2019 Bay Area FSHD Family Day Conference
Li Ka Shing Center, Stanford Medical School
Sunday, April 28, 2019, 1:00 to 6:00 pm

Agenda

1:00 – 1:30 pm	**Welcome.** Mark Stone, CEO of FSH Society, & John Day, MD PhD, director of Stanford FSHD research center • Ice-breaker activity
1:30 – 3:00 pm	**Advances in FSHD treatments** (20 mins for talks + 30 mins Q&A) • Getting ready for FSHD clinical trials, Jeff Statland, MD • Why we think FSHD is treatable, Stephen Tapscott MD PhD, • FSHD research at Stanford, Antoine de Morree, PhD • Q&A
3:00 – 3:15 pm	**Break**
3:15 – 4:00 pm	**Best medical practices for FSHD** • FSHD 101 – Jacinda Sampson, MD PhD • Genetic diagnosis – Carly Siskind, MS LCGC • Q & A
4:00 – 5:00 pm	**Living with FSHD** • Speech and swallow issues – Sarah Stranberg, CCC-SLP • Strength and Function: Updates on iFSHD study and staying active throughout the lifespan – Tina Duong, MPT • Well-being in chronic disease - Kent Drescher, PhD • Q & A
5:00 – 5:30 pm	• **Mark Stone.** Our vision and strategy • **Kent and Sue Drescher.** Bay Area Chapter Directors call to action • **Q&A**
5:30 – 6:00 pm	**Reception**

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Bay Area FSHD Family Day Conference Speaker Profiles

John Day, MD PhD, is professor of neurology and neurological sciences and director of the Stanford Neuromuscular Disorders Program. He has over 25 years of experience in diagnosing, treating and supporting patients with neuromuscular diseases. He is involved in ongoing research defining causes, diagnosis and novel treatments of genetic neuromuscular disorders. Prior to his arrival at Stanford in 2011, Dr. Day directed the Paul and Sheila Wellstone Muscular Dystrophy Center at University of Minnesota. Dr. Day graduated from the Albert Einstein College of Medicine and completed his Residency and Fellowship at University of California Medical School in San Francisco. Dr. Day is Board Certified in Neurology by the American Board of Psychiatry and Neurology.

Antoine de Morree, PhD, is a postdoctoral fellow in the laboratory of Tom Rando at Stanford University, and instructor in neurology and neurological sciences. His research interest is in the molecular mechanisms underlying neuromuscular disorders and the molecular regulation of satellite cell quiescence and activation in relation to normal aging.

Kent Drescher, PhD, is a licensed clinical psychologist. He recently retired from many years of service at the National Center for PTSD, which is part of the part of the VA Health Care System. In his role there he conducted research, provided clinical care for veterans, and educated clinicians about post-traumatic stress. Dr. Drescher has FSHD and personally understands the physical and emotional impact the disease has had on him and his family. His clinical specialty is Acceptance and Commitment Therapy, which is a trans-diagnostic behavioral treatment designed to increase human flourishing in the face of diverse life challenges.

Tina Duong, MPT, is a research physical therapist at Stanford with over 10 years of clinical experience in neuromuscular and pediatric research. She is instrumental in optimizing clinical endpoints for study design and assessments through the CINRG network and research in exercise regimens in mouse models of muscular dystrophy. She trains and educates clinicians worldwide on implementing and designing clinical outcome measures for international multi-site trials. She is a member of the International Outcomes Working Group in which she collaborates with experts to assess and develop novel outcomes and approaches toward function-based outcome measures in Duchenne muscular dystrophy. She continues to work on outcomes development with neuromuscular diseases and pursues her work on the benefits of rehabilitation and exercise as conjunctive therapies.

Jacinda Sampson, MD PhD, received her MD and a PhD in biochemistry from University of Alabama at Birmingham, and completed her neurology residency and neurogenetics fellowship at the University of Utah. She served at Columbia University Medical Center prior to joining Stanford University Medical Center in 2015. Her areas of interest include myotonic dystrophies, Duchenne muscular dystrophy, and neurogenetic disorders such as neurofibromatosis, hereditary spastic paraparesis, spinocerebellar ataxia, among others. She is interested in clinical

trials for treatment of neurogenetic disorders and is the clinical application of next-generation genomic sequencing to genetic testing.

Carly Siskind, MS LDGC, is Senior Genetic Counselor and Clinical Associate Professor (Affiliated) at the Stanford Neuromuscular Disorders Program. She sees patients both at Stanford Hospital and Clinics and Lucile Packard Children's Hospital. Her main research focus being Charcot-Marie-Tooth disease. Ms. Siskind obtained her Bachelor's degree from the University of Michigan with a major in Biology, minor in Global Change and a teaching certificate for high school science. She obtained her master degree from Northwestern University in Chicago. She was board certified by the National Society of Genetic Counselors in 2009 and licensed by the state of California in 2011.

Jeffrey Statland, MD, is Associate Professor of Neurology with tenure at the University of Kansas Medical Center. His research has centered on describing the natural history of and response to therapy of neuromuscular diseases. He currently serves as principal investigator for research studies in FSHD, DMD, SMA, and Myotonic Dystrophy. His specific research interest over the last 8 years has been preparing for clinical trials in FSHD. He has worked with collaborators to develop new disease-relevant outcome measures to assess biomarkers, physiological changes in muscle structure or function, and patient-reported FSHD impact. Currently he has assembled an FSHD Clinical Trial Research Network, comprised of 8 U.S. academic centers and three collaborating sites in Europe, and a research team with decades of experience in conducting clinical trials, with the overarching goal of hastening therapeutic development for FSHD.

Mark Stone is President and CEO of the FSH Society. He joined the Society in 2017. Mark has served as an executive leader of research-focused patient advocacy nonprofit organizations for the past 13 years. Prior to joining the FSH Society, he was the chief executive officer of NephCure Kidney International. Passionate about helping organizations work to accelerate treatments and cures in diseases that are both rare and of a genetic origin, Stone has successfully engaged patients, families and friends in raising up an "army of activists" while advocating with pharmaceuticals, the National Institutes of Health (NIH), and the FDA to increase funding levels and enlarge the pipeline of potential therapies.

Sarah Stranberg, CCC-SLP, is a senior speech-language pathologist in the Outpatient Neurologic Rehabilitation Program at Stanford Health Care. She has degrees in communication sciences and disorders from Northwestern University and the University of Minnesota. Sarah has extensive experience and interest in working with individuals with communication and swallowing disorders due to neurologic conditions, including ALS, Myasthenia Gravis, brain injury, stroke and Parkinson's disease. Sarah is dedicated to helping individuals with neuromuscular disorders and their families achieve their greatest functional abilities and quality of life. She enjoys involvement with activities outside of clinic such as support groups, professional conferences, and community events. In 2005, the Muscular Dystrophy Association presented Sarah with the Leon Poliachik Humanitarian Award for her comprehensive care and work with individuals with neuromuscular diseases.

Stephen Tapscott, MD PhD, is a board-certified neurologist and associate director of the University of Washington Medical Scientist Training Program, a full member in the Human Biology and Clinical Research divisions at Fred Hutchinson Cancer Research Center and a UW professor of Neurology and adjunct professor of Pathology. Dr. Tapscott has clinical expertise in neuromuscular and neurogenetic diseases. He strives to create active partnerships with his patients to achieve the best possible outcomes. Dr. Tapscott's research team broke a decades-long stalemate with FSH muscular dystrophy when they identified the mechanisms that cause it. This leads to the possibility of developing future treatments for the disease and potentially some cancers.