

Miami FSHD Family Day Conference
Saturday, February 23, 2019, 12:00 pm to 6:00 pm
Newman Alumni Center University of Miami
6200 San Amaro Drive, Coral Gables, FL 33146

Agenda

- Noon-1:00 pm **Informal welcome. Lunch** (included with registration)
- 1:00 pm—1:05 pm **Welcome**
Mario Saporta, MD PhD
Mark Stone, CEO of the FSH Society
- 1:05-2:00 pm **Best practices in caring for FSHD** (15 mins each + 10 mins Q&A)
- **FSHD 101 – What every patient needs to know**
Mario Saporta, MD PhD
 - **Genetic testing and counseling**, Stephanie Bivona, MS, CGC
 - **Physical therapy and exercise**, Kayla Chomko, DPT
- 2:00-3:00 pm **Panel session and patient roundtable**
- Your legal rights—Matt and Debbie Dietz
 - Assistive devices and technology – Gene Harkey
- 3:00-3:15 pm **Break**
- 3:15 – 4:15 pm **Advances in FSHD treatments** (45 mins for talks + 15 mins Q&A)
- **Clinical trials today**, Mario Saporta, MD PhD
 - **The Future of FSHD therapies**, Peter Jones, PhD
 - **An engaged and empowered community**
June Kinoshita, Chief Strategic Programs Officer, FSH Society
- 4:15-5:00 pm **Family: Living, loving, caring, sharing**
Ora and Isaac Prilleltensky, introductory remarks
Open conversation with all attendees
- 5:00-5:15 pm **What’s next for the Miami community? Vision and strategy**
Mark Stone, President and CEO, FSH Society
- 5:15-6:00 pm **Reception and social hour**

Miami FSHD Family Day Speaker Bios

Stephanie Bivona is a board-certified genetic counselor who specializes in neurogenetics. She is the genetic counselor for the Muscular Dystrophy Association clinic at the University of Miami. She will be discussing the role of a genetic counselor for families with FSHD, genetic causes of FSHD, and genetic testing for FSHD.

Kayla Chomko, DPT, received her bachelor's degree from the University of Florida in 2005 in the area of Health Science and earned her doctor of physical therapy degree from the University of Miami in 2008. Since graduation, Dr. Chomko has focused on the treatment of neurological disorders, specifically working with patients and loved ones affected by neurodegenerative diseases. Kayla is the lead physical therapist within the Neurology department with a focus on muscular dystrophies, ALS and CMT among many other neurological conditions. She joined University of Miami Department of Physical Therapy as voluntary adjunct assistant professor and is assisting with establishing their Neurological Residency program. Kayla is eager to educate the community regarding how physical therapy can assist patients with FSHD and best practices within the field regarding this diagnosis.

Matthew and Deborah Dietz are the founders of Disability Independence Group (DIG), a nonprofit advocacy center for disability rights. The mission is to expand opportunities for participation, education, employment, and acceptance of persons with disabilities through advocacy, litigation, education, and training. Matthew Dietz, litigation director, has been practicing in the arena of civil rights litigation since 1996 and has handled hundreds of matters involving the Americans with Disabilities Act, the Fair Housing Act and other civil rights laws. Deborah Dietz, executive director, is the creator of The Wallet Card Project that helps individuals with intellectual/developmental disabilities safely disclose their disability to law enforcement and is working on a project that looks at the intersection of domestic violence/sexual assault and disability.

Peter L. Jones, PhD, is the Mick Hitchcock Endowed Chair of Medical Biochemistry and Associate Professor of Pharmacology at the University of Nevada, Reno School of Medicine. He earned a bachelor's degree in microbiology from Miami University in Oxford, OH, and then his PhD in genetics and molecular biology at Emory University School of Medicine in Atlanta, GA. He went on to a post-doctoral fellowship at the National Institutes of Health, Bethesda, MD, where he expanded his training into biochemistry, chromatin and epigenetics. Since 2003, Dr. Jones' lab has been investigating epigenetic dysregulation underlying FSHD pathogenesis to identify therapeutic targets, develop more affordable and accessible diagnostics, and aid in the generation of cell and animal models of the disease for better therapeutic development and pre-clinical testing. He currently runs the lab with his wife and Co-PI, Dr. Takako Jones.

June Kinoshita is Chief Strategic Programs Officer at the FSH Society. She joined the Society in 2012. Her focus is on the intersection of patient engagement and therapy development. As part of the Society's research team, she works closely with academic and industry partners on the Therapeutic Accelerator project. She organizes FSHD Family Day conferences across the U.S. as

well as the annual FSHD International Research Congress. June is also Editor of the FSHD Advocate and the FSH Society's educational publications, website and social media content.

Ora Prilleltensky, PhD, obtained her doctorate in counseling psychology from OISE at the University of Toronto. She is the former director of the major in Human and Social Development at the University of Miami and has taught various graduate and undergraduate courses. She has also worked in a variety of clinical settings, including a child guidance clinic, a university counseling center and a rehabilitation hospital. She is the author or co-author of four books dealing with well-being, change, and disability.

Isaac Prilleltensky, PhD, is the former dean of the School of Education and Human Development at the University of Miami. He currently serves as the vice provost for institutional culture at the University of Miami, where he is also a professor of educational and psychological studies. He is a community psychologist with interests in the connection between wellness and fairness. He has published ten books and over 130 scientific papers and chapters. He is the recipient of awards by the American Psychological Association for his scholarly work and by the National Newspaper Association for his humor writing.

Based on their research, recent books on well-being and change, and on their family and professional experience, the Prilleltenskys will lead a discussion on the emotional and relational side of FSHD. Ora, who uses an electric wheelchair, was diagnosed with FSHD at age 18. She and Isaac have been married for 36 years. For them, FSHD is a family affair that is best managed within the context of a loving and supportive relationship. The goal of the session is to provide a safe space for participants to share experiences, connect with others, and explore solutions that fit their needs and circumstances.

Mario Saporta, MD, PhD, MBA is an Assistant Professor of Neurology and Human Genetics at the University of Miami. He is the director of the Muscular Dystrophy Association Care Center and Charcot-Marie-Tooth Center of Excellence at the University of Miami where he coordinates a multidisciplinary team providing specialized care for patients with neuromuscular genetic disorders. He is also a principal investigator leading a research group focused on the use of human stem cells to study neuromuscular genetic conditions. He will review the basics of FSHD and provide an update on the current clinical trials 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.