A Message from Dr. John W. Day

We would like to thank you for joining our Neuromuscular Recruitment Database. We are writing to update you on the progress of our Neuromuscular Division over the last year. Our Database aids recruitment to research studies, drug trials, and outreach events. We have now enrolled more than 1100 people, initiated more than 40 studies, and organized multiple conferences and outreach groups annually. Stanford Neuromuscular Research studies have also led to 11 journal publications, and 6 presentations at conferences. Progress in our studies would not be possible without the support of participants and their families, so we appreciate your continued involvement in our research endeavors in 2018.

Our previous year held quite a lot of excitement for our research and clinical teams as we transitioned three research drugs into FDA-approved clinical treatments. The increased use of Spinraza to treat Spinal Muscular Atrophy, Lumizyme to treat Pompe disease, and Eteplirsen to treat some forms of Duchenne Muscular Dystrophy has been a great learning experience on how to transition from implementation of research trials to providing treatment to all. Furthermore, the approval of Soliris (Eculizumab) for Generalized Myasthenia Gravis this past year added a fourth success to our neuromuscular research community. We are committed to transferring our lessons learned during these trials and transitions into improvements in research of other neuromuscular conditions, as well as developing alternative treatments for Spinal Muscular Atrophy such as AveXis’ gene therapy and Roche’s small molecule.

We are poised to have yet another exciting and productive year in 2018, as our team grows and we launch more studies. We have provided a list of projects currently running, and many more will be added over the next few months. We encourage you to stay in touch with our team through the following:

- **Recruitment Database and study questions:** Call our Neuromuscular research phone at (650) 725-4341 or email NeuromuscularResearch@stanford.edu

- **Social Media:** Follow @Stanford_Neuro on Twitter, join “Stanford Neuromuscular Disorders” on Facebook, or visit our “Stanford Neuromuscular Program” YouTube Channel

Again, thank you for joining the Recruitment Database, and we wish you a safe and happy 2018.

Sincerely,

John W. Day, MD, PhD
Research programs organized by condition:

**All Neuromuscular Conditions**
- Recruitment Database: Enrolling people with neuromuscular conditions into a database for recruitment to upcoming studies (recruiting)
- Biobank: Enrolling people with neuromuscular conditions who are interested in donating biological samples (recruiting)
- Exercise tolerance study: Observational study to assess aerobic capacity for people ≥18 years with Pompe Disease or other Neuromuscular Disorders (recruiting)

**ALS**
- Cytokinetics: Drug trial testing safety and efficacy of CK-2127107 in adults with ALS (recruiting)
- CReATe Natural History Study: Observational study of patients with ALS, collecting samples, medical history, and tracking progression (recruiting in clinic)
- Drug trial for adults with C9orf72 ALS mutations (coming soon)

**Charcot-Marie Tooth (CMT)**
- Observational study of symptoms, progression, and genetics of CMT (recruiting in clinic)

**Duchenne Muscular Dystrophy (DMD)**
- Sarepta ESSENCE: Drug trial testing safety/efficacy of drug in boys 7-13 with DMD mutations amenable to skipping exon 45 or 53 (recruiting)
- Roche Spitfire: Drug trial testing safety/efficacy of drug in boys 6-11 with DMD (recruiting)
- Wave: Drug trial testing safety/efficacy of WVE-210201 in boys 5-18 with exon 51-skippable DMD mutations (recruiting)
- PTC: Drug trial testing the long-term outcomes of Ataluren in ambulatory boys age 5+ with nonsense DMD point mutations (recruiting)

**Limb Girdle Muscular Dystrophies (LGMD)**
- COS: Observational study of adults with LGMD2B, measuring symptom progression (recruitment closed, new COS2 study recruiting soon)

**Myotonic Dystrophy (DM)**
- END-DM1: Observational study of symptoms and biomarkers in adults with DM1 (recruiting soon)
- Sleep and gastrointestinal questionnaires for people with DM and family controls (recruiting)
- Drug trial for children with DM1 (coming soon)

**Neuromuscular Junction Conditions**
- Catalyst: Open-label expanded access protocol for Firdapse (Amifampridine Phosphate) treatment in Lambert-Eaton Myasthenic Syndrome (LEMS), Congenital Myasthenic Syndromes (CMS) and Downbeat Nystagmus (recruiting)

**Pompe Disease (PD)**
- Pompe Registry: Observational study tracking outcomes of people with PD (recruiting)
- IPANEMA: Natural history study of possible undiagnosed PD patients (recruiting in clinic)
- Sanofi/Genzyme COMET study: Drug trial testing neoGAA and alglucosidase alfa in treatment-naïve patients with late-onset PD (recruiting)

**Spinal Muscular Atrophy (SMA)**
- AveXis: Gene therapy trials STRONG (kids aged <60 months, recruiting), STRIVE (age<6 months, recruiting closed), and SPRINT (age <6 weeks, recruiting soon)
- Ionis CS11: Drug trial extension measuring safety/efficacy of Nusinersen in children with SMA types 1 and 2 (closed to recruitment)
- Roche: Drug trial of RO7034067 FIREFISH (age <7 months, recruiting) and SUNFISH (recruiting only ages 2-17 with SMA2 or SMA3)
- PNCRN: Observational study of patients with SMA, collecting samples, medical history, and tracking progression (recruiting in clinic)

***For all conditions with studies listed as “closed” and other neuromuscular conditions not listed, we are still enrolling participants in our Recruitment Database for studies and trials that are currently in the planning phase.