- ▶ CETT – Collaboration, Education & Test Translation for Rare Genetic Diseases
- ▶ Create win/win
  - Research funds used for research
  - Researcher aware and involved with new findings
  - Capture genotype and phenotype data
  - Identify research candidates



# Summary

- ▶ Validation of Information
    - Understand importance
  - ▶ Questions to Ask
    - What
    - How
    - When
  - ▶ Develop plan
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