A Message from Dr. John W. Day

We thank you for joining our Neuromuscular Database and are writing to update you on our progress during the last year. The Database is essential for research studies, drug trials, and community conferences. We have enrolled almost 1800 people, have more than 50 active studies, and continue to organize many annual conferences and outreach groups. We have published more than 40 articles, and make annual presentations at many national and international conferences. Progress in our studies is impossible without ongoing support of participants and their families, and we greatly appreciate your continued involvement in our research in 2020.

The previous year held a lot of excitement for our clinic and research teams as we increased use of FDA-approved drugs for various neuromuscular conditions, as well as welcoming new FDA approvals of Zolgensma and Evrysdi for spinal muscular atrophy and Vyondys as well as Viltapso for appropriate Duchenne muscular dystrophy patients. These groundbreaking treatments are great news for all neuromuscular disorders, verifying that the same novel treatment approaches work that are in clinical trials or late preclinical studies for Myotonic Dystrophy, SMA, ALS, Myasthenia, LGMD, CMT and many other nerve, muscle and neuromuscular junction disorders.

Active trials include exciting new treatments for Duchenne muscular dystrophy, ALS, SMA, myasthenia gravis and Pompe disease, all of which are teaching us many things that will help in establishing new treatments and will develop new clinical trial methods that will strengthen approaches to all neuromuscular diseases. We are also continuing to learn new lessons from our earlier trials of SMA, which are providing additional approaches to our upcoming studies. Additionally, our unique repository of specimens from SMA, myotonic dystrophy, Duchenne, ALS and many disorders has provided a unique resource for our ongoing work with investigators around the world to better understand neuromuscular disorders and develop measures for upcoming trials.

Research progress was definitely impacted early in 2020 with the outbreak of Covid-19, but we have learned how to safely open our program and many essential drug trials have continued apace through this challenging time. Our team has also learned to drive studies forward by performing regulatory groundwork and virtual assessments from home so that studies can be quickly ramped-up as research restrictions are lifted.

To move new treatment trials forward rapidly, so we can get treatments to patients around the world as soon as possible, we are eager for your help establishing and validating the disease-specific measures needed for all clinical trials of these exciting new treatments.

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To be successful we need the help of you and your family members – affected and unaffected! – to define biological markers that will allow us to measure disease activity and response to treatment. This research obviously can only be done in patients and we’re eager to work on it with you; we would ask for blood or other body fluids – and also benefit from donated tissues from family member who undergo surgery or die of the disease. If you and your family members sign up for our database and our tissue repository, we will let you know the specific studies that are open to you.

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:** 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 2020.

Sincerely,

John W. Day, MD, PhD

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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)
- MDA MOVR registry of some neuromuscular conditions (recruiting in clinic)

**Amyotrophic Lateral Sclerosis (ALS)**
- Alexion: Drug trial testing safety and efficacy of ravulizumab in patients with ALS (recruiting soon)
- Biogen: Drug trial testing safety and efficacy of BIIB078 in adults with C9orf72 ALS mutations (recruiting)
- Biogen: Drug trial testing safety and efficacy of BIIB105 adults with ALS (recruiting soon)
- CReATe CAPTURE: Observational study of patients with ALS (recruiting in clinic soon)
Research programs organized by condition: (continued)

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

**Duchenne Muscular Dystrophy (DMD)**
- Pfizer: Gene therapy for young ambulatory boys with DMD (recruiting soon)
- PPMD registry (recruiting in clinic)
- PTC: Drug trial testing the long-term outcomes of Ataluren in ambulatory boys age 5+ with nonsense DMD point mutations (recruiting)
- Sarepta 4045-302: Testing safety and efficacy of Golodiresen and Casimersen (recruitment closed)
- Wearable Technology to Assess Gait Function in SMA and DMD (recruiting soon)

**Facioscapulohumeral Dystrophy (FSHD)**
- Natural history study (recruiting soon)

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

**Myasthenia Gravis (MG)**
- Alexion: Drug trial testing safety and efficacy of ravulizumab in adults with generalized MG (recruiting)
- ArgenX: Drug trial testing safety and efficacy of ARGX-113 in adults with generalized MG (recruiting closed)
- Takeda: Drug trial testing safety and efficacy of TAK-079 in adults with generalized MG (recruiting soon)

**Myotonic Dystrophy (DM)**
- AMO: Drug trial testing safety and efficacy of Tideglusib in children with congenital DM (recruiting soon)
- END-DM1: Observational study of symptoms and biomarkers in adults with DM1 (recruiting)
- Stanford: EEG and spinal fluid studies to define new measures of neurological change (recruiting)

**Pompe Disease (PD)**
- Audentes Seroprevalence Study: Examining the prevalence of antibodies to AAV8 (recruiting)
- Audentes FORTIS: Drug trial testing safety and efficacy of AT845, an AAV8-Delivered Gene Transfer Therapy in Patients With Late Onset PD (recruiting)
- Pompe Registry: Observational study tracking outcomes of people with PD (recruiting in clinic)
- Sanofi/Genzyme COMET study: Drug trial testing neoGAA and alglucosidase alfa in treatment-naïve patients with late-onset PD (recruiting closed)

**Spinal Muscular Atrophy (SMA)**
- AveXis: Gene therapy trials STRONG (kids aged <60 months, recruiting reopens soon), STRIVE (age<6 months, recruiting closed)
- Biogen DEVOTE: Drug trial testing safety and efficacy of higher doses of nusinersen (recruiting)
- Biogen SHINE: Drug trial extension measuring safety/efficacy of Nusinersen in children with SMA types 1 and 2 (recruiting closed)
- CureSMA registry (recruiting in clinic)
- iSMAIC: Observational study of patients with SMA, collecting samples, medical history, and tracking progression (recruiting in clinic)
- Roche: Drug trial of RO7034067 FIREFISH (age <7 months) and SUNFISH (age 2 to 25) and JEWELFISH (ages 6 mo to 60 yrs) (recruiting closed)
- Scholar Rock: Drug trial testing safety and efficacy of SRK-015 in SMA (recruiting closed)
- Stanford: Observational study of outcomes in adults with SMA (recruiting in clinic)
- Wearable Technology to Assess Gait Function in SMA and DMD (recruiting soon)

***For all conditions with studies listed as “recruiting 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.