

FSH Watch



CONNECTING THE COMMUNITY OF PATIENTS, FAMILIES, CLINICIANS, AND INVESTIGATORS



ADVOCACY

FSH muscular dystrophy receives ICD-10 codes

Critical diagnostic classification standard raises visibility of FSHD

by DANIEL PAUL PEREZ
FSH Society

The average civilian has never heard of the ICD codes, so you can be forgiven for greeting this news with a shrug, but believe me, this is a huge deal. Read on.

In December 2017, the FSH Society, Parent Project Muscular Dystrophy (PPMD), and the Foundation to Eradicate Duchenne (FED) announced that the nominations to create more specific ICD-10 codes for Duchenne/Becker muscular dystrophy (Duchenne/Becker) and FSH muscular dystrophy (FSHD) had been accepted by the International Classification of Diseases (ICD-10) Coordination and Maintenance Committee. The new codes will be included in the Centers for Medicare and Medicaid Services (CMS) Fiscal Year 2019 Coding Addenda, effective October 1, 2018.

Until now, Duchenne/Becker and FSHD have been lumped into a broad category of diagnoses in the standard International Classification of Diseases (ICD). The ICD is the foundation for the identification of health trends and statistics, and the international standard for reporting diseases and health conditions. Owned, developed, and published by the

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GOVERNO DO ESTADO DE SÃO PAULO, CC BY 2.0, curid=66581281

The DUX genes turn other genes on and off, rather in the way a conductor like maestro Marin Alsop commands an orchestra. Photo credit: Governo do Estado de São Paulo, CC BY 2.0, curid=66581281



RESEARCH

DUX may be a master switch of the genome

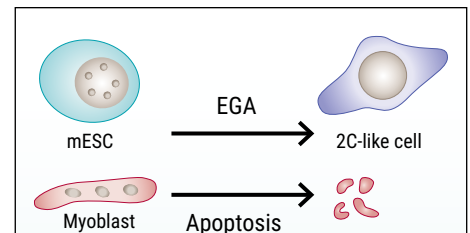
Recent papers thrust FSHD gene into the spotlight

by JUNE KINOSHITA
FSH Society

The DUX4 gene in FSH muscular dystrophy is typically described as a rogue actor, a genetic oddball that is never supposed to be active in adult muscle, and is rendered harmless by an elaborate lockdown system. It is only when several parts of the “lock” mechanism break that DUX4 emerges to cause damage to muscles.

Yet researchers have also known that

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Dr. Jekyll and Mr. Hyde roles for DUX genes? In a mouse embryonic stem cell (mESC), DUX sets off embryonic genome activation (EGA) in the two-cell stage (2C-like cell). But if expressed in an immature muscle cell (myoblast), DUX triggers cell death (apoptosis).

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Charting a bold course

When John F. Kennedy made his historic declaration that we were going to the moon, he cast a vision of a better future. A future where those things that were thought to be impossible, unachievable, and out of reach become possible, achieved, and grasped. In that statement, he began to rally the resources—innovation, intelligence, willpower, commitment—that would chart a bold course to achieve this vision.



Mark A. Stone

To paraphrase a statement by John Schaar, “The future is not some place where we are going, but a place we are creating ... first in mind, next in will, then in activity.”

Because of the continued support of the FSHD community, we have come to a place in history where we can chart a bold course toward a better future. A future where the ravages of FSHD are mitigated and eliminated. A future where therapies for muscle regeneration allow our families to restore what was lost. A future that actually cures FSHD. We have drawn a “line in the sand,” setting the date of 2025 when we will have disease-modifying therapies available to our families. This is our future—a place we are creating, first in mind, next in will, then in activity.

Our best and brightest researchers have validated the possibility of it. And we are beginning to put plans in place to achieve it. While there will be more details in the coming months, I have outlined some of the elements of our bold course below:

- **We have agreed to launch a therapy accelerator initiative called FSHD Therapeutics**, which will be a series of initiatives aimed at filling gaps that currently prevent successful clinical trials and regulatory (FDA) approval and hinder therapies

from reaching our families. We will be seeking less expensive diagnostic tests and ways to measure effectiveness of a therapy in a trial.

- **Additionally, within FSHD Therapeutics we will be seeking to create a venture capital fund** to fast-track the most promising therapies through the development pipeline. Many of our leading researchers and small biotech companies have promising treatments and cures but are working in a cash-strapped environment. We want to be prepared to fill the funding gaps through strategic investments, if necessary, to avoid delay.
- **Over the next three years, we need to raise \$15 million for these initiatives**—above our regular annual giving. While the challenge seems as insurmountable as going to the moon must have seemed when JFK cast that bold vision, we know that together we can do it, and together we will do it.

The clock is ticking. The target date is 2025. Our organization’s leadership has asked and answered the questions: “If not us, then who? If not now, then when?” Without you, these challenges would seem overwhelming. But through your willingness to support, influence, and be involved, you join an army of activists who believe that together anything is possible—including a world free from the devastating impact of FSHD.

On behalf of the families we serve and stand alongside, thank you.

Working until a cure is found.

Mark Stone



President and CEO
FSH Society

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It is our editorial policy to report on developments regarding FSHD, but we do not endorse any of the drugs, procedures, treatments, or products discussed. We urge you to consult with your own physician about any medical interventions.

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The truth set me free

A call to action for June 20, 2018

by TAYLOR QUIGG

Boyertown, Pennsylvania

When you learn to count your blessings and not your problems, you will realize how beautiful your life truly is.

This post wasn't easy for me to write. In fact, it's been one I've been working on, thinking about for just about five years. June 20 is World FSHD Day, and on this day I want to spread awareness about a huge part of my life.

Five years ago, I began to notice symptoms, and three years ago I was formally diagnosed with facioscapulohumeral muscular dystrophy (FSHD). Friends who saw my pictures on social media probably had no idea that I have a disability. That's because for the past five years, I have tried to hide it. I was embarrassed. I didn't want to be labeled, defined, or controlled by a diagnosis.

By not talking about it or addressing it, by suffering alone and in silence, I thought I would make it go away. But it didn't. In fact, I ended up hurting myself more. This last year has changed me for the better, personally, physically, and spiritually. I slowly started to open up about my disability, and with every person I shared it with, I felt lighter. I felt free. Having people help me took away some of my pain. Using products to support my body took away pain that I had been living with for years. I didn't have to hide behind a wall that I had built. We should all be able to be different and accepted for who we are.

For most patients, FSHD is not a life-threatening disability. However, it does affect everything that I do. I had two choices: I could continue to hide it, suffer, and feel sorry for myself—or I could take control of my life. I wanted to show my daughters that no matter what life hands you, you can handle it! I wanted them to know that we are capable of so much, and that having a disability does not mean that you are unable to do things. It only means that you have to work a little harder and do things a little differently to reach the things you want most in this life.

I used to let my disability control me. But that ends today. Over the last year I



Taylor Quigg

have been deeply inspired by so many people and their stories. I realized that it's time to share my own. I'm sharing this because I want people to see past any physical weakness, a stumble or fall, to see the person. To see strength and determination. To remind everyone to be more compassionate and more readily lend a helping hand, because you never ever know what struggles people are facing behind closed doors. This jour-

This June 20, raise global awareness of FSH muscular dystrophy by sharing a selfie or photo—with a smile, orange-slice smile, whatever expresses YOU. Or share a video or caption sharing a story about what you are grateful for with hashtags #OrangeYouGlad #WorldFSHDDay.

ney that I am on has made me realize that I am meant to be here. I have a purpose, a story, and I am ready to take charge.

Raising awareness about FSHD and people living with disabilities is now my goal. Over the last year,

I have found peace, healing, and strength through this journey. No matter what you are going through in your personal life, you are not alone. Share your story—you never know who needs to hear it. You will make a difference in the lives of others.

One more thing: smile. Help me spread awareness of this rare disease by taking a selfie smiling. The "F" in FSHD is for "facio"; one of the symptoms of this disease is that your face muscles become weak and it affects your smile. Post your selfie, make it public, and use the hashtags #FSHD and #WorldFSHDDay. Help us raise awareness, and let's finally find a treatment and a cure! [FSH Watch](#)

Editor's note: Adapted with permission. This essay first appeared on Taylor Quigg's Facebook page on June 20, 2017.

Shower sandals

REDUCING THE RISK OF SLIPPING AND FALLING

Living with FSHD has caused great difficulty for me in keeping my balance. I'm sure you've heard this many times from others who have this condition.

I was about to have my shower converted into a walk-in because I was afraid of falling while showering.

But then I found Vertico shower and pool sandals. I purchased them from Amazon, but they are also sold in many other places. I found them to be very helpful for standing in the shower, as they reduce the risk of slipping on water or soap. You still need to be careful, but they do help a great deal with keeping your footing and balance. I hope you find this tip to be useful!

—ANTHONY MANNUZZA, Woodhaven, New York



Vertico Shower and Pool Sandals (Remember to use your [smile.amazon.com](#) account when purchasing online!)



Ask Beth

Become a sustaining member by making a monthly gift

by **BETH JOHNSTON**
FSH Society

Monthly giving is an easy way to support a cause that is near and dear to your heart by having a contribution sent to a charity automatically every month. This can be done using a credit card or, if your employer has a program, by a payroll deduction.



Beth Johnston

Jeff and I have found it easier to send a smaller amount every month to the FSH Society than to donate a larger lump sum at one time. Not only do we not have to remind ourselves to give at some point during the year, but it's also much easier on our budget!

Best of all, we know that our monthly



gift is providing the FSH Society with a reliable stream of support.

So, what are the benefits?

- It's easy! Regular donations can be made in affordable monthly payments.

- Giving options include preauthorized payroll deductions, recurring credit card transactions, or good old-fashioned checks.
- **Tip:** Your employer may have a matching gift program, which would double your donation! Find out by asking your HR department or checking online at <https://doublethedonation.com/fshsociety>.
- Save postage and time with automatic transactions.
- You can easily increase, decrease, pause, or stop your gift at any time.
- You will receive an annual tax receipt for your records.

To learn more, and to set up your monthly gift, visit our donation page at www.fshsociety.org/support-us/make-online-donation, and click the link to make a recurring monthly gift. **FSH Watch**

Jim Chin elected chair of FSH Society Board of Directors

PERSONAL PASSION AND BUSINESS ACUMEN DRIVE OUR NEW LEADER

During its meeting in February 2018, the FSH Society Board of Directors unanimously approved the executive committee's recommendation for Jim Chin to be our next Board chair. The vacancy was created by the retirement of William R. Lewis Sr., MD, from his position as the Society's Board chair, a role in which he served for the past quarter-century (see related story, page 5).



Jim Chin

"Jim brings a sharp business expertise that will be valuable to the Society as we refine our focus on catalyzing potential therapies through creative partnerships with the biotech industry," said Mark Stone, president and CEO of the FSH Society.

Jim has sat on the FSH Society's Board for more than 10 years, serving as the chair of both the development and investment committees. Recently, he also served as chair of the search committee assigned to facilitate staff transition and hiring of the FSH Society's new president and CEO. Jim is passionate about the Society's mission to accelerate research toward treatments

and a cure, as his son Jimmy and his wife Barbara, both now deceased, suffered from FSHD.

"I am committed to our goal of having a disease-modifying drug to our families by 2025," said Chin. "Our chair emeritus Bill Lewis will be an inspiration in working with our committed Board and staff as we move forward under the leadership of our president and CEO Mark Stone."

Until his retirement in 2014, Jim worked at UBS Financial Services in White Plains, New York, where he was managing partner of the Chin-Meador Team and senior vice president. Prior to his role as a financial consultant, Jim focused his 17-year career in broadcasting sales, predominantly with the CBS television stations, before he switched to finance in 1987.

Jim graduated from the Baruch School of the City College of New York in 1970 with a BBA in economics and marketing. A native New Yorker, Jim now resides in Jenks, Oklahoma. He is a former financial lecturer, volunteer firefighter, baseball coach, umpire, and regional director for the Fresh Air Fund. He is a doting grandparent, and when he is not spending time with his family, he is busy volunteering for the FSH Society, following the stock market, and playing golf.

William R. Lewis Sr., MD, retires as chair

The “lowly neurosurgeon” who guided the Society for 25 years

by DANIEL PAUL PEREZ

FSH Society, CSO and Cofounder

Bill Lewis Sr. and I have worked side by side for nearly 25 years in our endeavor to understand, with clarity and accuracy, the mechanism of FSH muscular dystrophy. As Bill steps down from his long tenure as chair of the FSH Society Board of Directors, I have a few thoughts I would like to share with you about Dr. Lewis and give you a sense of the quiet giant he is.

In many high-level interactions, Bill would often say in his soft, raspy voice, “Well, I’m just a lowly neurosurgeon,” and go on to offer gem after gem of insight with a clean and pragmatic sense of thinking. Bill has a self-deprecating style, and such a simple, direct presentation that it often took me a while to grasp the true impact of his statements. He is simply a lovely man—kind, compassionate, caring, and deeply aware of others’ feelings and the power of simply listening to what they have to say. He is a consummate human being.

As chair of the Board and member of the Scientific Advisory Board, Dr. Lewis has always been supportive of our mission. He has been present at nearly every Society event that has marked our organization’s high and low points—a quiet, comforting presence we will miss. His stability and emotional constitution are rock solid, and we have all come to rely on his strength. He is loyal to the people and principles he values.

Bill is comfortable with himself and the choices he makes. His style of speaking is thoughtful and deliberate. In our time together over the past 25 years, I have never heard him raise his voice, swear, or express hate toward anyone or anything. He has always been alert, present in the moment, and happy to live and to let live.

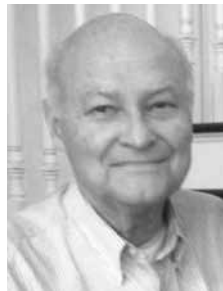
Bill smiles and laughs easily and often. I can hear his chuckle right now. He listens actively and carefully, and seldom offers advice even when it is sought. He lives by actions, by doing, and by setting an example. When you meet Bill, the first thing you notice is his tallness and physical constitution. He is a strapping fellow with intelligent blue eyes and a firm handshake. You look forward to seeing him and spending time with him.

Bill engages people easily. When you speak with him, you know he has heard and considered what you have to say. He makes the world a better place through work, education, and the practice of medicine—always with a strong moral and religious compass.

Bill understands the learning process, how to gain wisdom from experience, and how to apply that wisdom in practice. Having been there when the National Institutes of Health was founded, he comprehends the organization’s history and perspective. When we think of “lions” in medicine, Dr. Lewis truly fits the bill.

Although Bill’s retirement breaks one of the last major connections to the days of the FSH Society’s founding Board, in the near and distant future I will recall the kindness and wisdom of my colleague and dear friend William R. Lewis Sr., and draw on the strength those recollections bring me.

Please join me in thanking Dr. Lewis for his dedicated work. Our journey through FSHD is at a crossroads, and I believe the work we have accomplished together will allow us to move faster and further to accelerate therapeutics for FSHD. Bill, all of us at the FSH Society wish you and your family well, and sincerely thank you for all that you have helped us to achieve. FSH Watch



William R. Lewis Sr., MD

Honoring Carol S. Birnbaum, MD

FOR COMPASSIONATE SERVICE AND FSHD SUPPORT

In February of this year, Carol S. Birnbaum, MD, retired from the Board of Directors of the FSH Society. Carol joined the Society in 2012 shortly after the death of her mother at the age of 73 from complications related to FSHD.



Carol S. Birnbaum, MD

I first met Carol on Pearl Harbor Day in 2011 for lunch at a Jewish deli within a stone’s throw of her busy psychiatric practice in Cambridge, Massachusetts, where she works with people struggling with chronic diseases, as well as those trying to plan families in the face of genetic illnesses.

Carol’s service to the Society has always honored the memory of her mother and the wishes of her father to help others. With two sisters as well as a niece and nephew at risk for having FSHD, she has always kept a keen eye on the patient’s voice within the Society and the struggle to take meaning from an otherwise challenging and difficult situation.

Carol received her MD from the University of Connecticut School of Medicine, and did her residency in psychiatry at the Massachusetts General Hospital. Her insights and sharp mind have been incredible assets to the Society and our members. She served as chair of the nominating committee and has stewarded major changes in the Board, committees, governance, and communications within the Society.

The Board has greatly benefited from Carol’s combination of life and professional experiences. We hope that her work with the Society has helped with defining her own space around FSHD, and we are thrilled to have her brother-in-law Neil Solomon, MD, continue as a member of the FSH Society Board of Directors, where he serves on the science, technology, and research committee.

Carol, thank you for your compassionate service.

—DAN PAUL PEREZ
CSO and Cofounder, FSH Society



So damn lucky

I've found my calling, and it's one far beyond golf

by **MORGAN HOFFMANN**
West Palm Beach, Florida

In November 2016, I was diagnosed with muscular dystrophy, an incurable disease. The last few months have been the most trying of my life. I have a new reality now, and a new purpose.

In 2011, I started to notice that my right pec was deteriorating. I could see it in pictures and in my reflection in the mirror. So I started to seek the help of specialists all over the country, and even several in Canada. That was the beginning of a five-year period of misdiagnoses, frustration, and confusion. I visited more than 25 doctors. While they were “racking their brains,” my weakness progressed, my swing speed decreased, and I continued to lose muscle in my chest. Today my entire right pec is almost gone.

About a year-and-a-half ago, I visited a neurologist in New York City who decided to conduct blood tests. He said that, because he was looking for a specific analysis, the results wouldn't come back for three months. Six months later, I still hadn't heard anything. I called my trainer, Don Saladino, and we decided that reaching out to the doctor was the best idea. I hung up with Don and started searching for his number.

But then, seconds after Don and I had said goodbye, my phone started to vibrate. A New York City area code flashed on the screen.

It was the doctor.

“Morgan, the results came back,” he said. “You have muscular dystrophy.”

I was shocked.

“Well ... what does that mean?” I asked. “Am I going to be okay?”

“I'm not sure; I'm just telling you the results.”

The rest of the call was pretty much a blur. But before we hung up, the doctor told me one last thing.

“There's no cure.”

Before I tell you what this will mean for me going forward, I have to tell you where I'm coming from—for myself as much as anything. To be honest, my diagnosis still hasn't really sunk in yet. Maybe this is just a bad dream. Maybe my pec muscles will just grow back. Those were some of my first thoughts.

I know that I must fully accept this challenge, but doing that is so hard. The reasons why I know I'll be able to persevere—to conquer the future that's in front of me—are my childhood, my best friends, my family, my mentors, and my dreams.

Even though my life changed forever that November, that isn't when my story begins. It starts in the mid-1990s, in my hometown of Wyckoff, New Jersey.

My sister, Heather, and I grew up in a quaint house with a stern but understanding father and a caring, hard-working mother. I often think about our household and feel a sense of nostalgia for my childhood. Our lives were good, and I've always had an overwhelming sense of gratitude for the life I was born into.

When I reflect on it, everything was the way it should have been. Going to hockey practice at 5 a.m. on the weekends, baseball practice after school, and golf every other minute of the day. I can't



“Today I'm emerging from behind the scenes. Not because I want to talk about myself necessarily, but because I hope that by doing so I will be able to help find a cure for the disease that I and thousands of others are afflicted with.”

— MORGAN HOFFMANN

help but smile when I think about pretending to be Scott Stevens ripping slap shots from the blue line. Or Mariano Rivera striking out a big, mean-looking hitter to win the game. Or Tiger Woods hitting stingers into the net at the gym during golf practice. I made everything I did into a competition, into a game within a game.

When I think back to my childhood—to my “not a care in the world” days—that's what I remember. It was fun.

At night I'd lie awake dreaming about being a professional athlete—I just wasn't sure what sport I wanted to play. I ended up playing hockey for seven years and baseball for six, but I found my niche, my love, and my biggest challenge in golf. I loved everything about the game—the mental battles, the challenge of achieving

success on my own. I wanted to be just like Tiger Woods, but to have any chance of resembling him, I knew I had to fully commit to the sport. From my junior year of high school on, golf was the only game for me. It became my whole life.

I spent my junior and senior years of high school at the International Junior Golf Academy in Bluffton, South Carolina. I had already won two New Jersey state championships, and going to the academy gave me a chance to compete against the country's top talent. The warmer climate there meant that I was able to play year round, and combined with daily instruction from my coach Gary Gilchrist, the situation was ideal. Although leaving my sister, parents, and friends behind was one of the most difficult decisions I've ever had to make, in my mind, moving to the South was a path to the PGA Tour.

My time in South Carolina propelled me to Oklahoma State on a full golf scholarship. There, I played on a team with Rickie Fowler, Kevin Tway, and Peter Uihlein. You might think that my best memories of playing for the Cowboys are of being an All-America my freshman year and becoming the No. 1-ranked amateur in the world. And yes, it's true—those are great memories. But what really stands out is when I realized that I had the opportunity to give back to people who were less fortunate than myself. I have all my teammates to thank for that (we constantly reminded ourselves how lucky we were to be playing golf in college), especially Sean Einhaus.

Sean's mother is from Kathmandu. From Sean I learned of the hardships that many children in Nepal have to endure. While I was getting a free education and the chance to play golf, families halfway around the world were living on less than one dollar per day.

I was flabbergasted.

So Sean and I set up a tournament at Oklahoma State in which we played as many holes as possible in one day. Generous friends and family donated money for each hole we played. We finished 108 holes that day, and luckily we were able to raise enough money to send a number of Apple computers to a school that Sean's family had built in Nepal. A couple of years later I finally made my way to Nepal and visited the school; it was one of the happiest moments of my life. Seeing the smiles on the children's faces and experiencing how happy they were was astounding. I wish we had more loving, happy energy like that in the States.

I had interacted with a few local charities in Oklahoma, as well as back home in New Jersey, but this was the first initiative that I had started on my own. I was enthralled with the idea of giving back. I owe that to Sean. He taught me that there was more to life than golf and my own goals.

I worked my way onto the Web.com Tour in my first year out of school. That was when I embarked on my quest to engage with both private companies and nonprofit charities to promote health and wellness for kids in need. In my five years on the PGA Tour I've participated in many youth-development clinics around the country,

including for The First Tee foundation and the New Jersey Golf Foundation. I also partnered with St. Barnabas Health System and Blue Cross Blue Shield of New Jersey.

Through those collaborations, I've had the opportunity to visit children in hospitals, where I have met some of the most inspiring, kind, and motivated young minds. I was even extended the honor to be the starter at the Westfield 5K run presented by Blue Cross Blue Shield of New Jersey.

The children I've met who have diseases, terminal illnesses, or disabilities are truly incredible. They've taught me so much—how to fight, how to carry yourself, how to treat others when you are faced with a challenge that is beyond your control. Every one of them has one thing in common: happiness. They are why I want to give back. Even with all the stresses and hardships they experience every day, they are still able to find an uplifting smile and a contentedness with life. Fighting and never giving up is in my blood. No matter what happens to me, I will never stop doing everything in my power to make the lives of those around me better and to make the future healthier and brighter.

Since I earned my PGA Tour card in 2013, I haven't really spoken publicly about my involvement in these organizations, but it's truly been a passion of mine. I like to keep my personal life private—I don't have as many Twitter or Instagram followers as some other players on the Tour, and I definitely don't post all the time on social media about what I happen to be doing at any given moment.

But today I'm emerging from behind the scenes. Not because I want to talk about myself necessarily, but because I hope that by doing so I will be able to help find a cure for the disease that I and thousands of others are afflicted with.

The first thing I want to do is bring attention to our way of fighting. By our I mean the team I am lucky enough to be surrounded by. Don Saladino, one of my best friends, is both my health-and-wellness coach and my mentor, and has been the driving force behind my new way of life. He trains me to be an athlete, and we treat my body as if it were a machine. Everything that goes into it must have a purpose, must provide a benefit. My diet consists of all organic food, lots of water and vegetables, good carbs, and protein, and no dairy, gluten, or soda. My belief is that if you feed your body right it will run clean.

I've researched so many cases of people with diseases who made extreme overhauls of their diets. Often the changes they make halt and sometimes even reverse their symptoms. I know that there is no guarantee that anything so dramatic will happen because I am eating better, but I want to give myself the best possible chance of beating MD. And to do that, I've completely changed my way of living.

In my case, my muscular dystrophy is currently causing my right and left pecs to atrophy. Where the disease will attack next, I'm not sure. The characteristics of this specific type of MD (facioscapu-

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Announcing the Walk & Roll to Cure FSHD

A Signature Nationwide Fundraising Event

by **BETH JOHNSTON**
FSH Society

We are excited to announce the launch of our nationally branded Walk & Roll to Cure FSHD signature fundraising events in several locations around the country!

FSH Society Walk & Roll fundraising events have been held in the past (in San Diego, Cape Cod, and Colorado), but this is the first time we have coordinated the branding, messaging, and execution to be consistent and streamlined. Our brave Walk & Roll “pioneer” leaders have agreed to attend monthly training sessions and host a Walk & Roll in their area in order to beta-test all of the fundraising materials, the software platform, instructions, and procedures. With their help and leadership, we will be able to confidently launch our signature Walk & Roll fundraiser as a standard “toolkit” for people who would like to host a similar event in their area. We will launch this concept as part of our overall FSH Society chapter development program at the FSHD Connect Conference in Las Vegas this June. FSH Watch

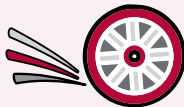
Editor’s note: Beth Johnston, our chief community development officer, is heading up the effort to create local FSH Society chapters around the country. If you would like more information about this new program and how you can participate, please contact her at *Beth*. Johnston@fshsociety.org.



Mark your calendars!

Here are our pioneering Walk & Roll to Cure FSHD events this year:

- **September 8, 2018: Castle Rock, CO**—Katie Ruckert, pioneer leader (www.FSHSocietyColorado.org)
- **September 15, 2018: Dublin, OH**—Susan Aumiller, pioneer leader (www.FSHSocietyColumbus.org)
- **September 16, 2018: Our Bay Area team** will participate in an existing run/walk event in Alameda, California, to raise awareness and funds—Nez Bennouna, pioneer leader.
- **September 22, 2018: Puyallup, WA**—Nancy Payton, pioneer leader (www.FSHSocietyNorthwest.org)
- **Date TBD, Raleigh, NC**—Meredith Huml, pioneer leader



2018 IRC Workshop & FSHD Connect

DON'T MISS OUT! THERE'S STILL TIME TO REGISTER.

FLAMINGO LAS VEGAS, JUNE 8-10, 2018
REGISTER AT www.fshsociety.org.

IMPORTANT DEADLINES:

May 18, 2018—Flamingo hotel room block reservation
May 25, 2018—Registration deadline

The International Research Conference is June 8 through noon on June 9. This invitation-only workshop is the premier annual platform for FSHD researchers.

The FSHD Connect Conference begins at 1:00 p.m. on June 9 and continues through June 10. This day-and-a-half of immersive learning and community building is for patients and families, researchers, physicians, and health experts.

The CureFSHD banquet on the evening of Saturday, June 9, celebrates the FSHD community.

We thank our sponsors: Acceleron Pharma, AFM Téléthon, FSHD Canada Foundation, Fulcrum Therapeutics, Genea Biocells, Genomic Vision, Sanofi Genzyme, Ultragenyx, Univ. of Nevada Reno School of Medicine, Wellstone Center for FSHD Research.



FLAMINGO HOTEL RESERVATIONS: Visit <https://aws.passkey.com/go/SFFSH8>, or call the Reservation Center at 1 (888) 373-9855 to secure a reservation in our FSH Society Connect Conference room block (group code SFFSH8). Please be aware that a processing fee of \$15.00 + tax per reservation will be incurred if you choose not to use the dedicated weblink.

SO DAMN LUCKY

... from page 7

lohumeral muscular dystrophy) are atrophy of the chest, back, neck, arms, and sometimes legs. Each case is different, and some muscles degenerate more quickly than others. As of now, the disease has progressed slowly—only the right and a minimal amount of the left pec have deteriorated since I first started noticing an issue six years ago. Doctors are searching for a cure, conducting stem cell research, and experimenting with growth hormone treatments. I'm hopeful that they are on the right track.

As you might imagine, I often reminisce about the days when I used to play every sport in the book, when I was just a kid and didn't have a care in the world. Now, I try to live every day to the fullest. I have so many hobbies that I love: paddleboarding, surfing, fishing, boating, working out, watching my massive puppy grow, playing hockey, and flying my own plane. I also recently helped start a clothing company with one of my best friends, Charlie Schaefer. It's called Greyson. I love doing anything that's active, fast, and adrenaline packed. And the thought of possibly not being able to enjoy an active lifestyle is extremely scary.

But I believe now that this is why I was put on this Earth—so that when a child is diagnosed with muscular dystrophy, there will be a cure; there will be people to help with mental, nutritional, and physical training guidance. And especially so that no disease will ever hinder a little boy's or girl's passion for life.

Even though the type of muscular dystrophy that I have doesn't pose an immediate threat to my life, there is a good chance that it will shorten it. I don't know when that will happen, because there's no way to gauge the speed at which the disease will spread.

But please know this: This disease won't keep me from achieving my dream of winning on the PGA Tour—and it shouldn't keep others from chasing their dreams, either.

My girlfriend, Chelsea, has definitely been my biggest supporter throughout this entire ordeal. After I was diagnosed, she and I scrolled through pages and pages of Google search results for muscular dystrophy. We tried to gather as much information as we could about prognoses and new treatments. Most of the technological advancements we read about were actually pretty exciting. But we also ran across something that we both found incredibly sad.

We found countless blog posts in which people were describing symptoms almost identical to mine. They seemed to be, at their core, cries for help.

These people had no idea what was wrong with them.

Muscular dystrophy is very difficult to diagnose. And when the disease is properly identified, those who suffer then have to confront the financial burden of paying for treatment from the few specialists in the field.

It's a difficult outlook.

I am determined to help make a difference. I cannot wait to start raising money and awareness to fight this disease! Soon, I will be announcing the date of a charity golf event that I will hold at my home course, the Arcola Country Club in Paramus, New Jersey.

Speaking of the Arcola Country Club, it's kind of strange how things come full circle. Because lately, one memory in particular keeps coming back to me. It was in 2010, after I had just finished my junior year at Oklahoma State. I was walking down the ninth fairway on a perfect, 73-degree cloudless evening at the very club at which I will host my charity golf event. At that point, I knew I

was going to be turning pro, and I knew I had the rest of my life ahead of me.

As I looked up at the green, I caught the setting sun through the trees and could see it beaming on the clubhouse perched on its hill. You know how when you just see the most beautiful thing in the world, and even if your mind has a million things to say, you can't really find the words to explain the picture? It was like this sunset had pulled me out of my body, and I could see myself looking at it. I began to breathe deeply, filling my lungs with oxygen.

I thought about my childhood, running around Wyckoff without a care in the world. I thought about how lucky I was to have gone to both the premier high school and college for golf. I thought about the tournament that Sean and I had hosted for the kids in Nepal.

I thought about how my life was going just as I had always planned—and how I was about to become a professional golfer.

Most of all, I thought about how I had it all—my health; an amazing, loving family; incredible friends; and the chance to play a game I love.

And as the sun continued to set, the feelings of gratitude flooded in like rushing water. I gazed around and took a few more deep breaths.

I was so damn lucky to experience this life.

I was so damn lucky.

And today I know that I am so damn lucky. Because I've found my calling, and it's one far beyond golf. **FSH Watch**

Editor's note: Morgan Hoffmann is a professional golfer. Originally published on The Players' Tribune, December 4, 2017, this article is reposted with permission from <https://www.theplayerstribune.com/morgan-hoffmann-pga-tour/>.

Information at your fingertips

The FSH Society is dedicated to making sure you have accurate, useful information to help improve the quality of your healthcare and daily life. Our publications are created and reviewed by patients and experts. Visit our website (www.fshsociety.org), and go to Understanding FSHD/ Brochures & More to download:

- About FSHD (also in Spanish and Chinese)
- Physical Therapy and FSHD
- FSHD: A Guide for Schools
- FSHD and Social Support: A guide for friends and family
- Evidence-based FSHD care guideline—Summary for clinicians
- Evidence-based FSHD care guideline—Summary for patients & families

You can also request printed copies by mail at FSH Society, 450 Bedford Street, Lexington, MA 02420, or by phone at (781) 301-6060.





Have MG sports car, will travel

Crossing the continent fulfills a life's dream

by LEE and LIZ NINER

Hilton Head Island, South Carolina

When you retire, going through your bucket list becomes a high priority. In 2017, we decided it was time to cross off an item—a long-held dream to travel across America in our beloved 1970 MGA sports car.

We have been involved with MG sports cars since 1981. Liz was diagnosed with FSHD in 1996 when we were living in suburban Philadelphia, where we kept a “stable” of three MGs. Our passion for MGs has taken us to all parts of the US, Canada, and the UK. As Liz began declining physically in the early 2000s, she was determined not to let her affliction keep her from indulging her passion for road trips.

Liz maintained a very disciplined and dedicated exercise program of floor, machine, and water aerobics. She tailored her exercise program to focus on her core muscles while not overusing her leg and shoulder muscles. As FSHD began its gradual progress, Liz went from walking slowly to using a cane, and is currently using a walker. Simply getting into and out of the MG requires focus and determination, and this is where the exercise regimen really pays off.

The first leg of our trip was to drive from Hilton Head Island 600 miles up the Atlantic coast to Virginia Beach, where we joined six other MGA sports cars taking part in a cross-country drive called

from other drivers, and also when we park in a handicap spot. You would be amazed at how many casual bystanders offer assistance as she makes her way into hotels, restaurants, and gas stations.

Westward across Virginia, Tennessee, and Arkansas, Liz navigated the MG. We crossed the Mississippi River and proceeded west into Oklahoma. Liz brought her rubber stretching bands with her, and nightly exercises kept her muscles in condition. Crossing into Texas, New Mexico, and Arizona, where temperatures regularly soared above 100 degrees, we cruised on. Lots of time was spent on the fabled Route 66, and we even stayed in an authentic 1950s motel. Liz’s mobility issues presented challenges, but with Lee’s assistance she navigated the room’s period bathtub shower. Arriving in California to be greeted by the Pacific Ocean was an unforgettable and fitting conclusion to our 4,200-mile journey.

Crossing America, with its diverse scenery, on mostly secondary roads is a trip definitely worth taking. The lyrics of “This Land Is Your Land” kept coming back to us as we crossed farmlands, rivers, prairies, mountains, and deserts. Enjoying local cuisine and talking with people from small towns during gas stops and daily motel stays offered so much more than you could ever get from watching a movie or reading a book.



Liz and husband Lee departing on their “Ocean to Ocean” adventure

“Crossing America, with its diverse scenery, on mostly secondary roads is a trip definitely worth taking. The lyrics of “This Land Is Your Land” kept coming back to us as we crossed farmlands, rivers, prairies, mountains, and deserts.”

— LIZ NINER

the Ocean to Ocean caravan. The plan was to travel between 300 and 400 miles a day on secondary roads, leaving enough time to take in local sights.

For some yet unexplained reason, Liz is quite comfortable in the MG when all logic says this should not be. Her walker is placed on top of two soft pieces of luggage affixed to the luggage rack mounted on the MG’s trunk. This arrangement draws lots of quizzical looks

We really wanted to send a message that somebody with FSHD can be very capable, meet people, and experience the sights and sounds of a cross-country trip—in a 57-year-old sports car with no air conditioning and no windows!

Our respect for the kindness of perfect strangers grew immensely as we crossed the US. We all live in a wonderful country and need to experience it, no matter what our disabilities are. Life is good. [FSH Watch](#)



Knowing others who share your diagnosis makes you happier

Findings from major study of long-term disability

by JUNE KINOSHITA
FSH Society

We have long believed that helping individuals and families with FSHD meet one another is one of the most important services the FSH Society provides. It's why we invest energy and resources in our peer support network, local gatherings, social media groups, and the biennial FSHD Connect Conference. Now there is scientific evidence to support our



hunch. According to a large, seven-year study conducted by the University of Washington, individuals with chronic health conditions are happier when they know friends who share their medical diagnosis.

This intriguing news reached us via our former Board member Ann Biggs-Williams, who was a volunteer participant in the Aging and Quality of Life Study. Begun in 2009 and lasting through 2017, it is the largest longitudinal study of individuals living with long-term disability. Seven years of data were collected from 1,864 participants, including 1,218 who had participated for the entire length of the study, led by Michael P. Jensen, PhD, and his colleagues at the Rehabilitation Research and Training Center at the University of Washington.

According to a letter reporting the study's findings to its participants:

- Middle-aged individuals reported greater distress than younger and older individuals.
- Individuals who reported finding purpose or meaning in their lives reported less distress.
- Individuals who showed greater resilience reported a higher quality of life and were more satisfied with their ability to participate in social relationships.
- Greater fear of falling correlated with lower rates of physical activity.
- Those who had friends who shared their medical diagnosis reported being happier; conversely, those who did not list any friends sharing their medical diagnosis reported having more severe physical limitations and lower quality of life.

So, if you have felt hesitant about whether to reach out to others in the FSHD community, we say yes, do it—it's good for you! There are many ways to make connections:

- Attend a get-together near you (see our Events Calendar).
- Join one of our private Facebook communities.
- Come to 2018 FSHD Connect.
- Or simply contact the Society, and we can connect you with a peer.

The Rehabilitation Research and Training Center has developed a series of evidence-based fact sheets for individuals and clinicians. They can be downloaded at <http://agerrtc.washington.edu/info/factsheets>. FSH Watch



Acceleron plans to advance to Part 2 of its ACE-083 trial

Increases in muscle size were seen in Part 1 of the study

by JUNE KINOSHITA
FSH Society



Acceleron Pharma Inc., of Cambridge, Massachusetts, has announced plans to initiate Part 2 of the ACE-083 Phase 2 trial in patients with facioscapulo-humeral dystrophy (FSHD) during the second quarter of 2018. The news follows positive preliminary results released this January for the first two cohorts in Part 1 of the Phase 2 clinical trial with ACE-083 in FSHD patients. There are currently no FDA-approved treatments for FSHD.

"Preliminary results of ACE-083 in FSHD patients demonstrated positive safety and tolerability, along with unprecedented mean increases in total muscle volume of more than 12 percent in the two distinct muscles evaluated," said Matthew Sherman, MD, chief medical officer of Acceleron. "These data support our decision to advance to Part 2 of the Phase 2 trial, which we expect to initiate in the second quarter of this year. We look forward to fully exploring functional outcomes in the larger, placebo-controlled Part 2 of the trial."

Part 1 of the Phase 2 trial was an open-label study of ACE-083 designed to evaluate safety as well as changes in total muscle volume in patients with FSHD. Magnetic resonance imaging (MRI) was used to evaluate 11 patients with tibialis anterior weakness and 12 patients with biceps brachii weakness (with greater weakness on one side of the body). Each patient received ACE-083 (150 mg or 200 mg) as an injection into the weaker muscle once every three weeks for 12 weeks. Three weeks after the last injection of ACE-083, the muscle volume was measured by MRI and compared to its volume at the beginning of the trial.

"ACE-083 is demonstrating encouraging activity in its ability to increase muscle volume, and I look forward to its advancement in Part 2 of the trial," said Jeffrey Statland, MD, associate professor of neurology at the University of Kansas Medical Center, and the ACE-083 FSHD Phase 2 trial principal investigator.

Effects of ACE-083 on strength and function versus a placebo control will be evaluated in Part 2 of the study. If you are interested in enrolling in this trial, please visit clinicaltrials.gov and search "ACE-083" for updates and contact information for the sites that are recruiting volunteers. FSH Watch



FSH Society awards \$690,894 in new research grants

Advancing therapeutics and biomarkers

by DANIEL PAUL PEREZ and JUNE KINOSHITA
FSH Society

A drug that helps muscles grow and regenerate. Molecules that occur naturally in our cells to repress DUX4, the “toxic gene” thought to cause FSHD. A deeper dive into why FSHD muscle may have trouble regenerating. These are some of the exciting ideas that the FSH Society has funded in its latest round of grant awards.

For the August 31, 2017, round of grant submissions, we received 11 applications (nine new, one resubmission) and one request for a one-year extension for ongoing research projects. Six were awarded funds totaling US\$690,894. Here are the funded projects:

► NATURAL MICRORNAS AS POTENTIAL MODIFIERS OF DUX4 TOXICITY

Nizar Saad, PhD, The Research Institute at Nationwide Children’s Hospital, Columbus, Ohio, USA

FSH Society Sylvia & Leonard Marx Foundation fellowship, US\$80,000 for one year



Nizar Saad, PhD

Facioscapulohumeral dystrophy (FSHD) is a complicated disorder. After many decades of study, the FSHD research field now has focused on mis-expression of DUX4 as a primary factor in the disease. Therefore, Dr. Saad and colleagues are focusing on developing therapeutic strategies targeting DUX4. In order to reach this goal, they are testing a molecular screen that will allow them to identify natural molecules produced normally in all human cells that may operate to reduce DUX4. These molecules are called “microRNAs” and are known to turn off specific genes by activating natural cellular gene silencing pathways. As a next step, they will look for drugs that are capable of boosting the presence of these microRNAs, thereby reducing the presence of DUX4 in skeletal muscles.

► HYPERMORPHIC SMCHD1 VARIANTS

Jessica C. de Greef, PhD, Leiden University Medical Center, the Netherlands

FSH Society Sylvia & Leonard Marx Foundation fellowship, US\$171,000 for three years (US\$57,000 per year)



Jessica C. de Greef, PhD

DUX4 suppression is a promising therapy for individuals with facioscapulohumeral muscular dystrophy. Dr. de Greef’s group has recently identified several genetic variants in the chromatin protein SMCHD1 that potentially increase the activity of SMCHD1 at the D4Z4 repeat array. Thus, naturally occurring SMCHD1 variants may exist that protect muscle from DUX4 expression. In this project they will determine if selected SMCHD1 variants increase DUX4 repression in several

disease models. Dr. de Greef expects this project to contribute to our understanding of the FSHD disease mechanism and possibly lead to the development of a functional test to screen additional genetic variants in SMCHD1. Importantly, this line of research may also offer us novel options for DUX4 suppression by modulating SMCHD1 activity at the D4Z4 repeat array.

► STRYKA-001 TREATMENT IN THE FSHD-LIKE MOUSE MODEL

Ryan Wuebbles, PhD, and Takako Jones, PhD, University of Nevada, Reno School of Medicine, USA

US\$190,000 for one year

The laboratories of Dean Burkin and Peter Jones at the University of Nevada, Reno School of Medicine are collaborating on an exciting project funded by the FSH Society to explore the therapeutic use of a new small molecule, Stryka-001, for FSHD. This will be one of the first projects to explore a therapeutic treatment in the novel FSHD-like mouse model created by Dr. Jones’ lab. Stryka-001 is not expected to directly affect Dux4 expression, the direct cause of FSHD; however, it will instead promote muscle regeneration and recovery after damage. Preliminary results suggest that treatments using Stryka-001 result in reduced muscle loss and increased strength recovery after DUX4 expression has caused extensive muscle damage in this mouse model of FSHD. The hope is that Stryka-001 will offer an effective treatment option to mitigate further muscle damage and improve muscle strength while DUX4-modifying therapeutics are becoming viable treatment options. Stryka-001 can also be used in combination with other treatment options to speed muscle recovery for FSHD patients. Stryka-001 is already FDA approved for another indication, and therefore these studies may quickly translate into clinical trials within a year. If successful, FDA approval of Stryka-001 for use in FSHD patients could occur within three years.



Ryan Wuebbles, PhD

► **CHARACTERIZATION OF A NOVEL INHIBITOR OF DUX4 ACTIVITY**

Davide Gabellini, PhD, Fondazione Centro San Raffaele, Milan, Italy
US\$85,000 for one year



Davide Gabellini, PhD

A possible new inhibitor of FSHD has been found. Dr. Gabellini has discovered a new molecule able to block some of the processes that go awry in FSHD. The project, supported by the FSH Society, will characterize the exact mechanism of action of the new molecule to obtain a better understanding of how its properties could be used for therapeutic purposes. The project will also test the ability of the molecule to block disease-relevant symptoms using cellular and animal models of FSHD.

► **INTERPLAY BETWEEN MYOGENESIS AND THE IMMUNE SYSTEM IN FSHD PATHOLOGY**

Peter Steven Zammit, PhD, and Maryna Panamarova, PhD, King's College London, UK
US\$99,894 for one year



Peter Steven Zammit, PhD



Maryna Panamarova, PhD

This project examines the interplay between myogenesis and the immune system in FSHD pathology. When skeletal muscle is damaged, repair mechanisms are induced to heal the injury. Such repair mechanisms at the site of muscle injury include local inflammation, which contributes to controlling muscle stem cell proliferation and eventual differentiation to repair/replace damaged myofibers. These repair mechanisms are impaired in facioscapulohumeral muscular dystrophy, where damaged muscle becomes abnormally inflamed, often causing pain and discomfort. Inefficient muscle repair contributes to muscle weakness and wasting. However, our understanding about why muscle repair is compromised in FSHD is limited. Through analyzing which genes are active in muscle cells from FSHD patients and unaffected individuals, we are able to determine the molecular changes occurring in FSHD. Using RNA sequencing, Dr. Zammit and colleagues found that a major muscle-repair pathway in healthy muscle is specifically suppressed in FSHD. They will investigate this pathway further to better understand why it is repressed in FSHD, and also test a range of drugs to try to augment the activity of this pathway in FSHD muscle cells. We hope that these findings will give us

a better understanding of FSHD pathogenesis and provide a therapeutic strategy that could improve patients' quality of life and degree of disability.

► **BIOMARKER IDENTIFICATION BY HIGH-RESOLUTION PROTEOMIC APPROACH IN FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY**

Giorgio Tasca, MD PhD, Università Cattolica del Sacro Cuore, Rome, Italy
US\$65,000 for one year

The identification of sensitive molecular biomarkers of disease activity and progression is one of the major trend-topics in FSHD research. FSHD is indeed a relatively slowly progressing disease, and the ability to track subtle changes in the disease status is of major importance to assess the efficacy of a potential treatment.



Giorgio Tasca, MD PhD

One way of acquiring insights into molecular changes at the single muscle level is by studying muscle biopsies, which are, of course, invasive (and sometimes painful) procedures. Moving forward following the concept of the so-called "liquid biopsies," Dr. Tasca and colleagues developed a novel approach, i.e., long-term microdialysis with large pore membranes, which allows the continuous sampling of muscle interstitial fluid with a minimally invasive procedure consisting of the insertion of a tiny (1-mm diameter) catheter in the muscle through an intramuscular injection. The current project focuses on the characterization, by a high-throughput proteomic approach, of the interstitial fluid obtained from muscles showing radiological signs of early damage, identified by muscle magnetic resonance imaging (MRI). Muscle MRI is indeed able to detect the muscles that will more likely undergo faster degeneration. The analysis of these fluids (called microdialysates) could also provide new insights to understand disease mechanisms through the identification of biochemical pathways—possible targets for new therapies—that are dysregulated in FSHD muscles. Finally, the molecules that will be found in the microdialysates, locally produced at the site of muscle damage, have the potential to be also secreted into the bloodstream. Therefore, starting from the investigation of selected target molecules in muscle interstitial fluids and then moving to the analysis of blood samples could allow greater sensitivity in the detection of circulating markers of subclinical disease activity. FSH Watch



Bed yoga

Four easy poses to start or end your day

by KATHY SENECA
Cromwell, Connecticut

I became a yoga practitioner in the early 1990s, and over time I received certification and training in hatha and Svaroopa® yoga, taught classes, and ran workshops in those traditions for 10 years. I went on for advanced training as a yoga therapist, and after being diagnosed with FSHD, I began working with individuals with specific physical needs.

As traditional poses became difficult and harmful to my body, I found bed yoga beneficial and a way to maintain a practice. There is always a way to adapt and modify a pose, so yoga and yoga breathing can become part of a daily routine. These poses are designed to release the spine and provide the body/mind with the benefits of a yoga practice.

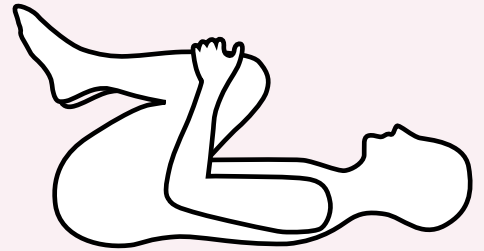
It's important to remember to breathe throughout the stretches. I recommend three-part breathing: inhaling—chest, belly, abdomen; and exhaling—abdomen, belly, chest, like riding a long, slow elevator. You can also reverse the direction: inhaling—abdomen, belly, chest; and exhaling—chest, belly, abdomen. Whatever feels comfortable to you.

One of the benefits of deep breathing is that it oxygenates the blood and helps calm the mind. If you get tired of this yoga breathing, take a break and simply do natural breathing, following your breath as it goes in and out. Yoga breathing is traditionally done through the nose, but again, if this is uncomfortable, do what works for you.

The recommended time to hold poses is at your discretion. Listen to your body, and do what feels good. Holding for 30 seconds brings benefits. Remember, pain or discomfort with a pose signals it's time to move out of the position and consider avoiding that position moving forward, depending on what your body is telling you. There are no hard and fast rules here, only guidelines, and, after all, you are in bed! Use any assists that work, from other people to pillows, straps, and anything in between. [FSH Watch](#)

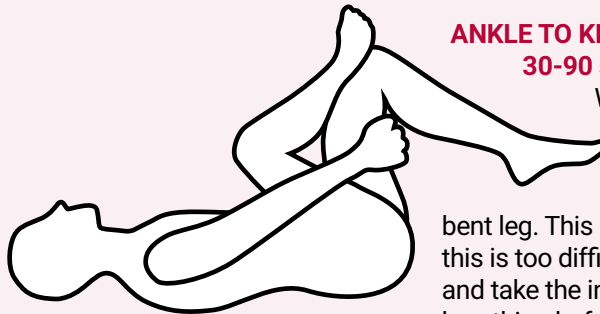
KNEES TO CHEST— hold pose one to two minutes.

Draw both knees to your chest using your arms. This lengthens the spine and helps relieve stiffness. You may hold your knees, behind the knees, or use a strap. A nice variation is to do each knee individually, then both knees together. This reaches deeper into the muscles and allows for greater release.



ANKLE TO KNEE—hold each side 30-90 seconds.

With one knee bent and that foot on the bed, cross your other leg so the ankle rests just above the knee of the bent leg. This pose helps open the hips. If this is too difficult or uncomfortable, skip it and take the interlude to do a minute of yoga breathing before proceeding to the last pose.



KNEE DOWN TWIST—hold each side 30-90 seconds.

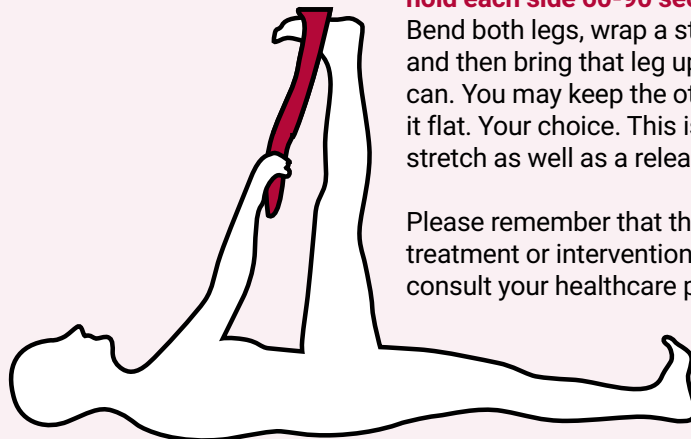
This pose is done on the right and left side. Try to be aware of creating a right angle at the knee with the lower leg and thigh (see diagram). If you find your legs are uncomfortable, stuff a pillow between them. This allows the body to relax and soften into this spinal release. When ready, bring knees to chest and shift slowly to the second side.



HAND TO BIG TOE— hold each side 60-90 seconds.

Bend both legs, wrap a strap around one foot, and then bring that leg up as straight as you can. You may keep the other leg bent or stretch it flat. Your choice. This is a great hamstring stretch as well as a release of the lower back.

Please remember that this is not medical treatment or intervention. If you have concerns, consult your healthcare provider.





Dear Neurologist

An open letter about newly diagnosed FSHD patients

by JIM ALBERT
Eldersburg, Maryland

As an FSHD patient diagnosed in the early 1990s, I'm writing this letter to provide some insight from a patient's viewpoint about working with newly diagnosed FSHD patients. It is my hope to provide an opportunity to strengthen the relationship among neurologists, FSHD patients, and FSHD research as a whole.

Like many FSHD patients, when I first developed weakness in my right shoulder, I made about a dozen visits to doctors trying to determine what was wrong. I visited a variety of specialists, from internists to orthopedists, but didn't find any of their answers to be convincing. I finally found one doctor who was both intelligent and humble enough to admit he had no idea what was wrong. He advised me to consult a neurologist, and this finally led to a diagnosis of FSHD. Genetic testing for FSHD was not available at that time, but the diagnosis made the most sense to me. An initial journey such as mine is quite common for FSHD patients.

Unfortunately, my diagnosing neurologist was not the most sensitive individual. He asked if I would come to his medical school class, and I agreed. Being young and somewhat naive, I was under the misimpression that I would be seeing more neurologists so that someone could actually "fix" me. Instead, I sat shirtless in front of about 30 students as the neurologist pointed out which muscles on me were strong and which were affected. I was a weight lifter, so the difference between atrophied and hypertrophied muscles was significant and very easily seen. I felt like a lab rat.

The experience was traumatic, and it really turned me off from seeing other neurologists. In fact, I never wanted to talk about the disease again because I hated what FSHD was taking away from me.

Neurologists should understand that the patient's experience at the time of diagnosis is essential for keeping people engaged with a disease that has no treatments and is both frightening and angering.

Based on my more recent visits to

clinics, I believe the FSHD patient's experience has improved significantly. In 1990, all I was told was that there was no treatment, nor was the disease well understood, other than that it was likely to be genetic. I was handed a piece of paper with a local support group's phone number that I never used.

Some young people diagnosed today with FSHD are too angry and afraid to see what the future might hold. They would rather live and adapt as best they can. It's only when adaptation becomes too difficult that some will decide to get involved.

Some will argue the path I took isn't

one of the struggles of FSHD advocacy groups is to get young, mildly affected people involved. Newly diagnosed patients have much to contribute because they are often still energetic and strong, and will remain so for a good number of decades. I feel it would be healthy for patients to strive for a balance, continuing with life while championing the cause that will someday help everyone who is stricken with this disease.

After my diagnosis, I did not visit a neurologist for the next two decades. I felt, "Why bother?" There was nothing that could be done, and my initial experience



“Neurologists should understand that the patient's experience at the time of diagnosis is essential for keeping people engaged with a disease that has no treatments and is both frightening and angering.”

— JIM ALBERT

necessarily a bad thing. However, having been on the sidelines for several decades, I now know that being involved with advocacy and research is far more rewarding than letting others do all the hard work.

With FSHD, as with any rare disease, patient involvement is vital and should be encouraged as much as possible. Currently,

had been so unpleasant. That just wasn't smart on my part. A support group would have kept me better informed and actively involved in finding answers. One wonderful day when that cure comes, it will be very rewarding to be able to say that I helped, or I was at least part of the solution.

... continued on page 17



Tissue modeling may cut clinical trial time

FSH Society biospecimen registry helps to speed progress

by ED MISETA
Clinical Leader, Chief Editor

Biotech company Fulcrum Therapeutics was founded in July 2016 with a focus on discovering small molecules to regulate genes. The company currently has three primary areas of interest: neurodevelopmental disorders, neuromuscular disorders, and neurodegenerative disorders. In the neuromuscular space, the company is using tissue donated from patients with facioscapulothoracic (FSH) muscular dystrophy, an incurable form of the disease, to find a treatment. The tissue is used to create research models that look and respond much like natural human tissues.

“Our approach involves identifying small molecules which can turn disease genes on or off, depending on the deficit associated with the disease,” says Owen Wallace, PhD, chief scientific officer for Fulcrum. “We are looking to either stop or turn on the expression of a particular gene. With FSH muscular dystrophy, we know the disease is caused by a protein called DUX4. Our hope is to ultimately discover and produce a pill that will reduce the production of DUX4.”

Examine patient samples

Discovering the mechanisms that will lead to a cure is a difficult process that involves focusing on the biology most relevant to the disease. The process also involves looking at patient-derived samples.



For muscle diseases, biological research is performed in myotubes, which are a developmental stage of a muscle fiber and the most representative samples that can be accessed from patients. Fulcrum is looking at myotubes from both healthy individuals and those who are afflicted with the disease. Researchers at Fulcrum are hoping to identify compounds and mechanisms that might correct the abnormal synthesis of DUX4. The *in vitro* cellular systems capture many mechanistic aspects of the disease and allow researchers to correct molecular deficits in ways that were not possible 10 to 15 years ago.

“There are two different approaches we are taking,” states Wallace. “First, we are dis-



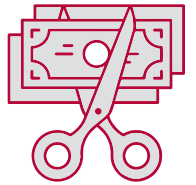
The Fulcrum Therapeutics team on World FSHD Day 2017.

covering what we believe to be the relevant mechanisms to correct the molecular deficits in patients. We believe the cellular systems I mentioned will be relevant to the disease. These are not highly engineered systems, but we believe the biology is quite close to the biology that is going on in patients. Second, we can derive cellular systems from a variety of different patients and explore how our molecules and mechanisms work across those cell lines. This will allow us to reproduce some of the heterogeneity that we would ultimately see when we get into a patient population, and ensure we are bringing forward the medicines that we think are going to have the greatest impact on the broadest patient population.”

Fulcrum will still need to perform *in vivo* experiments using animal models. But by using donated tissue samples from humans, models can be created that are similar to muscles in the human body. Performing tests on those models allows Fulcrum to develop its medicines to the point where there is increased confidence of the impact on humans. When the company brings forward a medicine for clinical testing, it will have the greatest opportunity to have the outcome Fulcrum is hoping to achieve in patients.

Reducing the time and cost of trials

Performing tests on tissue models prior to performing clinical trials on patients should reduce the time and cost of performing trials. But more importantly, Wallace notes the biology more closely reflects what is actually happening with the disease.



“Preclinical research done in overly engineered cell systems is often not very reflective of what is actually happening in a patient,” says Wallace. “For example, the signaling pathways being followed in these cellular systems can be quite different from the signaling pathways that might occur in a patient with a given disease. Clinical trials performed in many therapeutic areas are not successful and, in many cases, it is because the biology did not actually pan out in humans.”

In other words, the efficacy results expected in Phase 2 or 3 trials are often not achieved due in part to the fact that the biological systems being explored in the preclinical phase were not as connected to the human disease as originally thought.

“We believe that by using these patient-derived cellular systems, the biology

will be a lot closer to what exists in patients,” says Wallace. “Therefore, we think there is a higher probability of achieving the clinical results we expect to see.”

A more patient-centric approach



Until now, most of what we know about the effects of muscular dystrophy in patients has come from studies using muscle biopsies. Biopsies are a painful process made even worse in patients whose muscle tissue is deteriorating. By using models created from donated patient tissue, Fulcrum is able to study what goes on in a patient's cells over time without having to go back to the same patient for additional biopsies.

“We are able to see how things change and evolve, and how we may be able to fix them,” says Wallace. “But we are able to do so without having to repeatedly poke and prod patients. We also allow them to preserve the muscle they have, which is important to patients.”

Muscles have stem cells specific to the muscle. When a piece of muscle is removed from a patient, enzymes are used to dissociate it so that researchers can obtain cells that are protected inside the muscle. These stem (satellite) cells repair and regenerate muscle tissue to help people become bigger and stronger.

The cells can be cultured in a petri dish and grown. Eventually, they are used to make the multinucleated myotubes, which will later mature into myofibers (fiber-like muscle cells), which are the tissues we have inside our bodies.

“The microtissues that we are able to develop in the lab can be profiled and explored functionally,” says Wallace. “So we can actually look at things like twitching and contraction, as these systems spontaneously twitch the way a normal muscle would. This allows us to look at how much force a particular myofiber would be able to sustain, or perhaps look

at the effect of compounds to see if we can restore its function. These are aspects of the disease we would ultimately examine in a trial, only we are able to mimic them in microtissues in in-vitro situations.”

Partnerships speed discovery

Since Fulcrum opened for business in 2016, it has built a relationship with the FSH Society, a patient advocacy group championing FSHD drug development and clinical trials, as well as providing funding for research. The group has been critical in helping Fulcrum understand the patient community and develop clinical outcome targets that are meaningful to both patients and families.



In addition to helping Fulcrum connect with the patient community, the FSH Society has worked to draft a tissue protocol for patient donations through the National Disease Research Interchange (NDRI). The FSH Society partnered with the NDRI to create a nationwide registry of individuals with FSHD that has been integral in allowing the NDRI collaboration to occur.

While the vast majority of the cellular biology work is being performed in-house, Fulcrum collaborates with a broad network of experts. Wallace notes that numerous academic experts are helping to bring different capabilities to bear on the problem. For example, academic clinicians are helping Fulcrum examine clinical end points the company may want to measure. “We do not currently have a candidate in clinical development,” says Wallace. “Still, we are thinking carefully about what clinical measures we would want to assess once we get into the clinic. The clinicians are helping us to determine possible end points.” FSH Watch

Editor's note: Follow Ed Miseta on Twitter @EdClinical. Reprinted with permission from *Clinical Leader*. To read online and subscribe, go to <https://www.clinicalleader.com/doc/tissue-modeling-may-cut-clinical-trial-time-0001>.

For neurologists who aren't already aware, as of 2015 there are evidence-based guidelines for diagnosing and managing FSHD, published by the American Academy of Neurology. The guidelines are available at <https://www.fshsociety.org/resources/>. They include emphasis on physical therapy and the right kind of exercise. They provide strategies that didn't exist when I was diagnosed.

Regrettably, I felt really alone when I was diagnosed. Today, with access to modernized care guidelines, coupled with the FSH Society's community outreach efforts along with the aforementioned research and trials, I definitely feel less isolated.

With the generally accepted premise that aberrant expression of the DUX4-fl gene is the primary mediator in causing FSHD, there has been a momentum shift from general research in understanding FSHD toward therapeutic research. Adding to this research shift have been significant advances in animal models of FSHD, including both human xenograft and transgenic models that are suitable for both academic and biotech research.

Only five years ago, the number of biotechs working on FSHD therapies was zero. Today, I'm aware of at least a half-dozen that publicize FSHD somewhere in their pipeline from drug discovery to clinical trials, and I'm told the actual number is closer to a dozen. In addition, the FSHD Clinical Trial Research Network (CTRN) has been established with the foresight that in the near future, there will be a significant increase in investigational new drugs entering FSHD clinical trials.

With FSHD being a rare disease, now more than ever we need an increase in FSHD awareness and patient involvement in order to help ensure successful clinical trials. I took part in an FSHD clinical drug trial in 2016. The trial required six sites around the world in order to secure the 16 FSHD patients necessary for the trial.

In addition, there are a number of clinical studies, patient registries, and opportunities to donate blood and tissue samples for research. The FSH Society (<https://www.fshsociety.org/>) is an excellent source to assist anyone looking to participate in any of these activities.

As with any rare disease, it is important to gather as many patients as possible to

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DUX MAY BE A MASTER SWITCH OF THE GENOME

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DUX4 is not simply a toxic accident, but is expressed in the male germline (sperm development) and embryo. What its role might be has become clearer. A quartet of papers has recently asserted that, far from being a bit of toxic genomic trash, the DUX family of genes has a role at the very dawn of embryo development. (DUX4 is the human version; the mouse version is called simply DUX; in this story we refer to them collectively as “DUX genes.”)

In the earliest stage of development, a fertilized egg divides into a cluster of cells that are featureless, quivering orbs of protoplasm. The genome in these undifferentiated cells is silent, like a symphony orchestra waiting for the maestro to pick up the baton. A process called embryonic genome activation (EGA) causes the genome to switch on, and the orchestra to start playing. Inside the cells, networks of genes wake up and drive the development of cells down various pathways, leading to the formation of nerve, muscle, gut, skin—all the diverse cell types needed to build the tissues and organs of a fully functioning organism.

Research teams at the University of Washington, University of Utah, and Karolinska Institutet, Stockholm, Sweden (Hendrickson et al.; Whiddon et al.; Töhönen et al.), reported that DUX turns on a set of genes that is expressed in two-cell (in mouse) or four-cell (in human) embryos. The third paper, by Didier Trono and his colleagues at École Polytechnique Fédérale de Lausanne (EPFL), Switzerland, goes further, making the claim that DUX is the master switch—the Toscanini that wakes up the genomic orchestra, setting into motion the biological program that gives rise to a whole human being.

“We had good knowledge that DUX was doing some germline and stem cell programming, but the new papers really establish this,” said Stephen Tapscott, PhD, of the Fred Hutchinson Cancer Research Center in Seattle, Washington. He is a coauthor on two of the papers (Hendrickson et al. and Whiddon et al.). “In both situations, DUX is expressed at a time point that is important for setting up a permissive genome”—when many genes in the “orchestra” are allowed to make noise—“and that will give rise to an embryo.”

What DUX4 might be doing at the two- or four-cell stage is conferring “totipotency”—the power to become anything—which in

molecular biology terms means “the ability to access all of your genes,” Tapscott explained. “We’ve known for a long time that many of the genes that DUX4 regulates are expressed in FSHD, and at these early time points.”

Why would turning on this early program be bad for adult muscle cells? Tapscott noted that the early-stage development of an embryo involves turning genes on and, just as importantly, turning them off when they are no longer needed. To go back to our orchestral analogy, the conductor not only rouses the violin section, but also hushes the trombones. Tapscott speculates that DUX4 may turn off the “skeletal muscle program,” thereby impairing the muscle’s ability to sustain and rejuvenate itself.

One of the surprising findings reported (in Whiddon et al.) is that mouse DUX has some significant differences from human DUX4. This is quite unexpected, because master genes in embryonic development are typically conserved with few changes from species to species. That’s because they are mission-critical, and any mutations in such a key gene would likely be an evolutionary dead end (that is, the embryo would die, and the mutation would not be passed on to future generations).

This insight could have important implications for developing a better mouse model of FSHD. “When you put human DUX4 into mouse cells, the mouse may not respond in the same way as it would to mouse DUX,” Tapscott said. “Using mouse DUX rather than human DUX4 might be better for modeling some aspects of DUX4 gene activity in the animal.”

In summary, these fundamental studies of the DUX gene family may yield at least two benefits to FSHD families who are waiting for a treatment. They suggest that DUX4’s role in early development could point to what goes awry in patients’ muscles—and how to reverse it. And this research could also contribute to building better FSHD mouse models, which are essential for studying any new therapies.

There may also be a third benefit. The discovery that DUX4 is not a genomic oddity but may play a central role in early development should attract attention from life scientists everywhere. That can only help speed up progress in understanding FSHD.

“It’s all fitting together. What’s nice is that pieces that seemed harder to connect are now getting connected more easily,” said Tapscott.

“The pieces of the puzzle are starting to give us an idea of where things go wrong.” **FSH Watch**

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Many ways to engage

NOT GETTING FSH SOCIETY EMAILS?

Don't miss out on important news and current events. Sign up via our website by clicking “JOIN.” If you are certain you are on our email list, please check your spam or junk folder.

GET SOCIAL!

Find our Facebook, Twitter, and Yahoo! Groups by visiting www.fshsociety.org and clicking on the logos at the foot of the page. Our online communities are great sources of news, advice, and social support.

HAVE YOU MADE A GIFT TO THE SOCIETY IN 2018?

Your donations are tax deductible, and they make a real difference. Please send your gift in the enclosed envelope. Or contribute online at www.fshsociety.org. Thank you!

COMMIT TO THE FUTURE

Take your support to the next level by including the FSH Society in your will. Your bequest sustains our work for future generations. Questions? Please contact June Kinoshita at (781) 301-6649 or june.kinoshita@fshsociety.org.

FSH MUSCULAR DYSTROPHY RECEIVES ICD-10 CODES

... from page 1

World Health Organization (WHO), it is the diagnostic classification standard for all clinical and research purposes. The healthcare industry, clinical providers, information technologists, data administrators, insurance providers, government agencies, and many other stakeholders utilize ICD codes to properly populate electronic health records, track epidemiological trends, and support medical reimbursement decisions.

“The lack of an ICD code specific to Duchenne/Becker and FSH muscular dystrophy has proven a barrier to diagnosis, care, surveillance, research, and access,” explained Pat Furlong, founding president and CEO of PPMD. “While we have established surveillance through the passage of the MD-CARE Act, we have also had to develop costly and time-consuming processes to discern Duchenne, Becker, and FSH muscular dystrophy cases from those extracted using previous ICD codes. With approved therapies, payer decisions are now being affected as overestimates of economic impacts are made when applying the calculations to their datasets. And while care standards have been established, the Centers for Disease Control and Prevention (CDC) has been unable to assess whether those standards are being implemented. The implementation of these new ICD-10 codes will create a systematic and sizeable impact on the diagnostic, care, research, and outcomes landscapes for our Duchenne/Becker and FSHD communities.”

For two years, PPMD had lobbied for

ICD codes specific to Duchenne/Becker. After the second try failed, PPMD teamed up with the FSH Society. Working closely with the CMS and CDC's National Center on Birth Defects and Developmental Disabilities, the joint effort proved successful.

“This is very important news. It shows the value of the FSH Society's scientific and policy work with its nonprofit and agency partners,” said Dan Perez, cofounder and chief science officer of the FSH Society. “Nothing could be more technically specialized from a healthcare perspective than this effort to get an ICD-10 code for each type of primary muscular dystrophy. The benefits down the road for this entire class of muscle diseases will be substantial, and this opens up possibilities for greater clinician awareness, outcomes research, and policy.”

We are grateful to FSH Society Scientific Advisory Board members Kathryn Wagner, MD PhD, of the Kennedy Krieger Institute, and Katherine Mathews, MD, of the University of Iowa, as well as the American Academy of Pediatrics (AAP); Christina Westfield, MD, of the New York State Department of Health; Emma Ciafaloni, MD, of the University of Rochester; and additional representatives of the scientific advisory boards of both PPMD and the FSH Society for their technical expertise. FSH Watch

Editor's note: This article incorporates material from a joint PPMD and FSH Society press release.

DEAR NEUROLOGIST

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get a useful set of data. One example is at the NIH-funded Wellstone Center study, for which patients can donate blood and tissue samples at various clinical sites, including the Kennedy Krieger Institute in Baltimore, Maryland (<https://www.kennedykrieger.org/sites/default/files/patient-care-files/facioscapulohumeral-disease-study.pdf>).

Why is this important? First, involvement with support groups can be very good for your mental health. Newly diagnosed patients may have only a few muscles affected by FSHD, but the disease often progresses substantially with age. It is helpful to have the support of people who have experienced the same path of progression. Outside of a support group, it's unlikely that most FSHD patients will ever meet another patient.

Second, emphasizing support, good care protocols, and physical therapy at the time of diagnosis helps patients get back to living the healthiest life possible while managing the disease.

A person who has just been given the very bad news of diagnosis is confused and often feels helpless. Most times, patients know nothing about this disease and will have many questions. What can I do? How do I live my life? What should I avoid? Am I even asking the right questions? It is important for neurologists to provide guidance on such questions, even if the patient doesn't ask them.

I hope my experiences will serve as lessons learned, and help doctors and patients build productive relationships. FSH Watch

EVENTS CALENDAR

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WEBINARS

All webinars are noon to 1 p.m. US EST. Register online via our Events Calendar.

May 9, John Bach, MD. Dr. Bach is a leading clinical expert on noninvasive mechanical ventilation for individuals with FSHD. He is a professor of neurology and medical director, Center for Noninvasive Mechanical Ventilation at the Rutgers University New Jersey Medical School.

August 1, Kathryn Wagner, MD PhD. Dr. Wagner is director of the Center for Genetic Muscle Disorders at the Kennedy Krieger Institute and a professor of neurology



and neuroscience at the Johns Hopkins School of Medicine. In addition to seeing FSHD patients, Dr. Wagner focuses her research on understanding the genetics and progression of muscle pathology in FSHD and on developing therapeutics.

November 7, Stephen Tapscott, MD PhD. Dr. Tapscott's lab studies gene transcription and expression in normal development and disease, including FSHD. Other research interests include gene and cell therapies for muscular dystrophy. He is a professor of neurology at the University of Washington School of Medicine and has an appointment at the Fred Hutchinson Cancer Research Center.



PROJECT 2025

Spring campaign launches with \$100,000 matching gift challenge



At the FSH Society, our moonshot is to get a treatment for FSHD—an intervention that will slow or halt the muscle deterioration. We have set a target year, 2025, a date our scientific advisors agree is realistic to get the first disease-modifying drug to our families. It is an ambitious goal, but we will succeed as long as we have your support, our shared vision, and the team that will get us there.

You have helped us reach this tipping point. Together, we have come very far. This year, with your continued support, we will go so much further! We will:

- launch a major FSHD therapeutics initiative designed to accelerate the pace and volume of drug development, getting disease-modifying treatments to our families faster;
- equip and empower you to become an effective “army of activists,” because the deter-

mination of a community of patients and families is the only thing that has ever made a difference in the outcome of drug discovery and development.

We expect 2018 to be a transformational year for the FSHD community. This is why a group of visionary benefactors has pledged to match up to **\$100,000 in gifts made by May 31, 2018**. By donating to our spring campaign, you will give us the fuel to reach our moon—a treatment to slow or halt FSHD—by the year 2025. Thank you for being on our team!

To join the spring campaign:

- Donate at www.fshsociety.org.
- Mail a check to FSH Society, 450 Bedford Street, Lexington, MA 02420.
- Call (781) 301-7301.



EVENTS CALENDAR

FUNDRAISING EVENTS

June 2, Fischer's Pine Lake, OH
Cosie Laurello Memorial Run & Walk

September 8, Castle Rock, CO
2018 Colorado Walk & Roll to Cure FSHD

September 15, Dublin, OH
2018 Columbus Walk & Roll to Cure FSHD

September 22, Puyallup, WA
2018 Pacific Northwest Walk & Roll to Cure FSHD

October 21, Los Angeles, CA
Ghostly Gala to Vanish FSHD

CONFERENCES

June 8-9, Las Vegas, NV
2018 FSHD International Research Conference (IRC)

June 9-10, Las Vegas, NV
2018 FSHD Connect Conference



September 30, Baltimore, MD
Mid-Atlantic FSHD Family Day Conference

November 10, Kansas City, KS
Kansas FSHD Family Day Conference

MEMBER MEETINGS

April 28, Leesburg, VA
Mid-Atlantic Spring BBQ

April 29, Boston, MA
New England Member Meeting

May 5, Los Angeles, CA
LA Member Meeting

May 6, Palo Alto, CA
Bay Area Member Meeting

May 19, Tampa, FL
Tampa Member Meeting

May 19, TBD
Minnesota FSHD Spring Gathering

May 19, Denver, CO
Colorado Members Meeting

May 19, Salt Lake City, UT
Utah Members Meeting

May 20, Grand Rapids, MI
Michigan Member Meeting

May 22, Sacramento, CA
Sacramento Member Meeting

October 20, Denver, CO
Colorado Member Meeting

December 2, Palo Alto, CA
Bay Area Member Meeting

VIRTUAL MEETINGS

Open to all!

FSH Society Talk Radio broadcasts live on the last Wednesday of every month at 9 p.m. EST (8 p.m. CST). 2018 dates: April 25, May 30, June 27, July 25, August 29, September 26, October 31, November 28, December 26.

Connecticut Connections meets via webinar on the first Thursday of each month, 7-8:30 p.m. EST. 2018 dates: May 3, June 7, July 5, August 2, September 6, October 4, November 1, December 6.

Western Washington FSH Community meets via Skype on the fourth Saturday of each month, 10-11 a.m. PST. 2018 dates: May 26, June 23, July 28, August 25, September 22, October 27, November 24.

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