The Role of Patients in Developing Treatments

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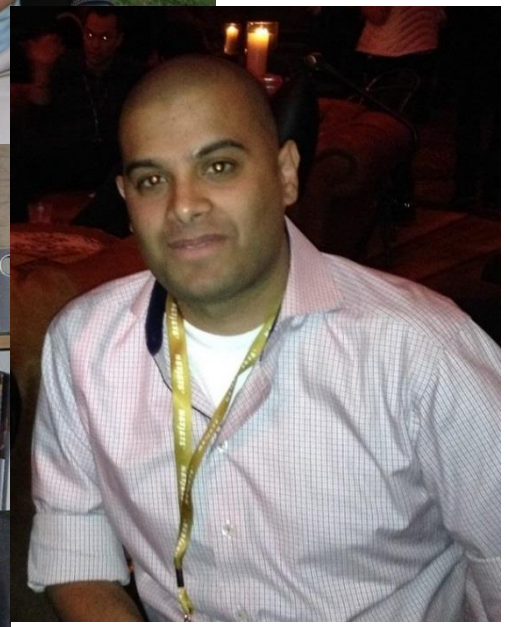
You are important!!!

- 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 it 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

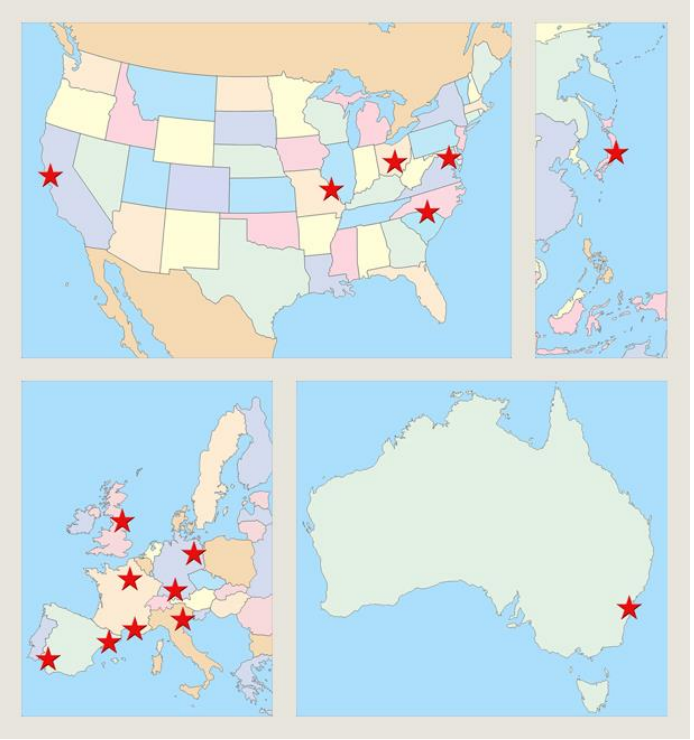
Phase 3

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)

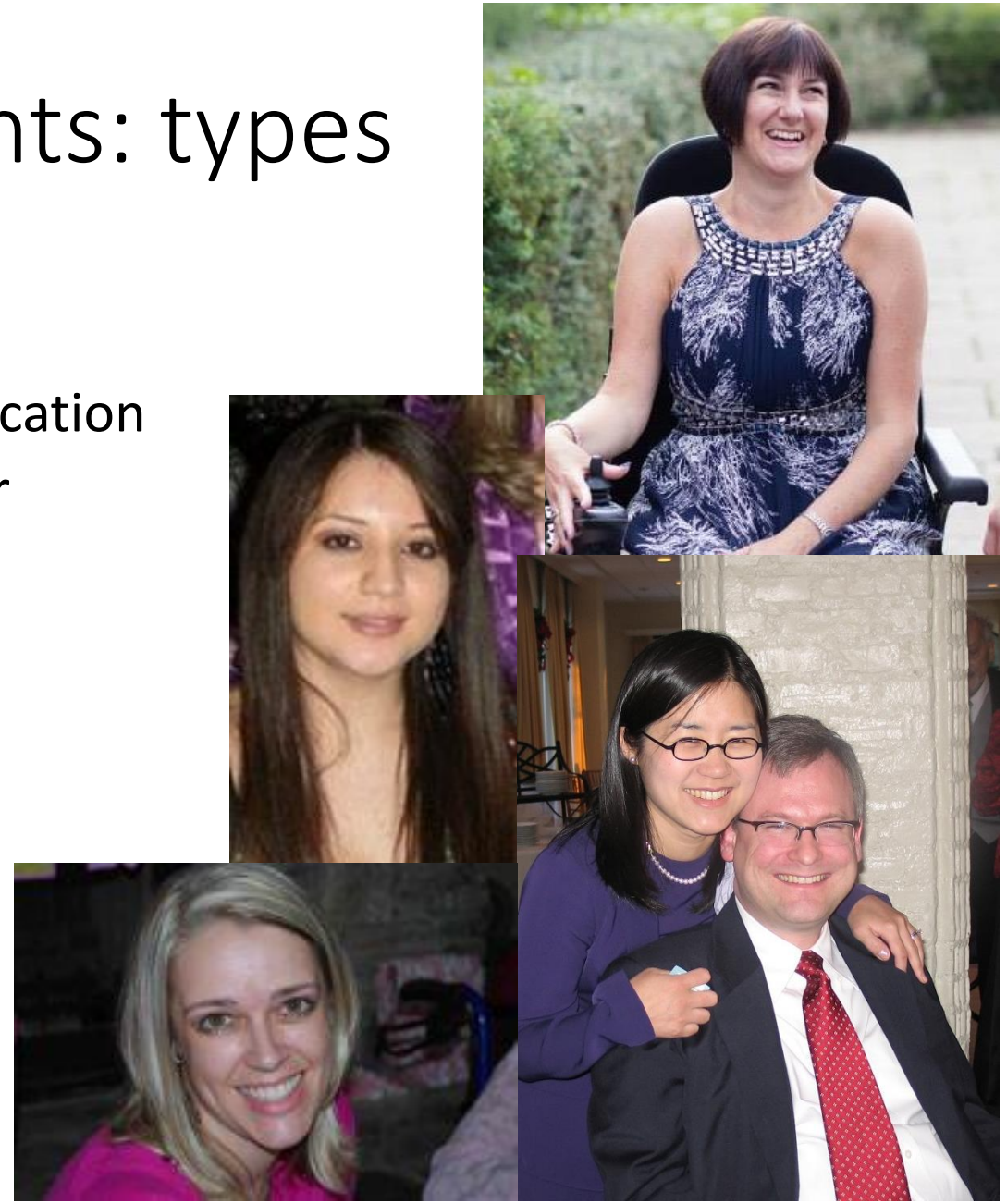


Categorization of treatments: applicability

- 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

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Analysis of Patient Blood Samples

How to Interest a Drug Company in a Rare Disease

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  - Prevalence? Age of onset? Progression?

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How to Interest a Drug Company in a Rare Disease

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- 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!

- **S**pread information about the disease and research.
- **P**articipate in patient groups (online or in person).
- **E**ncourage patients to get a genetic diagnosis.
- **E**ducate clinicians who are unfamiliar with LGMD.
- **D**ocument your experiences as a patient.