The Role of Patients in Developing Treatments

Brad Williams
Jain Foundation
2B Empowered Conference
May 2015
You are important!!!

• Clinical research is how treatments are developed
• It can’t be done without patients!
• Some clinical trials will have very specific requirements for enrollment
  • You may benefit from the treatment after it’s approved even if you don’t qualify for the clinical trial
• Many of us will participate in more than one study, data may be collected multiple times

Thank you in advance for participating!
Drug Development Pathway

Key questions for approval:
Is it safe?
Does it work?

Candidate Drug → Preclinical studies
- Studies of effectiveness and toxicity in animals

Recombinant DNA Advisory Committee
- Applies only to gene therapies

Investigational New Drug Application (FDA)
- FDA reviews preclinical data, decides whether to approve clinical trials

Clinical Trials
- Phase 1: Small number of patients, looks at safety only
- Phase 2: Look at safety and initial indication of effectiveness
- Phase 3: More patients, need to demonstrate statistically significant benefit and acceptable risk

Approved Drug
What do we need to go through the pathway?

• Candidate drugs: role of research is to develop them
• Clinical trials need
  • Patients to participate in them
  • Clinicians to conduct them
  • Outcome measures to interpret them
• Interest of drug developers (pharma companies)
Categorization of treatments: applicability

- For muscle diseases in general (muscle growth promoters)
- For various muscular dystrophies (steroids)
- For dysferlinopathy specifically (DYSF gene therapy)
- For specific mutations
  - In DYSF gene only (exon skipping)
  - In DYSF or other genes (Ataluren)
Categorization of treatments: types

• Repurposed drugs
  • Already FDA approved for some other indication
  • Safety data already known, but efficacy for dysferlinopathy not established
  • May need to conduct clinical trial
• New drugs
  • “Small molecules”
  • Biologics
  • Genetic therapies
To be ready for clinical trials, we need to...

• Know Cause of Disease
• Understand Mechanism of Pathology
• Build Patient Registry
• Characterize Natural History of Disease
• Develop Readouts for Clinical Trials
• Identify Biomarkers to Monitor Progression
To be ready for clinical trials, we need to...

• Know Cause of Disease ✔ Dysferlin gene discovered in 1998
• Understand Mechanism of Pathology
• Build Patient Registry
• Characterize Natural History of Disease
• Develop Readouts for Clinical Trials
• Identify Biomarkers to Monitor Progression
To be ready for clinical trials, we need to...

- Know Cause of Disease
- **Understand Mechanism of Pathology**
- Build Patient Registry
- Characterize Natural History of Disease
- Develop Readouts for Clinical Trials
- Identify Biomarkers to Monitor Progression
To be ready for clinical trials, we need to...

• Know Cause of Disease
• Understand Mechanism of Pathology
• **Build Patient Registry** ✔ Jain Foundation Registry and IDR
• Characterize Natural History of Disease
• Develop Readouts for Clinical Trials
• Identify Biomarkers to Monitor Progression
To be ready for clinical trials, we need to...

- Know Cause of Disease
- Understand Mechanism of Pathology
- Build Patient Registry
- Characterize Natural History of Disease
- Develop Readouts for Clinical Trials
- Identify Biomarkers to Monitor Progression

Dysferlin Clinical Outcome Study
To be ready for clinical trials, we need to...

• Know Cause of Disease
• Understand Mechanism of Pathology
• Build Patient Registry
• Characterize Natural History of Disease
• Develop Readouts for Clinical Trials  Dysferlin Clinical Outcome Study
• Identify Biomarkers to Monitor Progression
To be ready for clinical trials, we need to...

- Know Cause of Disease
- Understand Mechanism of Pathology
- Build Patient Registry
- Characterize Natural History of Disease
- Develop Readouts for Clinical Trials
- Identify Biomarkers to Monitor Progression

Analysis of Patient Blood Samples
How to Interest a Drug Company in a Rare Disease

• Characteristics of Disease: (things we should know)
  Prevalence? Age of onset? Progression?
How to Interest a Drug Company in a Rare Disease

• Characteristics of Disease: (things we should know)
  Prevalence? Age of onset? Progression?

• How well understood is disease: (role of research)
  Cause? Source of pathology? Natural history study?
How to Interest a Drug Company in a Rare Disease

• Characteristics of Disease: (things we should know)
  Prevalence? Age of onset? Progression?

• How well understood is disease: (role of research)
  Cause? Source of pathology? Natural history study?

• Engagement of patients and advocates: (your role)
  • Visibility and awareness of disease
  • Advocacy organization and patient community
  • Patient Registry to help recruitment for trials
  • Willingness of patients to participate in research
The Current Situation

• COS is providing data on symptoms, progression, outcome measures, and familiarizing clinicians with dysferlinopathy

• Several prospective treatments are undergoing pre-clinical studies

• We don’t know which treatments will work best
  Investigate all of them!!!

• Future: multiple clinical trials for different treatments
What you can do TODAY!

• Spread information about the disease and research.
• Participate in patient groups (online or in person).
• Encourage patients to get a genetic diagnosis.
• Educate clinicians who are unfamiliar with LGMD.
• Document your experiences as a patient.