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Stanford Neuromuscular Research Program Newsletter: Issue 1

A Message from John W. Day, MD, PhD

We would like to take a minute to thank you for joining our Stanford Neuromuscular Recruitment Database, and update you on the progress of our Neuromuscular Division over the last year. Our Database was created in 2013 to aid recruitment to research studies, drug trials, and outreach events. We have enrolled more than 380 people, initiated more than 20 studies and trials, and organized several conferences and outreach groups.

We are poised to have an even more exciting and productive year in 2015, as our team grows and we launch more studies and trials. A list of projects is found on the back of this page. We encourage you to stay in touch with our team through the following:

- **Recruitment Database and study questions** - Contact Katharine Hagerman at (650) 723-9574 or email khagerma@stanford.edu.

- **Social Media** - Join our "Stanford Neuromuscular Disorders" group on Facebook or stay tuned as we post videos to our new "Stanford Neuromuscular Program" YouTube Channel.

Again, thank you for joining the Recruitment Database, and we wish you a safe and happy 2015.

Sincerely,

John W. Day, MD, PhD

Dr. Day is a Professor of Neurology, Pediatrics and Pathology at Stanford University. He is focused on both improving clinical care and driving research in the field of neuromuscular disorders.

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Research studies, programs, and trials 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)

ALS

- Genetics of ALS: Collecting DNA from adults to identify genes influencing ALS (recruiting soon)
- DPS: Determining whether diaphragm pacing systems improve survival or function in adults with ALS (recruiting)

Charcot-Marie Tooth (CMT)

- Development/validation of Disability Severity Index by retrospective chart review (closed)
- Chart review of pulmonary functions (closed)
- Observational study of symptoms, progression, and genetics of CMT (recruiting)

Congenital Myopathies

- Valerion study: Observational study of symptom progression in adults with Myotubular Myopathy (recruiting)
- Audentis study: Retrospective chart review of adults with myotubular myopathy (closed)
- Nemalin myopathies: Recruiting to our database for upcoming studies

Duchenne Muscular Dystrophy (DMD)

- CINRG: Collecting samples from boys with DMD to better understand the condition and aid therapy development (recruiting)
- Sarepta: Drug trial testing safety/efficacy of Eteplirsen in boys aged 7-16 with DMD (recruiting)

Facioscapulohumeral muscular dystrophy

- Recruiting to our database for upcoming studies

Limb Girdle Muscular Dystrophies (LGMD)

- COS: Observational study of adults with LGMD2B, measuring symptom progression (closed)
- LGMD 2C and 2D: recruiting to our database for upcoming studies

Myotonic Dystrophy (DM)

- DMCRN: Observational study of symptoms and biomarkers over 1 year in adults with DM1 (recruiting)
- Isis CS2: Drug trial testing safety/efficacy/dose of DMPKRx in adults with DM1 (recruiting)
- CHRI: Observational study of children ages 8-17 with DM1, measuring neuropsychological symptoms (recruiting)
- Sleep Study: Observational study of sleep and neuropsychological symptoms of adults with DM1 (recruiting soon)
- Metabolism: Observational study of metabolism in adults with DM (recruiting)

Neuromuscular Junction Conditions

- Lamberton-Eaton Myasthenic Syndrome (LEMS) trial: Drug trial testing safety/efficacy of Amifampridine Phosphate in adults with LEMS (closed)
- Myasthenia Gravis (MG): Alexion Drug trial testing safety/efficacy of Eculizumab in adults with refractory generalized MG (recruiting soon)

Pompe Disease (PD)

- Pompe Registry: Observational study tracking outcomes of people with PD (recruiting)

Spinal Muscular Atrophy (SMA)

- Isis CS3B: Drug trial testing safety/efficacy of SMNRx in babies ≤6 months with SMA type I (recruiting)
- Isis CS4: Drug trial testing safety/efficacy of SMNRx in children ages 2-12 with SMA type II (recruiting)
- Isis CS3A (closed)
- PNCRN: Observational study of people with SMA, collecting samples, medical history, and tracking progression over 3 years (recruiting soon)

***For all conditions with studies listed as “closed”, we are still recruiting to our database for many studies starting in the next year that may not be listed