



Stanford Neuromuscular Research Program Newsletter: Issue 5

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 helps us recruit individuals to research studies, drug trials, and community conferences. We have now enrolled more than 1500 people, initiated more than 60 studies, and organized multiple conferences and outreach groups annually. Stanford Neuromuscular Research studies have also led to nearly 30 journal publications, and presentations at many international 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 2019.

Our previous year held quite a lot of excitement for our research teams as we increased use of four FDA-approved drugs for various neuromuscular conditions, as well as welcomed in new FDA approvals of Firdapse for Lambert-Eaton Myasthenic Syndrome, Keveyis for periodic paralysis, Soliris for myasthenia gravis, Tegsedi and Onpattro for amyloid neuropathy, and Radicava for ALS. We are now eagerly awaiting the FDA's response to another dramatic leap for Spinal Muscular Atrophy, Zolgensma.

We are also excited about new trials in development to address the root issue of many conditions, as well as gene therapy studies already underway. However we are cautious when technologies have the ability to edit the genome, such as CRISPR-Cas9, as there is a risk of causing unintended mutations in DNA throughout the body, and even in germline cells which have the ability to be passed on to the next generation. Aside from gene therapies, we are motivated to alleviate symptoms of all neuromuscular conditions, and therefore are testing compounds that may improve strength and stamina in many different neuromuscular conditions

As we start new trials, we encourage you to stay in touch with us through:

- **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 2019.

Sincerely,





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)

Amyotrophic Lateral Sclerosis (ALS)

- Cytokinetics: Drug trial testing safety and efficacy of CK-2127107 in adults with ALS (recruitment closed)
- CReATe Natural History Study: Observational study of patients with ALS, collecting samples, medical history, and tracking progression (recruiting in clinic)
- Biogen: Drug trial testing safety and efficacy of BILB078 in adults with C9orf72 ALS mutations (recruiting)

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 (recruitment closed)
- Roche Spitfire: Drug trial testing safety/efficacy of drug in boys 6-11 with DMD (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)

Myasthenia Gravis (MG)

- Two trials starting in 2019

Myotonic Dystrophy (DM)

- END-DM1: Observational study of symptoms and biomarkers in adults with DM1 (recruiting)
- Sleep and gastrointestinal questionnaires for people with DM and family controls (recruiting)
- Pilot trials for sleepiness and brain fog (recruiting soon)
- EEG and spinal fluid studies to define new measures of neurological change (recruiting soon)
- A new drug trial in children with congenital DM (recruiting soon)

Pompe Disease (PD)

- Pompe Registry: Observational study tracking outcomes of people with PD (recruiting)
- Sanofi/Genzyme COMET study: Drug trial testing neoGAA and alglucosidase alfa in treatment-naïve patients with late-onset PD (recruitment closing March 2019)

Spinal Muscular Atrophy (SMA)

- AveXis: Gene therapy trials STRONG (kids aged < 60 months, recruiting reopens soon), STRIVE (age < 6 months, recruitment closed), and SPRINT (age < 6 weeks, recruiting soon)
- Biogen CS11: Drug trial extension measuring safety/efficacy of Nusinersen in children with SMA types 1 and 2 (recruitment closed)
- Roche: Drug trial of RO7034067 FIREFISH (age < 7 months, recruitment closed) and SUNFISH (recruitment closed) and JEWELFISH (ages 6 mo to 60 yrs, recruiting)
- iSMAC: Observational study of patients with SMA, collecting samples, medical history, and tracking progression (recruiting in clinic)
- Scholar Rock: Early trial to make muscle stronger in individuals with type 2 or 3 SMA who are either already on nusinersen or not (recruiting soon)

***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.