



RARE PATIENT ADVOCACY SYMPOSIUM

A partnership of
Penn Medicine Orphan Disease Center
and Global Genes



Sheraton University
City Hotel

Saturday, May 19, 2018

7:00 am - 4:30 pm

Why and How Should Our Community Start A Patient Registry



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#PennMedMDBR2018

#GGPennRareSymposium



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Rare Disease Patient Registries: *Roles in Supporting Rare Disease Clinical Development Programs*



the
orphan
disease center
at the University of Pennsylvania

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Why are patient registries important?

Potential goals:

- A mechanism for elevating the patient voice
- Expedite trial launch: recruitment & site identification
- Data collection & sharing (better disease understanding)
- Enable better clinical study design
- Safety monitoring



Common pitfalls

- Sustainability
- Transparency
- Resource for the entire community
- Data sharing
- The shifting privacy landscape
- Timing



Registry Types

Patient Portals (to collect PROs)

- Contact registry
- Patient or caregiver reported outcomes (PROs): important for understanding clinical needs and determining whether therapeutics are ‘clinically meaningful.’

The FDA calls for the integration of the patient voice throughout the life cycle of investigative products to better inform outcome measures.

Research Registries (integrating PROs and clinical data)

Clinical data is important for understanding the standard-of-care and to better characterize each disease. These data are collected on the same patients that have PRO data, making for a more holistic view of the disease and the patient healthcare experience.

Natural History Studies

Goals: 1) Characterize the disease with respect to diverse symptoms, heterogeneity in presentation, and progression. 2) Development of a comparator arm dataset where placebo controls are not advised

NHS collect clinical data under formal clinical protocols during a special research visit

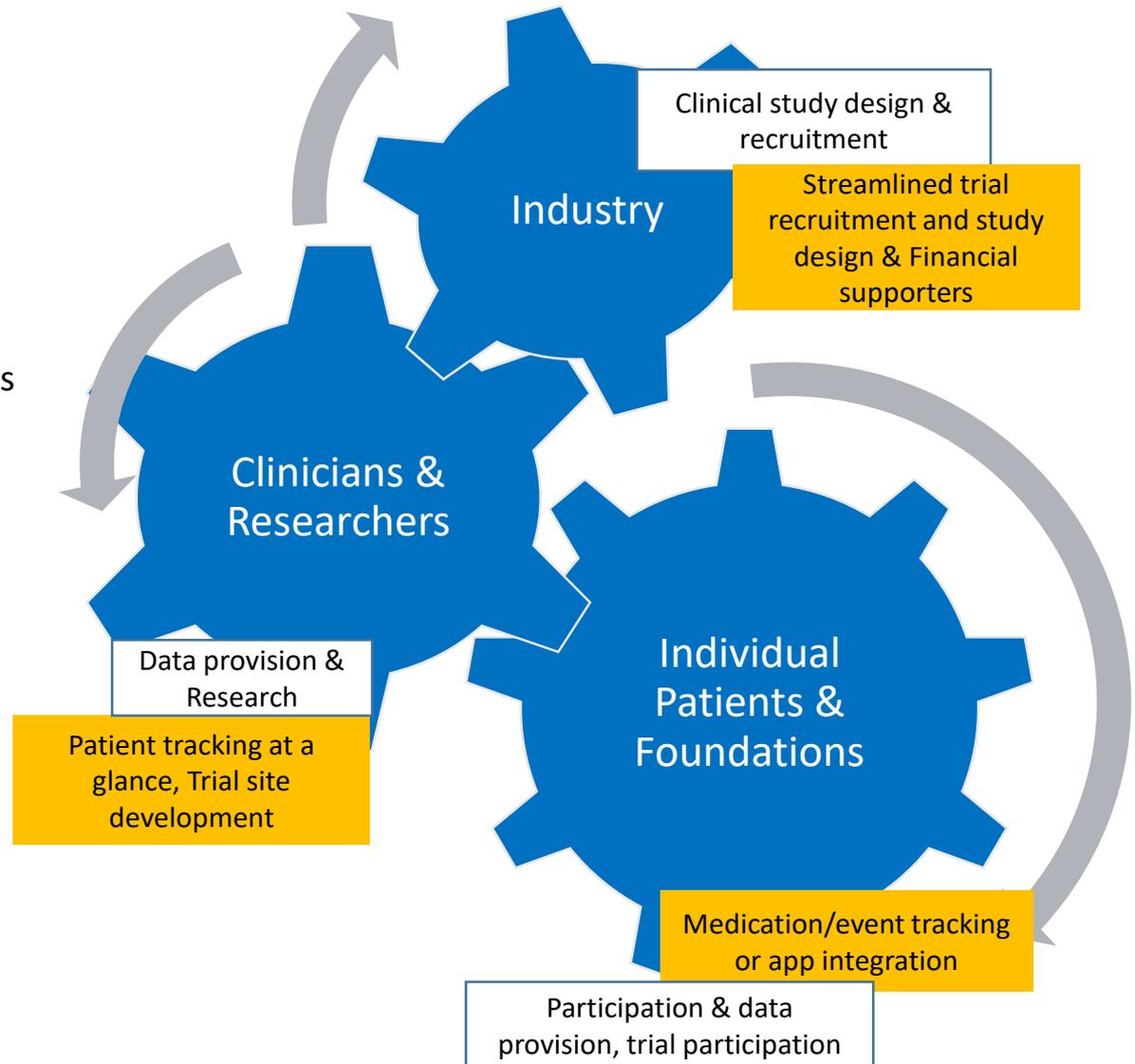
Roles, Responsibilities, (and Incentives...)

Who are the primary drivers in the community?

What are their expectations?

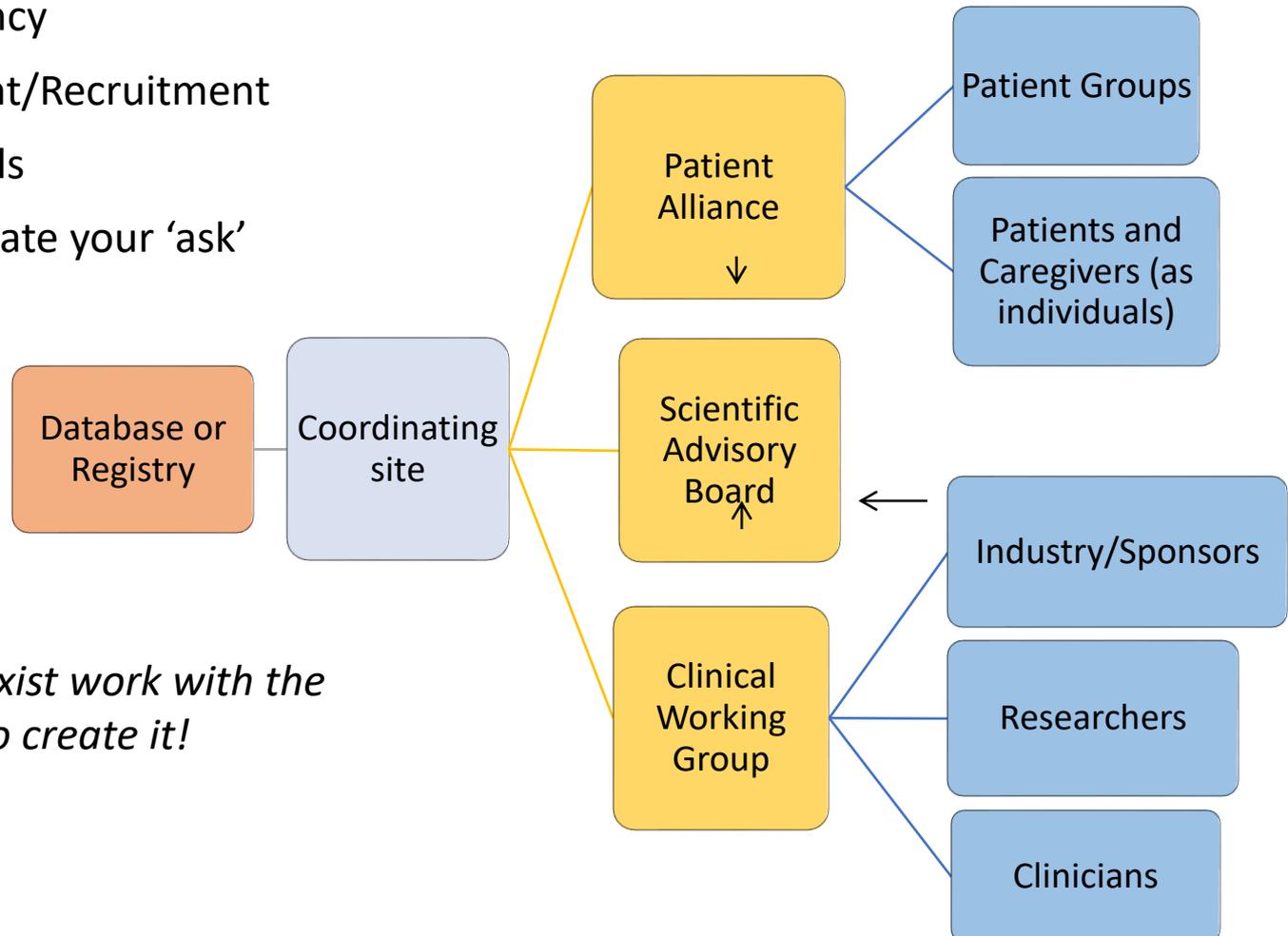
How can you engage these individuals or groups effectively?

What are the primary motivators?



Coordination and communication

- Transparency
- Advisement/Recruitment
- Clarify goals
- Communicate your 'ask'



If it doesn't exist work with the community to create it!



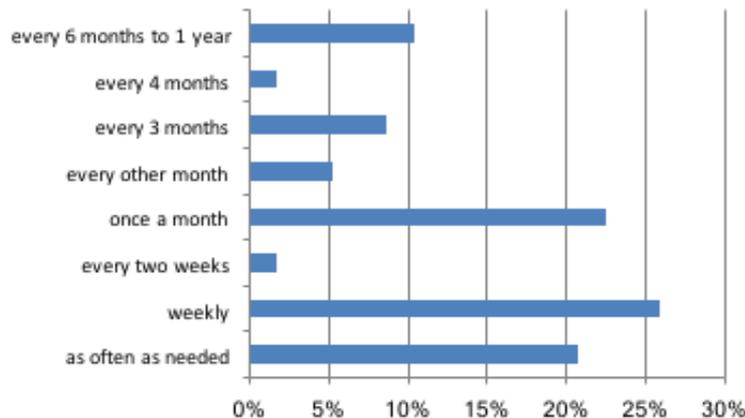
Early feasibility testing

Do you currently use a seizure log to track your child's seizures?

No- 47.3%

Yes- 52.6%

How often would you be willing to go to the Patient Portal to enter data?



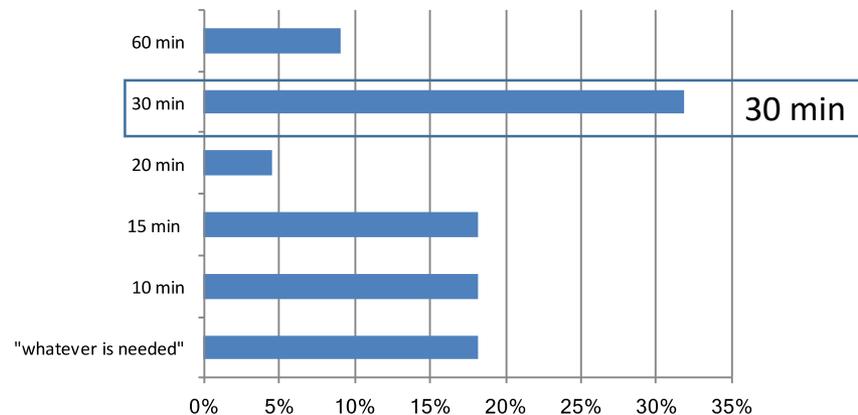
Are you interested in being notified about new Clinical Trials or Research Studies that your child may be eligible for?

Yes- 96.49%

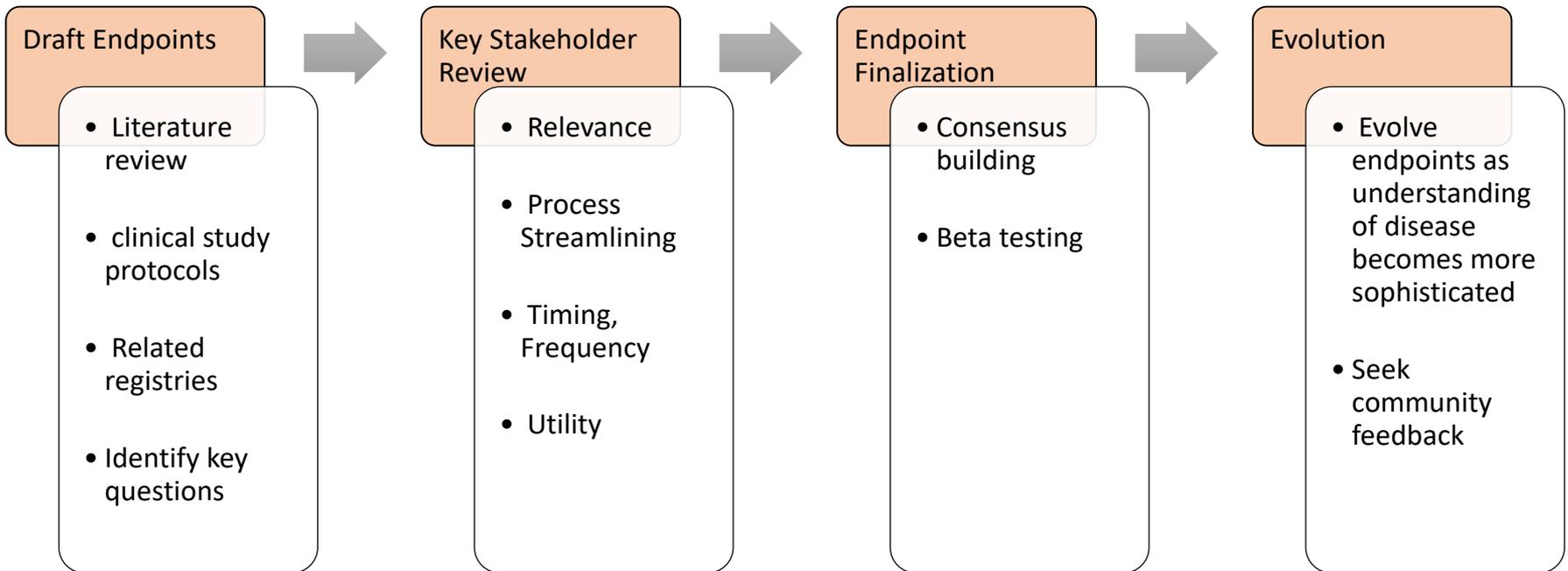
No- 0%

Maybe- 3.51%

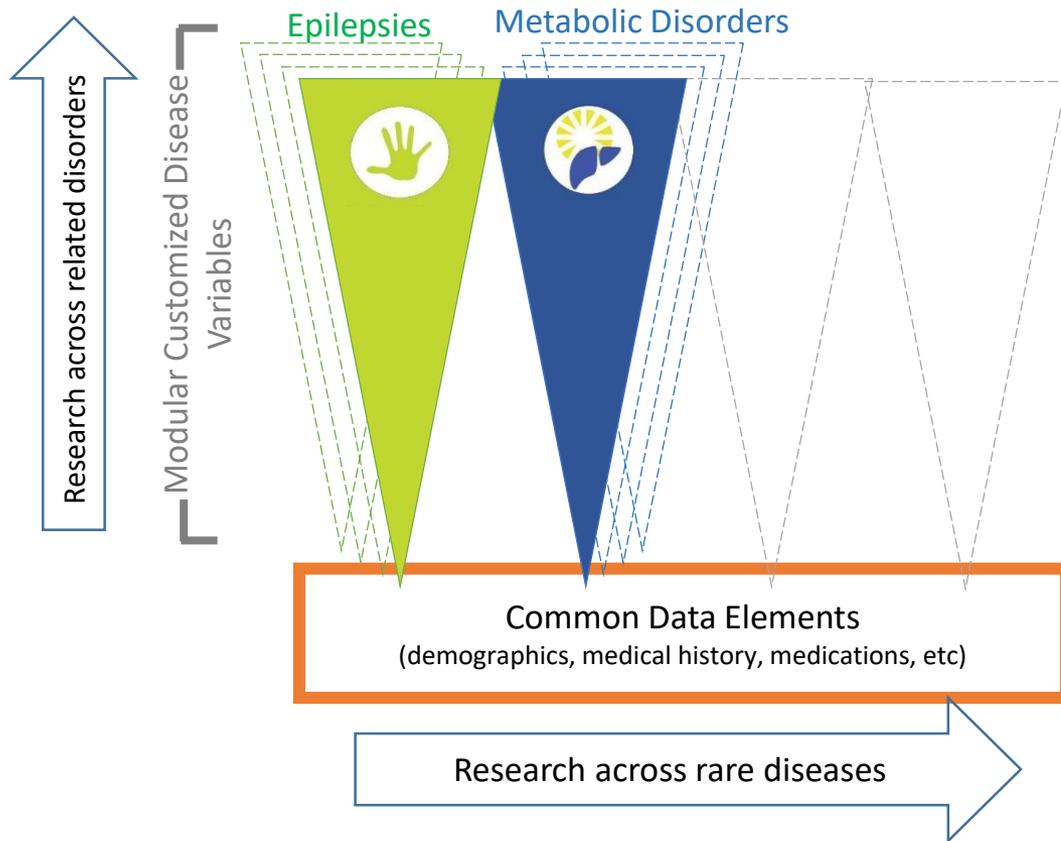
What is the most amount of time you would be willing to spend during one sitting to enter data?



Community Feedback: Building Data Dictionaries



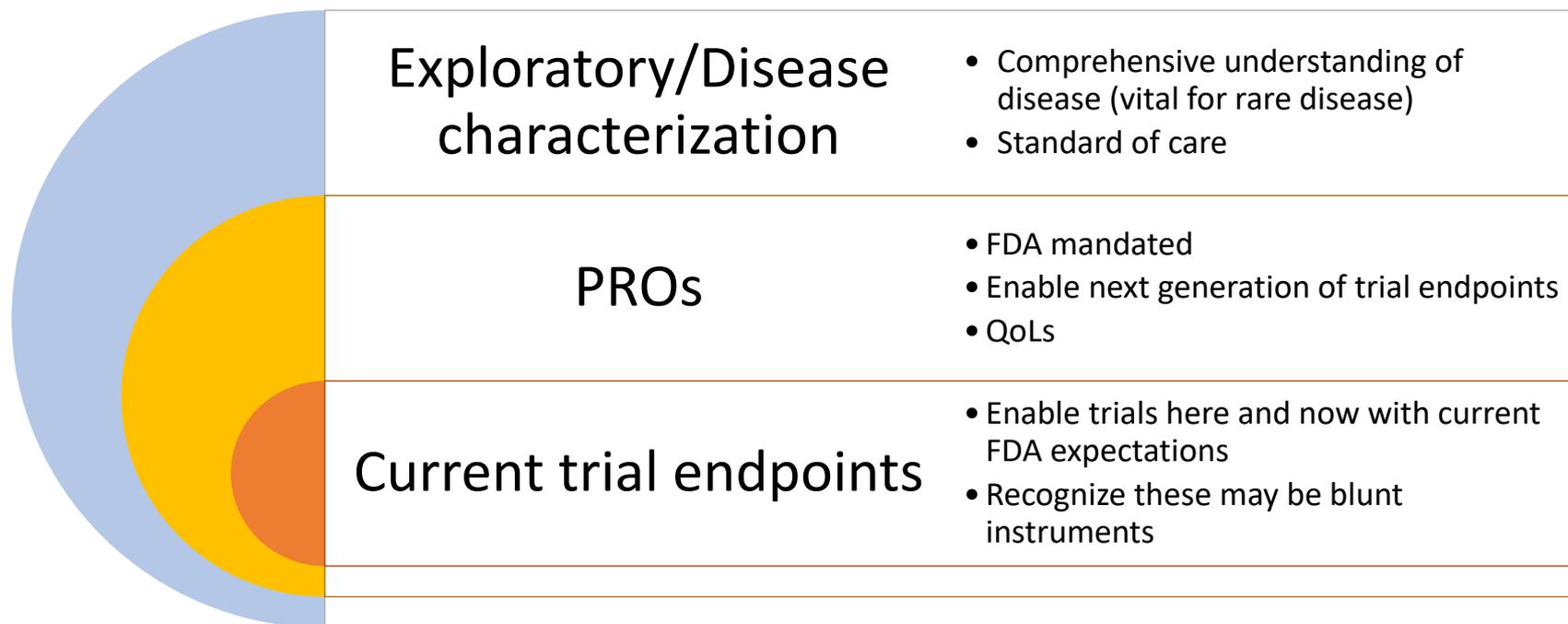
Interoperability & Research



- Use GUIDs (Global Unique Identifiers) to enable data integration
- Utilize common data elements based on established ontologies
- Involve patients and caregivers- understand their motivations, expectations, and what matters



Nesting-Doll Model



1. Prioritize (i.e., by importance or immediate needs)

2. Where possible, enable exploratory areas

3. Consider data collection and analysis as well as feasibility throughout the process



Avoid obstacles

- Community registries should support **multiple** investigational interventions
- Collect PROs & clinician-reported outcomes, when possible
- Find a platform that can **evolve** with community needs
- Be transparent about funding, intentions, and data use

Sustainability

- Consider funding sources at the start: Government, Industry, Foundations/Patient Groups
- **Consortia Model**: Pool resources, community driven effort that is open & inclusive
- What are the primary goals and features? Scale appropriately but **enable future growth**



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To learn more visit us at

<http://orphandiseasecenter.med.upenn.edu/>

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The material presented in this presentation is for informational purposes only. The views expressed in presentations are those of the speaker(s) and not, necessarily, of Ovid Therapeutics.

Functional Registry

- 1. Find an educated team, not a stock platform**

Consistent and reliable people collecting disease-specific and relevant information.
- 2. Seek game-changing phenotypes**

Breakthroughs happen by connecting novel characteristics of a complex community.
- 3. Freely accessible to the research community, including pharma/biotech**

Scientists and clinicians must have the ability to re-contact each family.
- 4. Make it clinical**

Know the primary purpose of your registry and make it cross-functional.
- 5. Efficient Data Collection**

Merge Patient Registry and Natural History.
Activates community by giving each family a vital role in research.

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A Registry Built For Discovery

First 15



Mission: 100 / WES



Family Meeting



Celebrate



Study



Organize



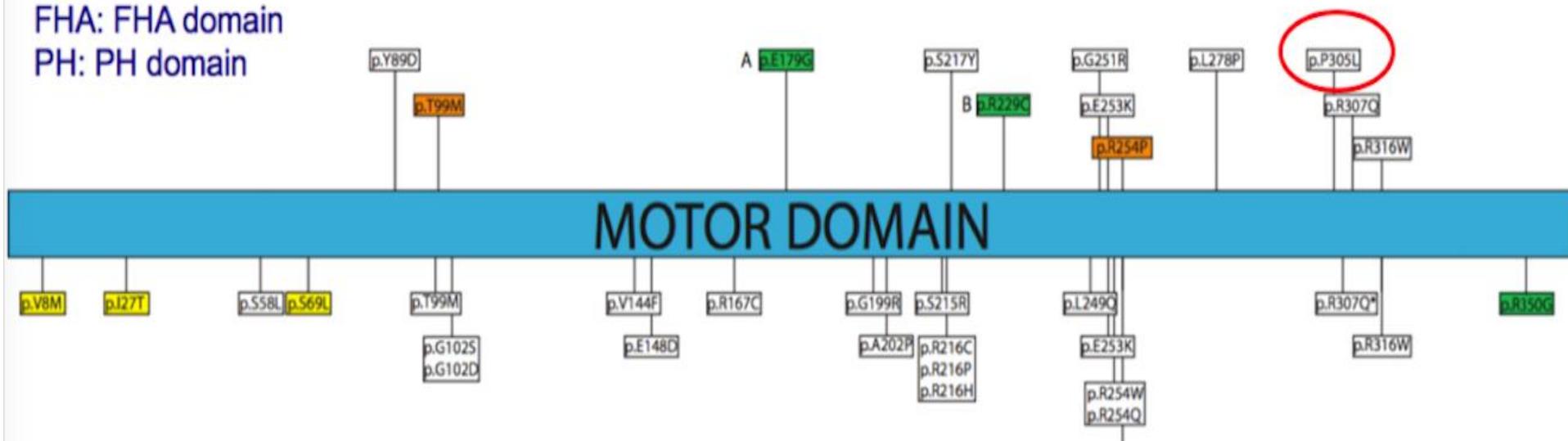
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Understanding the Gene

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CC: Coiled coil
FHA: FHA domain
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