Successful collaboration between researchers and patient advocacy results in:

1. Effective Study Design
2. Integrated Patient Perspectives
3. Efficient Patient Recruitment and Study Startup

For remote & in-clinic natural history study of rare disease

PATIENT RECRUITMENT

PATIENT ENROLLMENT DETAILS (as of Sept 14, 2021)

<table>
<thead>
<tr>
<th>No. of Patients</th>
<th>Further Enrollment Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>24</td>
<td>Consented and enrolled</td>
</tr>
<tr>
<td>5</td>
<td>Pending remote only study</td>
</tr>
<tr>
<td>4</td>
<td>Unwilling to travel</td>
</tr>
<tr>
<td>1</td>
<td>Pending scheduling</td>
</tr>
<tr>
<td>2</td>
<td>Not interested or didn’t qualify</td>
</tr>
</tbody>
</table>

**STUDY DESIGN & MILESTONES**

**PHASE 1**
AUG 2020 — NOV 2020
- Design – 2 months
- Study visits – 1 month
- Analysis – 1 month

**PHASE 2: Design**
DEC 2020 — MAR 2021
- Contract and IRB approval – 3 months

**PHASE 2: Launch & Enrollment**
MAR 2021 – SEPT 2021
- 24 baseline remote & onsite visits completed by July 2021
- Six-month remote only visits scheduled to begin in October 2021
- Launching arm of study – remote only with no travel involved

**MOTIVATION:** Prospective longitudinal natural history is urgently needed to establish clinical trial readiness for the ultra-rare VCP disease population.

**AIM 1:** Define the natural history of VCP disease

**AIM 2:** Validate data collected via remote versus in-person clinic methods

**AIM 3:** Identify any divergent disease trajectories

**METHODS**
1. Partnered with Alfano Lab at Nationwide Children’s Hospital to design and conduct functional measures study
2. Conducted phase 1 study with 5 diverse patients using tele-health methods
3. Integrated learnings and patient feedback into phase 2 protocol design

**PATIENT ADVOCACY ROLE**
1. Provided patient perspective in study design
2. Advertised study in the patient community
3. Assembled remote patient kits to standardize equipment for measurement
4. Provided patient prep and technology training meeting for remote study
5. Provided study funding and travel stipends to participants for travel to Columbus, OH

**SLEEP 2021 RESULTS**
- Patient recruitment began at IRB approval
- Baseline remote & in-clinic visits launched 3 weeks from start
- Exceeded patient enrollment goal at 8 weeks
- Exploring expansion of study using remote only

**PATIENT REPORTED OUTCOMES:**
- **UPPER EXTREMITY:** Performance of Upper Limb 2.0 (PUL)
  - Hand Grip Dynamometry
  - Nine-Hole Peg Test
  - ACTVE WSV (in-clinic only)
  - Forced Vital Capacity (FVC)
- **LOWER EXTREMITY:**
  - North Star Assessment for limb-girdle type dystrophies (NSAD)
  - Timed Up & Go (TUG)
  - 100-meter walk (in-clinic only)
  - 4-stair climb (in-clinic only)

**SPIROMETRY:**
- Forced Vital Capacity (FVC)
- Forced Expiratory Volume in 1 sec (FEV1)
- MIP/MEP (in-clinic only)

**Cognitive Predictors of Student Success:**
- Non-Cognitive Predictors of Student Success:
- Patern Issues
- Additional Support
- Self-Efficacy
- Motivation
- Study Methods
- Cognitive Methods
- No. of Patients (2021)

**Phenotypes of VCP associated multisystem proteinopathy (MSP)**

**Inclusion Body Myopathy**
- Paget’s Disease of Bone
- Frontotemporal Dementia
- ALS

- **Cure VCP Disease**
  - Rare Disease Coalition
  - Global Genes
  - NORD

- **Study Measures**
  - Rare Disease Coalition
  - Global Genes
  - NORD

- **Cure VCP Disease**
  - Rare Disease Coalition
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