2017 Scientific Workshop #9

Emerging Technologies for Rare Diseases: Clinical and Regulatory Case Studies and Approval Pathways

No Disease Is Too Rare to Deserve Treatment
Disclosures

- Founder, executive employee and stock owner, Ultragenyx
- Prior employee and stockowner, BioMarin
- President, EveryLife Foundation for Rare Diseases
Thank you to today’s sponsors!
Mission and Core Principles

Accelerating biotech innovation through science-driven public policy

What We Believe:

- No disease is too rare to deserve treatment
- Rare disease therapies should be safe and effective
- We could do more with the science we already have

What We Do:

- Advocate for evidence-based changes in public policy, development strategies & regulatory review

How We Get it Done:

- Grassroots action
- Scientific and policy expertise
Public Policy Objectives

The Foundation seeks practical policy solutions:

• Close the innovation gap for the 95% of rare diseases that have no FDA-approved treatment
  • Support initiatives and new technologies that foster novel and innovative treatment

• Ensure patients receive earliest access to diagnostic and treatment opportunities

• Improve the development/regulatory process and advance regulatory science for rare disease therapies

• Enhance the patient voice in policymaking, drug development and regulatory decision-making
Policy and Advocacy Initiatives

**RARE DISEASE LEGISLATIVE ADVOCATES** clearinghouse to train patients and parents on how to be effective in changing policy.

**EXPANDING NEWBORN SCREENING** state legislation to require a state to screen for a disease once it’s on the federal RUSP.

**COMMUNITY CONGRESS** Collaboration between patient organizations and industry representatives to seek policy solutions.

**INCENTIVIZING RARE REPURPOSING** federal legislation to double the number of rare disease therapies approved by FDA.
Rare Disease Congressional Caucus Briefing Tomorrow

Curing Rare Disease: Policy and Regulation Needed for Emerging Technology

Wednesday, September 13th, 2017
12:00 – 1:00pm
Senate Visitors Center, Room 203-02

Lunch provided
Co-Chairs Lance and Klobuchar to speak
# Workshop Series Topics

*Designed to bring policy leaders together with experts in the field to build the scientific capacity needed to create science-driven policy solutions*

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*Slides from prior workshops are posted at [www.everylifefoundation.org](http://www.everylifefoundation.org).*
Golden Age of Rare Disease Treatments

- Nearly 600 rare disease therapies in development
- Cures are now possible with emerging technologies
- Congress passed 21st Century Cures Act with broad bipartisan support in 2016

Orphan Designations

Orphan Approvals
New Treatment Modalities

- Gene Therapy
- Protein Replacement Therapy
- Transcription Induction
- Splicing Modification (exon skipping)
- Nonsense Suppression
- Chaperones Folding
- Gene Editing
- Antisense or siRNA
- Substrate Inhibition
- Protein Replacement Therapy
New Development and Regulatory Strategies

- New sources of evidence being considered
  - Patient-reported, patient-generated data, & real-world evidence (RWE)

- New trial designs and endpoints
  - Increased understanding of underlying Nat. Hx of disease
  - Alternative control groups, novel single arm designs
  - Novel endpoints being considered
  - Use of biomarkers as endpoints for dosing and efficacy

- New approaches to nonclinical evaluations
  - Guidance simplifying testing for enzyme replacement
  - Consideration for platform versus individual drugs
Goals for Today

The Foundation seeks small, scientifically sound policy changes that have a large impact and translate to investment and innovation for rare diseases treatments.

What areas can we identify today that need policy or strategy improvements?

- Development strategies
- Regulatory evaluations
- Commercialization

What are the 2 or 3 most important tractable and practical issues that are the most critical that we should seek to change in drug policy or development?