Lisa Heral

Fibrous Dysplasia Foundation
The NIHR rare disease *musculoskeletal* translational research collaboration (NIHR RD TRC)

MK Javaid, R Luqmani, P Wordsworth, H Teare, K Melham J Kaye
National Collaboration…
NIH – Global Program

Goals for Current Phase of the PROGRAM, June 2014 – May 2015

1. Aggregate data from multiple disease groups into single *interoperable* database.

2. Establish a program and data governance model.

3. Create a secure query tool.

4. Determine resources needed to scale program to additional rare diseases.
NIH/NCATS GRDR™ Program

Global Rare Diseases Patient Registry Data Repository

- Registry owners notify identified participants and directed to study PI
- Patients join a registry and provide health information
- Registry managers de-identify collected patient data and biospecimens, and assign Global Unique Identifier (GUID)
- De-identified patient data is shared with GRDR™ program staff
- Patient data linked to biospecimens via the GUID Interfacing with Rare Diseases Human Biospecimens/ Biorepositories (RD-HUB)

Researchers conduct various biomedical studies within & across diseases

Researchers, Clinicians, Industry Pharma

- Patients
- Patient Registries
- GRDR™ Database
- Other RD Databases
- RD-HUB

GRDR aggregates, maps data to CDEs & national standards, integrates patient clinical information and provides access to approved researchers

NIH National Center for Advancing Translational Sciences
Mission Statement

“The Fibrous Dysplasia Foundation provides information, advocacy and support for patients, medical professionals and the general public and promotes research for diagnosis, treatment and a cure for Fibrous Dysplasia (FD) and McCune-Albright's Syndrome (MAS), Cherubism and related diseases. Our goal is to improve the quality of life for affected individuals and their families.”
The NORD Natural History Platform

October 25, 2014
NORD History
Registries and rare disease

• What’s a “registry”?  

• What’s a “natural history study”?  

• Why are registries so important for rare diseases?  
  – Improve care  
  – Increase efficiency in research efforts  
  – Support development of treatments
NORD’s role evolves

Thought leadership and advocacy

Direct support to rare disease patient organizations
What we believe

• Empower the community
• Empower the community
• Empower the community
• Empower the community
Overview

• Collaboration with FDA and NIH NCATS
  – Subject matter experts
  – Technologists
  – Epidemiologists

• Develop a safe, centralized, and reusable platform for constituents and stakeholders.

• Establish guidelines and best practices for natural history study design and execution
VHL Databank Pilot

- Secure
- NHS
  - Medical history
  - Patient Reported
- Project is controlled by VHLA
Registration

- Simple, straightforward design
- Respondents
- Participants
- Contact preferences
Account Management

- Multiple participant access
- Two-click consent revocation process
Survey Sample

- Branching logic to “Show/hide”. Only answer see questions that are pertinent to you!
- Real time data validation
- File uploads
- Draft/Final Submission

Property of NORD, Inc.
Mobile and Tablet Access

- Responsive design automatically adjusts to screen size
Instant feedback

- See how you match up with charts and scores
- Return to your completed surveys anytime
Plans for the future

- More clinical data, with less data entry
- Multiple language support
- Other features based on feedback we hear from you and the FDF board.
Why we’re working with FDF
Questions?
Questions?

Thank you!

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Dr. Amanda Konradi

Fibrous Dysplasia Foundation
QUESTIONS

• WE ARE AVAILABLE DURING LUNCH

• LISA.HERAL@YAHOO.COM