Virtual FSHD 360 Conference
University of Florida, Gainesville
Saturday, June 12, 2021, 10:00 am to 2:00 pm

10:00—10:05 am	**Welcome** Mark Stone, CEO, FSHD Society
10:05—10:25 am	**FSHD 101** S. Subramony, MD, University of Florida
10:25—11:35 am	**Genetics of FSHD** Lee Kugelmann, University of Florida
	New Approaches to FSHD Diagnostic Testing Erin Beaver, PerkinElmer Genomics
	Rehabilitation and FSHD Rachael Dorsey, MOT, OTR/L, University of Florida
	Q&A Panel with all of the morning speakers
11:35 am—12:05 pm	**Lunch break and chat about nutrition** Carley Rusch, nutrionist
12:05—12:25 pm	**Magnetic resonance imaging in muscular dystrophies** Krista Vandenborne, PhD, University of Florida
12:25—1:35 pm	**FSHD Research at UF and beyond** Eric Wang, PhD, University of Florida
	Small molecule therapies for FSHD Fran Sverdrup, PhD, St. Louis University
	RNA therapeutics for FSHD Kelly DiTripani, VP Medical Affairs, Avidity Bioscience
	Q&A with afternoon speakers
1:35—1:55 pm	**Activating our Florida communities** Beth Johnston and Anna Gilmore, FSHD Society Heloise Hoffmann, Jane Pollock, Christina Thissen, chapter directors
1:55—2:00 pm	**Concluding remarks** Mark Stone, CEO, FSHD Society
2:00-3:00 pm	**Social time**

FSHD 360 @ University of Florida, June 12, 2021

About our speakers

FSHD 101, S. Subramony, MD
Dr. Subramony is professor of neurology at the McKnight Brain Institute at the University of Florida and professor of pediatrics at the University of Florida College of Medicine. He is co-director of the MDA Clinic at UF College of Medicine and principal investigator for the MOVE study at the FSHD Clinical Trial Research Network site at UF. FSHD was initially recognized as a distinct muscular dystrophy apart from Duchenne dystrophy in the 1880's by two physicians, Landouzy and Dejerine and for this reason was named Landouzy Dejerine disease. For close to 100 years, research described families and studies using available technologies such as EMG and muscle biopsy but this led to little understanding of the real issues. Typically, the disorder begins with weakness of facial muscles, then shoulder and upper arm muscles progressing to muscles over the stomach, spine and legs. The majority do not progress to the need for a wheelchair and significant problems other than in muscles are uncommon other than perhaps hearing loss. In the 1990s, the genetic abnormality in the vast majority of FSHD families was found to be an unusual shortening of a repetitive element called D4Z4 repeat at one end of chromosome 4 with normal persons carrying 11 or more repeats and FSHD patients, 10 or fewer repeats. The size of the repeat influences some aspects of severity but not all. As real designer therapies become feasible, clinical research needs to understand the rate of progression of the disease better so that the effectiveness of drugs can be proven. Development of biomarkers of the disease also is a goal of clinical research. Ongoing natural history study under the auspices of the FSHD CTRN hopes to accomplish this and be ready for trials.

Genetics of FSHD, Lee Kugelmann, MMSc, CGC
Lee Kugelmann is a certified genetic counselor with the University of Florida Department of Neurology. She works in the Neuromuscular and Movement Disorders divisions, providing education and support to patients and families with genetic diagnoses. She also works with UF's Clinical Genomics Program to bring whole genome sequencing to the clinic. Her talk with discuss the genetic mechanisms of FSHD, how FSHD is inherited and what that means for other family members, and how to pursue genetic testing.

New Approaches to FSHD Diagnostic Testing, Erin Beaver, MS, CGC
Erin Beaver is a genomic testing consultant and licensed certified genetic counselor with PerkinElmer Genomics. Erin has over 8 years of clinical genetic counseling experience in addition to her experience in the laboratory industry. Erin is a member of the Advisory Board for Heartland Regional Genetics Network and a member of the Advisory Board for the Genetic Counseling training program at Washington University in St. Louis. She serves as an Adjunct Instructor and thesis advisor with the Genetic Counseling training program at the University of Arkansas for Medical Sciences. Erin's research interests include developing innovative models of care in genetic services, healthcare communication, and addressing disparities in healthcare and patient accessibility to genetic services.

Rehabilitation and FSHD, with Rachael Dorsey, MOT, OTR/L
Rachael Dorsey has been providing occupational therapy for adults with neurological conditions for 13 years in inpatient and outpatient rehabilitation settings. In the last 2 years, she has specialized in treating neuromuscular diagnoses (muscular dystrophy, ALS and others), movement disorders, and brain injury. In her talk, she will discuss the benefits of occupational, physical, and speech therapy for managing quality of life throughout the lifespan in people with FSHD. What to expect in regard to evaluation and treatment from your neurological therapists to maintain function, address self-care, daily activities, mobility, speech, and swallowing.

Talking about nutrition, Carley Rusch
During this lunch break chat, nutritionist Carley Rusch will discuss principles for a healthy diet for individuals living with FSHD. Carley received a bachelor's degree in Food Science and Human Nutrition from the University of Florida and remained at UF to complete her master's degree and dietetic internship. After becoming a clinical dietician and working at AdventHealth in Orlando, Carley was recruited to move back to UF to work toward a PhD in Nutritional Sciences and facilitates an outpatient nutrition program at the UF Health Norman Fixel Institute for Neurological Diseases. Currently, Carley splits her time between clinical care and research. Her research focus is related to the role of diet and gastrointestinal health in neurological diseases.

Magnetic resonance imaging in muscular dystrophies, Donovan Lott, PT, PhD, CSCS, and Krista Vandenborne, PhD

Donovan J. Lott is a Research Associate Professor within the Department of Physical Therapy at the University of Florida. He completed his undergraduate degree in Exercise Science at Brigham Young University and his MSPT degree at Washington University School of Medicine. After working in both outpatient and home-health settings as a physical therapist, he returned to Washington University to obtain a PhD in Movement Science with an emphasis in soft tissue mechanics. Donovan then went to the University of Florida (UF) as a post-doctoral fellow to work with Krista Vandenborne, PhD, PT, in the Muscle Physiology Laboratory. He has spent the past 18 years working in clinical research focused on patients with various forms of neuromuscular disease. At UF, his primary research has involved patients with muscular dystrophy (mostly Duchenne muscular dystrophy and Myotonic Dystrophy Type 1). His overall research goals are to better understand the impact of muscular dystrophy pathophysiology on skeletal muscle and its impact on functional mobility and to investigate optimal exercise prescriptions to minimize these deleterious effects and maximize independence and safety in this patient population.

Dr. Vandenborne is Distinguished Professor and Chair of the Department of Physical Therapy at the University of Florida. Her research has focused on the implementation of magnetic resonance imaging (MRI) and spectroscopy (MRS) to characterize skeletal muscle in disease. Her laboratory has applied Magnetic Resonance to the study of skeletal muscle in numerous animal models and a variety of patient populations. Over the last twenty years, she has been leading the efforts at the University of Florida to develop a strong translational research program dedicated to the development and validation of MR imaging biomarkers for muscular dystrophy. Dr. Vandenborne has created a large MR imaging clinical trial network for muscular dystrophies, with over 30 imaging sites across the US and Europe to include MR imaging biomarkers in phase 1-3 clinical trials.

FSHD Research at UF and beyond, Eric Wang, PhD
Dr. Wang is an Associate Professor in the Department of Molecular Genetics & Microbiology and the Center for NeuroGenetics at the University of Florida. His laboratory applies molecular, cellular, and computational approaches to study muscle and nervous system diseases, including FSHD, Myotonic Dystrophy, and ALS. He is personally motivated in this area because he has family members affected by genetic neuromuscular disease and works closely with academic, industry, and patient advocacy partners to translate scientific advances into therapies. He will provide an overview of FSHD research and various therapeutic approaches that are currently being developed.

Small molecule therapies for FSHD, Fran Sverdrup, PhD
Dr. Sverdrup is an associate professor in the Department of Biochemistry and Molecular Biology, Saint Louis University School of Medicine. His lab performs target identification and validation, drug screening, mechanism of action studies and preclinical evaluation of drug candidates. Currently, his research is

focused on drug discovery and the epigenetic control of gene expression in FSHD. His lab has employed chemical genetics and candidate gene knockdowns to identify druggable pathways that regulate DUX4 expression. For promising therapeutic approaches, a mouse xenograft model of human FSHD is utilized to establish pharmacokinetic/pharmacodynamic relationships of drug candidates. The xenograft model conserves the primate-specific DUX4 locus and allows evaluation of drugs that work at an epigenetic level.

RNA therapeutics for FSHD, Kelly DiTrapani, RN, BSN, BA
In September of 2020, Kelly DiTrapani joined Avidity Biosciences as Vice President of Medical Affairs. Kelly brings extensive commercial and medical affairs leadership in the life sciences industry, focused on bringing innovative therapeutics to patients in the oncology and rare disease spaces. Kelly received both her Bachelor of Arts and Bachelor of Science in Nursing from Duke University. Avidity Biosciences is a biotechnology company founded in 2012 and based in La Jolla, California. The company's mission is to profoundly improve people's lives by revolutionizing the delivery of RNA therapeutics. Avidity is pioneering a new class of RNA therapeutics called antibody oligonucleotide conjugates (AOCs), which combine the specificity of monoclonal antibodies with the potency and precision of oligonucleotides. The company is initially focused on advancing and expanding its pipeline in untreated rare muscle diseases, including FSHD. Kelly will share an overview of Avidity's AOC program in FSHD.

FSHD Society Team

Mark Stone, President & CEO
Mark has served as an executive leader of research-focused patient advocacy nonprofit organizations for the past 18 years. Prior to joining the FSHD Society, he was the chief executive officer of NephCure Kidney International. From 2004-2011, he was executive vice president and COO of the Polycystic Kidney Disease (PKD) Foundation. He has also served as the deputy director of an international relief and development organization and as a pastor within the Nazarene Church. Passionate about helping organizations work to accelerate treatments and cures in diseases that are both rare and of a genetic origin, he 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.

June Kinoshita, Director of Research and Patient Engagement
June joined the FSHD Society in 2012 and served as its Executive Director until September of 2017. Previously, June co-founded and served as Executive Editor of the Alzheimer Research Forum, the pre-eminent Web community for researchers in neurodegenerative disorders. June has worked closely with a variety of foundations to develop initiatives for multiple sclerosis, schizophrenia, amyotrophic lateral sclerosis, Parkinson's disease, and other disorders. She co-founded N-of-One, Inc., a pioneering targeted oncology company. June began her career as a science journalist, working as a writer and editor for *Scientific American*, *Science*, *The New York Times Magazine,* and many other national publications.

Jamshid Arjomand, PhD, Chief Science Officer
A neuroscientist with more than 15 years of pharmaceutical and biotechnology experience in chronic pain, neurodegeneration, neuromuscular disorders and human stem cell disease modeling, Jamshid came to the FSHD Society from Genea Biocells, a San Diego-based biotechnology company where he served for five years as Vice President of Business Development. Genea's pipeline included FSHD for which their lead asset, GBC0905, received orphan drug designation by the FDA in May 2018. From 2005 to 2013, he served as Director of Basic Research at CHDI Foundation. There he designed and managed a complex portfolio of

academic, clinical and industry driven projects, primarily related to biomarker discovery, stem cell development, and target discovery and validation efforts for Huntington disease.

Beth Johnston, Chief of Community Development
Beth joined the FSHD Society staff in August of 2016. Her long history with the Society began as a volunteer shortly after her husband was diagnosed with FSHD. She continues her commitment to raising awareness of the disease and building an "army of activists" that will help us achieve our mission of treatments and a cure. Prior to joining the Society, she was Founder & CEO of Social Bridges, a Denver-based social media marketing agency. She has also worked in information technology, project management, high-technology & telecommunications consulting, and real estate.

Anna Gilmore, Regional Director, Community Development
Anna joined the FSHD Society in March 2018. She comes to the organization from Northeastern University, where she worked in the City & Community Affairs division for the last 7 years. Her projects there included implementing co-curricular volunteering programs and facilitating student leadership development and training with a focus on civic engagement and social justice.

Christina Thissen, Northeast Florida chapter director
Christina was diagnosed with FSHD in 1998 when she was 19 years old. She owns an IT Managed Services company in Jacksonville with her big brother who also has FSHD. She looks forward to bringing others together in our community to share stories and resources.

Heloise Hoffmann, Southwest Florida chapter director
Heloise was diagnosed with FSHD at age 13 and is a co-convener of the GenZ of FSHD group. She says, “From the beginning, I knew I wanted to do something about [FSHD advocacy] and that I couldn’t just sit around and feel sorry for myself. The FSHD Society has given me so much hope, and I’m excited to give back and share this hope by leading the Southwest Florida Chapter.”

Jane Pollock, Tampa chapter director
Jane’s husband, George was diagnosed in 2006. He is the first person in his family to have FSHD. “We found the FSHD Society to be a great resource and found support among its members,” says Jane. “We wanted to help spread the word to other patients and their families about the work being done by the society. George is involved on the FSHD Society’s Board, and I wanted to contribute in my own way towards connecting families and helping to find and fund a cure.”