

## Agenda

- 8:30 a.m. Registration**
- 9:00 a.m. Welcome**  
Ronald Bartek — Friedreich's Ataxia Research Alliance (FARA)  
Annie Kennedy — Parent Project Muscular Dystrophy (PPMD)
- 9:15 a.m. Introduction and Demonstration of the Toolkit Portal**  
Petra Kaufmann, MD — NCATS
- 10:00 a.m. Break** (15 minutes)
- 10:15 a.m. Case Study #1**  
Megan O'Boyle — Phelan-McDermid Syndrome Foundation  
Discovery and Pre-Clinical Case Study: Developing a Registry for Phelan-McDermid Syndrome
- 10:45 a.m. Case Study #2**  
Jennifer Farmer, MS, CGC — FARA  
Clinical Trial Readiness Case Study: Friedreich's Ataxia Experience
- 11:15 a.m. Case Study #3**  
Kenneth Hobby — Cure SMA  
Translating Pre-Clinical to Clinical Case Study: Spinal Muscular Atrophy Experience
- 11:45 a.m. Lunch** (1 hour)
- 12:45 p.m. Case Study #4**  
Annie Kennedy — PPMD  
Post-Approval Activities Case Study: Duchenne Muscular Dystrophy Experience
- 1:15 p.m. Discussion Panel**  
Megan O'Boyle, Jennifer Farmer, Kenneth Hobby, and Annie Kennedy
- 2:15 p.m. Break** (15 minutes)
- 2:30 p.m. Next Steps for the Toolkit**  
Group discussion (all participants)  
Moderated by Ron Bartek, Annie Kennedy, Chuck Mohan, and Michelle Campbell (Food and Drug Administration)
- 3:30 p.m. Closing Remarks**  
Ron Bartek, Annie Kennedy, Anne Pariser (NCATS)
- 4:00 p.m. Adjourn**