OFFICE OF RARE DISEASES UPDATE

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> 7,000 rare diseases

25 M affected in US

Many undiagnosed

< 500 have any treatment
How can we all make a difference for rare diseases?

- Leveraging technology
- Creating a research continuum
- Partnering for success
- Engaging the next generation of researchers
How can we accelerate the path from discovery to health benefit?

Integrating care and research

Integrating data from multiple sources

Clinic

Research

Patient-reported outcomes

Harmonization

Collaboration
How can we make sure our data count?

For rare diseases:

- Continuity of data
- Data standards
- Keep “end-user” in mind

Registry

Natural History Study

Biomarkers

Trials

Post-approval

Photo credit: Michael and Rachel Harris
Partnering for Success

Patient Groups

Academics

Industry

Government
(in US: NIH, FDA)
Trans-NIH Rare Diseases Working Group

- Rare Diseases affect many organs, and are often multi-system diseases.
- Therefore, strong partnerships with the categorical NIH ICs are important.
- Coordination and cooperation stipulated in Rare Diseases Act of 2002.
- One way by which the ORDR facilitates coordination and cooperation is through the Trans-NIH Rare Diseases Working Group.
- The re-constituted working group met on December 12, 2016 and will meet quarterly with rotating meeting venue.
CURRENT ORDR PROGRAMS

NCATS

National Center for Advancing Translational Sciences
https://rarediseases.info.nih.gov

Online resource with:

- **Up-to-date, reliable and easy-to-understand information** on rare or genetic diseases
- **In English or Spanish**
- **For people with rare or genetic diseases**, their families, friends, care providers and wider communities
- **Contact information** for telephone and email queries
NIH/NCATS Rare Diseases Registry (GRDR) Program

- Common Data Elements (CDEs) for collecting data
- Informed consent templates
- Access to GRDR Global Unique Identifier (GUID)
- Map patient data to GRDR CDEs & national standards
- Information and tools
RARE CLINICAL DISEASES RESEARCH NETWORK

Initiative of the National Center for Advancing Translational Sciences (NCATS)
About the RDCRN Program

- Collectively, the RDCRN is studying 200 rare diseases in natural history and clinical trials at 418 active clinical sites located in the US and in 24 countries.
- There are more than 90 active protocols.
- 41,519 patients have enrolled in clinical studies.
- There have been 265 trainees.
- There are 3,545 collaborative consortium members.
- There are 144 PAGs as research partners, collectively formed a Coalition (RDCRN-CPAG).

http://rarediseasesnetwork.epi.usf.edu/
International Coordination

• Given the “rarity” of patients and investigators, the ORDR is engaged in coordination with international partners, including:
  ➢ IRDiRC
  ➢ GA4GH
  ➢ ICORD

• ORDR participated in a collaborative project with the NCATS New Therapeutic Uses (NTU) program and the European E-Rare initiative:
  ➢ RDCRN investigators were invited to seek support for participation in international clinical trials to repurpose drugs in collaboration with E-Rare (European investigators) [http://grants.nih.gov/grants/guide/pa-files/PA-16-183.html](http://grants.nih.gov/grants/guide/pa-files/PA-16-183.html)
  ➢ Applications were received in July, 2016 (including two RDCRN applications).
NCATS TOOLKIT PROJECT
Rare Diseases Toolkit

Why?

• Patient involvement and community engagement are vital throughout the translational research process

• A wealth of educational and informational tools have already been developed by and for the rare disease community

  ➢ Academia
  ➢ Disease foundations
  ➢ Government agencies
  ➢ Industry

• **But** existing resources are dispersed and difficult to discover, especially for newcomers
What will the Toolkit project do for the rare diseases community?

- Collaboratively create a **well-designed source for online educational and informational research resources and tools**.
- Provide a **single online portal with resources** that patient groups can readily access along with context.
- **Improve coordination** rather than re-create existing resources.
- Facilitate opportunities to **bring groups together, identify gaps** in online resources, and **disseminate information** to patient groups.
- Promote **continuity across the lifecycle of the drug development process**.
How we plan to develop the Toolkit

• Planning group driven by patient group representatives
• Inclusive*, transparent, collaborative
• Focus on tools that are useful for research, easily accessible and practical

Ascertaining needs of patient groups
Survey landscape of available tools
Develop & Demonstrate Sept. 20, 2016 workshop
Disseminate starting spring 2017, via larger meeting(s) & webinars

• Identify gaps & opportunities
• Organize tools based on exemplary use cases
• Educate & inform rare disease community
• Develop programs that assist with use-case based strategies for patient groups at different stages

* Tools are suggested for inclusion by the patient community. While we cannot capture everything, the initiative will be evolving and we invite comment and feedback.
Potential Future Directions

- Increasing collaborations between the ORDR and NCATS DPI programs such as TRND
- Shifting from “one-disease-at-a-time” to “rare disease-ome” approach
- Stimulating partnerships for rare diseases
- Registries and natural history studies that are trial and regulatory-ready
- Promoting new therapeutic modalities such as gene therapy and editing for rare diseases
- Harmonizing and internationalizing rare diseases clinical research networks
Take-home Messages

- Leveraging technology
- Creating a research continuum
- Partnering for success
- Engaging the next generation of researchers

IF YOU WANT TO GO FAR, GO TOGETHER.

- African Proverb
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