

**FSHD 360 Conference University of Colorado**

Douglas County Library, 20105 East Mainstreet, Parker, Colorado

Saturday, August 28, 2021, 10:00 am to 4:00 pm

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| 10:00-10:30 am | <b>Check in</b>  |
| 10:30—10:35 am | <b>Welcome</b><br>Mark Stone, CEO, FSHD Society  |
| 10:35—10:55 am | <b>FSHD 101</b><br>Matthew Wicklund, MD, University of Colorado  |
| 10:55—11:15 am | <b>Genetics of FSHD and the role of the genetic counselor</b><br>Kaitlin Smith, University of Colorado   |
|                | <b>Advances in genetic testing technology</b><br>Mary Colasanto, PhD, PerkinElmer Genomics   |
| 11:15-11:30 am | <b>Q&amp;A with all morning speakers</b>   |
| 11:30—12:15 pm | <b>Multidisciplinary panel with</b><br>Matthew Wicklund, MD, moderator<br>Physical therapy and exercise – Hannah Dwight<br>Occupational therapy – Karen Hookstadt<br>Mental health – Kristin Orłowski, Ph.D. |
| 12:15—1:00 pm  | <b>Lunch and group conversation</b>  |
| 1:00—1:20 pm   | <b>FSHD research at UC</b><br>Suja Jagannathan, PhD, University of Colorado  |
| 1:20—1:40 pm   | <b>The MOVE study and other clinical research at UC</b><br>Brianna Blume, University of Colorado   |
| 1:40-1:55 pm   | <b>Break</b>   |
| 1:55—2:55 pm   | <b>Drug development panel</b><br>Jenn McNary, Fulcrum Therapeutics<br>Lisa Ackermann, Avidity Biosciences<br>Molly White, Dyne Therapeutics  |
| 2:55—3:25 pm   | <b>Activating our Colorado community</b><br>Beth Johnston, FSHD Society, and Amanda Hill, chapter director   |
| 3:25—3:30 pm   | <b>Concluding remarks</b><br>Mark Stone, CEO, FSHD Society   |
| 3:30—4:00 pm   | <b>Social time</b>   |

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Speaker Bios



**Mark Stone, President & CEO, FSHD Society.** 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.



**Matthew Wicklund, MD, University of Colorado.** Dr. Matt Wicklund is the chief of the Neuromuscular Section and director of the Neuromuscular Pathology Lab, Clinical Neuromuscular Pathology Fellowship, Neuromuscular & Rare Disease Genetic Counseling Fellowship, and Muscular Dystrophy Association (MDA) Care Center at the University of Colorado’s Anschutz Medical Campus. He completed his neuromuscular fellowship at the Ohio State University. Dr. Wicklund sub-specializes in neuromuscular medicine, performs electrodiagnostic testing, and interprets muscle and nerve biopsies. He has a particular interest in the diagnosis and treatment of genetic and autoimmune muscle diseases, including FSHD and the limb girdle muscular dystrophies. Dr. Wicklund has vast experience providing multidisciplinary care to patients and is actively involved in clinical trials on the CU Anschutz campus.



**Kaitlin Smith, MS, CGC** is a certified genetic counselor for the University of Colorado’s Department of Neurology on the Anschutz Medical Campus. Kaitlin is a 2016 graduate of the University of California, Davis where she earned a Bachelor of Science in Neurobiology, Physiology, and Behavior, and Bachelor of Arts in Psychology with a developmental emphasis. She graduated in 2020 from Arcadia University with a Master of Science in Genetic Counseling where she was awarded the Marie Barr Genetic Counseling Award in 2019 and completed her thesis: ‘Not alone anymore: parent and sibling experiences finding support with an ultra-rare diagnosis’. Kaitlin was selected as the inaugural Rare Disease and Neuromuscular Genetic Counseling Fellow at the University of Colorado in 2020 where she received additional training and education in neurogenetics and neuromuscular medicine at the University of Colorado Hospital and Children’s Hospital Colorado. She currently supports CU’s multidisciplinary Muscular Dystrophy Association (MDA) and adult neurogenetics clinics, coordinates genetic testing, and provides genetic counseling to patients and their families.



**Mary Colasanto, PhD**, PerkinElmer Genomics. Mary is the Genomics Testing Consultant for the Great Lakes region for PerkinElmer Genomics. I earned my Doctorate in Human Genetics from University of Utah for making scientific contributions to the underlying etiology of several neonatal-onset genetic disorders and bringing to light previously unknown phenotypes. I then transitioned to genetic engineering where I developed animal models of human disease for both industry and academic researchers. As my motivation for graduate school was to aid providers in ending the diagnostic odyssey, transitioning to clinical genomics has allowed me to educate and assist clinicians in finding answers for their most challenging cases.



**Hannah Dwight, DPT**. Hannah is a graduate of Arizona State University with her BS in Kinesiology, and Doctorate of Physical Therapy from Regis University. She currently works as the research physical therapist for CU's Department of Neurology in which she is able to both coordinate and execute research studies and work with individual patients in the clinic setting. Hannah has a passion for neuromuscular disorders and in helping educate and improve quality of life for patients with a wide variety of diagnoses. In her free time, she enjoys running, skiing, hiking, and trying new recipes.



**Karen Hookstadt, OTR/L**, is a board certified senior occupational therapist at the University of Colorado Hospital. Karen graduated from the University of Illinois of Chicago and has extensive and in-depth experience in helping individuals improve their ability to function in daily living, school, and work environments. Before joining UCHHealth, she worked in a number of inpatient, outpatient, and home health settings treating a wide variety of neurological, post-surgical, and trauma diagnoses. Karen has developed and managed Adaptive Driving, Therapeutic Recreation, and Rehabilitation programs and currently supports CU's multidisciplinary Muscular Dystrophy Association (MDA) and Amyotrophic Lateral Sclerosis (ALS) clinics as the primary occupational therapist, assisting with treatment plan development and evaluations for adaptive equipment.



**Kristin Orlowski, PhD**. Dr. Orlowski is a Licensed Psychologist who specializes in health psychology within an integrated primary care setting. Dr. Orlowski believes in the whole health model of care, working collaboratively with patients and the integrated care team in order to identify meaningful and tangible goals while utilizing evidence-based practices that are tailored to the specific needs of patients. Dr. Orlowski provides therapeutic support to improve overall health and behavioral health functioning. With a compassionate and reassuring presence, Dr. Orlowski strives to help each patient feel validated, empowered and motivated in their abilities to live their best lives. When she is not engaging in professional endeavors, Dr. Orlowski finds pleasure spending time with her husband and two daughters. She creates balance through additional hobbies including maintaining an active lifestyle, enjoying the beautiful outdoors and traveling.



**Suja Jagannathan, PhD**, University of Colorado. Suja Jagannathan is an Assistant Professor at the Biochemistry and Molecular Genetics Department and the RNA Biosciences Initiative at the University of Colorado Anschutz Medical Campus. Her group studies how cells detect and degrade aberrant messenger RNAs (mRNAs), and how dysregulation of this surveillance process contributes to FSHD. Before starting her own lab at the University of Colorado in 2018, Suja was a postdoctoral fellow at the Fred Hutchinson Cancer Research Center in Seattle (2013 – 2017) and a graduate student at Duke University in Durham, NC (2006 – 2013).



**Brianna Blume**, University of Colorado. Brianna Blume is a Research Program Manager in the Neuromuscular Division of the Department of Neurology at the University of Colorado Anschutz Medical Campus. Her role is to expand the Neuromuscular Clinical Research Program to help patients with rare diseases in the Rocky Mountain Region gain access to cutting-edge therapies. Before joining the Department of Neurology, Brianna completed a master's degree at Colorado State University in Biomedical Sciences with a concentration in Neurobiology (2011-2012).



**Elizabeth 'Lisa' Ackermann, PhD**, Neuromuscular Therapeutic Area Head and VP Clinical Development, Avidity Biosciences. Lisa joined Avidity in January 2020 and serves as the Neuromuscular Therapeutic Area Head and VP Clinical Development. Lisa brings extensive experience in both the research and development of RNA-targeted therapeutics and their application to rare diseases. She has held leadership positions in industry and patient advocacy groups including Ionis Pharmaceuticals, Otonomy, Myotonic Dystrophy Foundation, and TorreyPines Therapeutics. Over her 8-year tenure at Ionis, she led the development of Tegsedi® (an approved treatment for TTR Amyloidosis with Polyneuropathy) and several other rare disease programs. Lisa holds a Ph.D in biochemistry from UCSD and completed a NIH postdoctoral fellowship in Pharmacology at UCSD.



**Molly White**, Dyne Therapeutics. Molly brings extensive experience in patient advocacy and community engagement to her role at Dyne. She previously served as chief executive officer of the Myotonic Dystrophy Foundation, an organization dedicated to advancing research and enhancing the quality of life of people living with myotonic dystrophy. In that role, she led strategic direction and administrative oversight for the comprehensive programmatic and fundraising efforts of the foundation and its staff.



**Jenn McNary**, Fulcrum Therapeutics. Jenn is a trusted voice in the rare disease community as a mother, public speaker and fierce advocate. Her work in the rare disease space as a thought leader earned her the Ryan's Quest Ryan's Hero award in 2013, a nomination for the Global Genes Champion of Hope award in 2014, and the prestigious 2017 Meyer-Whalley instrument of change award. Jenn has unique experience in the drug development field, as a parent of children enrolled in the clinical trials, an advocate engaging with the regulators and as a consultant helping to develop programing for patients. Currently, Jenn is consulting in the



biotechnology space with an expertise in caregiver/patient engagement, including bringing the patient voice to drug development and solving barriers to access. She is also the Founder of One Rare, a non-profit formed to meet the needs of young adults with rare and chronic conditions.



**Jamshid Arjomand, PhD**, chief science officer, FSHD Society. 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 community development officer, FSHD Society. 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.



**Amanda Hill**, Colorado chapter director, FSHD Society. Amanda grew up in Colorado and lives in Highlands Ranch with her husband, Justin, who was diagnosed with FSHD in 2016 at the age of 31. Amanda works in biomedical research and is passionate about health and scientific education and advocacy, so becoming involved with the FSHD Society was a natural and obvious step for her. Amanda has served as chapter director for Colorado for several years and has also contributed many wonderful articles about FSHD research for the Society's website and the FSHD Advocate magazine.



**June Kinoshita**, director of research and patient engagement, FSHD Society. 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.