

FSH Watch

INCLUDING
DONORS
2010



CONNECTING THE COMMUNITY OF PATIENTS, FAMILIES, CLINICIANS AND INVESTIGATORS

CLINICAL MANAGEMENT

Statins and FSHD

by WILLIAM R. LEWIS, III, M.D.

Clinical Professor of Internal Medicine
Director of Echocardiography
University of California, Davis

Coronary heart disease is the number one killer of Americans. As such, considerable attention has been focused on reducing the impact of heart disease in the United States and other industrialized nations. The best way to attain this goal is through preventative measures such as abstinence from smoking, maintaining healthy body weight, eating a diet with five servings of fruit and vegetables daily, and aerobic exercise. Aerobic exercise is important as it has been shown to lower blood pressure and raise HDL, a protective type of cholesterol.



William R. Lewis, III, M.D.

Patients with FSHD are a special population in that the muscle wasting limits or precludes exercise. Also, muscle is an important target of insulin, and as an individual loses muscle mass, the individual may become insulin resistant (type 2 diabetes). Insulin resistance is a risk factor for heart disease.

When lifestyle is not enough to control the risks for heart disease, we look to pharmacologic agents. The most commonly used and most effective agents are the HMG CoA reductase inhibitors, or statins. This class of drug has been shown to reduce the bad

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Dr. Tapscott addresses the Society's International Patient and Researcher Network Meeting, Las Vegas, Nevada, summer 2010.

RESEARCH

Uncovering the cause of a common form of muscular dystrophy: research team makes second critical advance

by KRISTEN WOODWARD

Reporting on the publication of Stephen Tapscott, M.D., Ph.D., PLoS Genetics, October 28, 2010
Fred Hutchinson Cancer Research Center, Seattle

An international team of researchers led by an investigator from Fred Hutchinson Cancer Research Center has made a second critical advance in determining the cause of a common form of muscular dystrophy known as facioscapulohumeral dystrophy, or FSHD.

In August 2010, the group published a landmark study that established a new and unifying model for the cause of FSHD. The current work, published October 28, 2011, in *PLoS Genetics*, shows that the

disease is caused by the inefficient suppression of a gene that is normally expressed only in early development. The work will lead to new approaches for therapy and new insights into human evolution.

The disease-causing gene, called DUX4, previously had been thought to be a completely inactive gene in humans. DUX4 belongs to a special class of genes called retrogenes, which usually represent unused byproducts of evolution that have no remaining biological function, sometimes called "dead genes."

In contrast, the researchers discovered that the DUX4 protein is abundantly expressed in human germ-line cells, the cells that form the sperm and eggs, which indicates a necessary function early in

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FSH Muscular
Dystrophy

Neuromuscular clinics

Editorial note: FSH Watch will occasionally highlight neuromuscular clinics that we believe might be of interest to patients and their families. This issue includes programs in Colorado and Iowa.

**UNIVERSITY OF COLORADO
 Anschutz Medical Campus
 Neuromuscular Clinics
 Denver, Colorado
 Steven P. Ringel, M.D., Director**



The Neuromuscular Program on the Anschutz Medical Campus of the University of Colorado includes comprehensive research, education and clinical services for patients from a six-state area. The neuromuscular clinic for adults is located at the University of Colorado Hospital (UCH), while the neuromuscular clinic for children is on the same campus at The Children's Hospital (TCH). One of the oldest programs in the United States, the neuromuscular program at UCH was established in 1950 and expanded to include TCH in 1988. Steven P. Ringel, M.D., Professor of Neurology, has directed the UCH clinic since 1976. Dennis Matthews, M.D., Professor and Chair of Physical Medicine and Rehabilitation, and Julie Parsons, M.D., Assistant Professor of Pediatrics, co-direct the TCH clinic. The adult clinic includes three full-time faculty neurologists (Drs. Steven Ringel, Dianna Quan and Yvonne Rollins) and three volunteer community neurologists (Drs. Marc Treihaft, Pierre Pavot and Robert Schabbing). TCH staff include two pediat-

ric physiatrists (Drs. Dennis Matthews and Joyce Oleszek) and three pediatric neurologists with specialty training in neuromuscular disorders (Drs. Julie Parsons, Audrey Yee and Michele Yang).

Both clinics offer comprehensive services, including diagnostic testing with electrophysiologic testing, muscle imaging, and muscle and nerve biopsy services. Genetic testing and counseling are available to help families interpret test results and understand the impact of these results on the rest of the family. Rehabilitation needs are addressed with the assistance of physical, occupational and speech therapists. Since the Anschutz campus is a large regional referral center for complex medical care, subspecialty consultation is readily available for pulmonary, cardiac, ocular and auditory symptoms. Because neuromuscular disorders can be associated with nutritional issues, both clinics offer consultation with a nutritionist. Durable medical equipment vendors are available on site. Muscular Dystrophy Association patient services coordinators are present at all clinic visits to answer any questions about support groups, services and activities for patients with neuromuscular disorders.

In addition to our comprehensive clinical services, we offer a wide variety of educational and research programs on our campus. Since its inception, our neuromuscular program has trained more than 40 neuromuscular physicians, many of whom provide services to neuromuscular patients not only in the U.S. but also in other countries. We conduct basic research studies to uncover the causes of neuromuscular disorders and are actively involved in a host of therapeutic trials designed to reverse the disability

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FSH Watch

WINTER 2011

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It is our editorial policy to report on developments regarding FacioScapuloHumeral Muscular Dystrophy (FSHD), but not to endorse any of the drugs or treatments discussed. We urge you to consult with your own physician about the procedures mentioned.

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Articles may be edited for space and clarity. Every effort has been made to ensure accuracy in the newsletter. If you wish to correct an error, please write to the above address.

Look for us on the internet at: www.fshsociety.org
 Editors: Daniel Paul Perez and Nancy Van Zant
 Editorial assistance: Howard Chabner, Carol Perez and Charles Perez
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caused by neuromuscular illnesses. The clinical trials, which are voluntary, include patients from throughout the Rocky Mountain region.

Our neuromuscular faculty frequently join faculty from other institutions to conduct large-scale research trials and the faculty regularly present their research findings at national meetings. We offer community educational symposia for patients and their families. These programs address both clinical and the latest research discoveries.

For an appointment: call Janet Jones at UCH 303-724-2188; or TCH 720-777-3928.

UNIVERSITY OF IOWA HOSPITAL AND CLINICS

Neuromuscular Clinics

Iowa City, Iowa

Katherine Mathews, M.D., Director

The Neuromuscular Clinic is multidisciplinary, with a core group of experienced health care professionals who provide diagnostic evaluation, consultation and ongoing care. The needs and interests of the individual patient and family dictate the content of the clinic visits. Katherine Mathews, M.D., is a pediatric neurologist who sees patients of all ages and particularly patients with muscular dystrophies. She has a longstanding interest in FSHD, including participation in genetic studies in the early 1990s.

Christina Trout, RN, MSN, provides care coordination as well as genetic counseling, which includes discussion about identifying family members who might inherit FSHD, and providing information about family planning options such as prenatal testing or pre-implantation genetic diagnosis. Linda Boehmer, RN, assists in care coordination.

Other key clinic personnel include a physical therapist, dietician, social worker and research coordinator. All these individuals are also available for consultation and assistance between clinic visits and have longstanding experience with the range of needs of individuals with neuromuscular disorders. Additional medical specialists, such as adult and pediatric pulmonologists, cardiologists, a respiratory therapist and occupational therapist are readily available and interact closely with the neuromuscular team. Colleagues in the department of ophthalmology, Suzanne Q. Longmuir, M.D., and Michael D. Abramoff, M.D., Ph.D., have developed a particular interest in the retinal findings associated with FSHD. Similarly, a speech pathologist, Anne Wallace, has worked with many children and young adults with FSHD on consultation from the neuromuscular clinic. Visits with various specialists are coordinated with the neuromuscular clinic appointments as needed. This is an MDA clinic, and the MDA Healthcare Service Coordinator is available to update families on MDA initiatives like online chat rooms and educational materials.

In addition to medical status updates and concerns, the clinic visits might include information about disease pathophysiology and discussion of research updates. Clinic personnel assist families in interactions with schools and workplaces as needed. It is common for school-aged children to need modified physical education or for teachers to request ideas to adapt classroom activities.



Susan Szmyrgala (nutrition), Carrie Stephan (research coordinator), Kathy Mathews, M.D., Christina Trout (advanced practice nurse, clinic coordinator), Shelley Mockler (physical therapist), and Jim Porter (social work); not in photograph, Karla Laubenthal (research physical therapy)


“ Colleagues in the department of ophthalmology, Suzanne Q. Longmuir, M.D., and Michael D. Abramoff, M.D., Ph.D., have developed a particular interest in the retinal findings associated with FSHD. ”

The neuromuscular physical therapist (PT) can provide consultation to the local therapist who may have limited experience with FSHD. Instruction is provided to patients and their caregivers for daily therapy. The PT is certified in wheelchair positioning/seating and can offer guidance about a variety of adaptive devices, orthotics, abdominal binders and durable medical equipment.

Dr. Mathews is also an investigator in the University of Iowa Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center, directed by Kevin Campbell, Ph.D., and Steve Moore, M.D., Ph.D., and funded by the NIH. The Wellstone Center facilitates close communication between basic scientists, including a monthly meeting to review cases and discuss diagnostic challenges. Dr. Moore directs a muscular dystrophy tissue repository that makes tissue samples available to researchers throughout the world.

The neuromuscular team (PT, social work, Ms. Trout and Dr. Mathews) also holds outreach clinics throughout the state of Iowa to facilitate patient access to specialty care.

For an appointment: call Christina Trout at 319-356-4017. Concierge services are available through the hospital to facilitate a visit to Iowa City.

Dr. Mathews is a member of the FSH Society's Scientific Advisory Board. 

From “children” to “strong adults”

by BARBARA AND JEFFREY BACHE, AND KEVIN BACHE

Over the past two decades, we have made the journey with our son, Kevin, diagnosed with FSHD at ten and a half years of age. He has been aware of his diagnosis from the very beginning. Kevin, now 33 years old, holds a masters degree, owns his own home, and works full time.

From an early age, Kevin has amazed us with how he adapts to adverse situations. In kindergarten, his teacher said he was acting out socially and felt he might not be hearing well. She was right. Until he was around other children in a formal setting, he was reading lips and no one was the wiser. Today, he does quite well in spite of some permanent hearing loss.

Kevin was always quite active. While watching him run the bases during a little league game, we felt something was strange in the way he sort of loped along. That observation led us to the FSHD diagnosis. Since we have no history of this disease in our families, we really had no idea what the future would hold.

From grades one through twelve, Kevin was in an Individualized Education Program (IEP) because of his hearing loss and later because of other issues with FSHD. Throughout the years, we worked closely with the school system to ensure he had tools to succeed such as speech therapy and preferential class seating. Miserable because he couldn't keep up in physical education classes in middle school, he was able to get an exemption. In high school, he was provided extra time to take SATs. For him, this was just one more thing to signal he was different. To say he fought being singled out is putting it mildly. Kevin finally accepted that extra time was just one way to “level the playing field.”

Around age twelve, controlling his physical actions became more difficult. After several years of classes, Kevin stopped taking martial arts; however, he credits martial arts with learning how to control his body to avoid getting really hurt when he falls. Instead of playing high school baseball, Kevin became the equipment manager so he could be part of the team. Eventually, he removed himself from this position. As his parents, watching this unfold was gut wrenching; we just wanted to protect him from being hurt.

Before graduating from high school, Kevin told us he wanted to take one semester off before starting college. Our first reaction was “No way.” Then we decided to negotiate. If he took one course at the community college where the credits would be transferable to a four-year college, we would go along with him. Frankly, we were afraid he



Barbara, Kevin and Jeffrey Bache

would like being away from school and would not want to go on to college. He agreed and began attending the University of Maryland in January.

In college, Kevin began having difficulty getting up from a sitting position. Classmates would assist him in class and on occasion when he fell walking across campus. He met with the campus disability coordinator and then withdrew a semester to re-group before deciding how to proceed.

During this hiatus, we visited Disney World. Watching how hard it was for him to get around, we wanted to rent a motorized chair but Kevin said “no.” He tired easily and frequently returned to the room to rest. After that trip, he decided it was time for his own motorized chair. During our next visit to Disney, he went everywhere in the park. An unexpected perk: we were escorted to the front of long lines!

After six years of stops and starts, during which he discovered he liked learning, Kevin received a much deserved bachelor's degree. Over the next few years while working full time, he completed course

work online and received a Masters in International Relations.

Our ongoing struggle is to find the balance between helping too much versus being there when Kevin needs (and asks for) assistance. We certainly do not always get that one right. Finding a place for him to live away from us to see what he could do on his own was probably the most difficult decision so far. Kevin's resilience surfaced big time when he weathered hospitalization from exhaustion and received approval to work some days from home all the while learning to balance what it takes to live on his own.

Helping our children develop independence is not easy. This disease will often make things more difficult for them. But, this doesn't change the fact that they are preparing for adulthood. If we do not provide freedom for them to make choices allowing for the inevitable mistakes, moving into adulthood could conceivably take much longer. We won't always be there to “help” or “protect” them.

At the point our children leave for college, they are young adults. They will face obstacles and thus we need to trust in their ability to find solutions. It is all part of the growing process. Easy to say; not so easy to do. It is hard to refrain from being a “helicopter” mother/father. But, we can trust they will come to us for help and we need to be there when they do. Making decisions using one's own ingenuity leads to the satisfaction of solving a problem. How

good that feels!

At what age should we talk with our child about FSHD? There is no right or wrong answer. Kevin has known since he was diagnosed, and for our family (with counseling) this has worked well. Together we learned about the disease and how his life is being altered going forward.

Children instinctively know when something is not quite right. They see their friends and may silently wonder “what’s wrong with me.” They don’t need to feel alone. So much of what we do as parents is a trust issue; our children need to trust us. Just as with sex, they could jump to wrong conclusions and think the worst. Without trust and communication, who really knows?

We joined a local FSHD support group shortly after Kevin’s diagnosis where we learned about and saw the effects of the disease and how diverse it is. Kevin didn’t want to attend; we didn’t force him. As we learned more of what he would likely face as he goes through life, we found ourselves shielding him from the realities. We probably told ourselves that maybe this would not be how he would progress. Once he decided to attend in his early twenties, he found some comfort in talking with others who experience many of the same problems.

Whatever balance we have achieved has occurred through discussion, tears, laughter and acceptance (and sometimes not) of our own personalities. A trained third party has been an invaluable asset to help us wade through the intricacies of this disease. People handle adversity differently: some simply ignore the facts; others want to know everything. The majority of us probably fall somewhere in between. Goodness knows, we have stumbled and bumbled and still do. Counseling has been an enormous benefit for all of us. We agree we are in a much better place today thanks to having someone bring things into the open and help us focus. Many times we internalize and push problems away pretending we are OK when

What we have learned over the past two decades:

- Plateaus will occur and adjustments need to be made (physically and emotionally). Practice patience.
- Pain management will be a vital part of the process
- Our children are adjusting to
 - Losing “abilities”
 - The idea of the disease and fears of the unknown
 - Fitting in
 - How others perceive them/how they perceive themselves.
- Accept that they will share with others and not us
- Don’t nag—they will only resist
- Respect their time and space
- Talk about how to work with each other
- LISTEN. Don’t simply impose your ideas/fixes
- Provide opportunities for counseling for them and you
- Provide assists (e.g., motorized scooters/chairs) as needs and wants arise
- In the meantime, let them “live their lives.”

we are not even aware of what we’re doing. Through counseling, we have learned much about ourselves and why we do things. We never expected to be dealing with FSHD in our lives.

We did not want this to be a how-to-do-it piece but rather one that shows an evolution through real life experiences and we hope we’ve done that. Comforting words some years ago from our counselor are now very real for us. Kevin “is a survivor.” That he is! FSH Watch

Kevin facilitated a session for young people at the Society’s 2010 International Patient and Researcher Network Meeting. See page 14.



S & L Marx Foundation awards FSH Society funds for “Living with FSHD” series

Special thanks to the S & L Marx Foundation for a generous grant of \$25,000 for the production of a new booklet in the Society’s series, Living with FSHD. The booklet, “FSHD: a guide for schools,” will focus on issues that young people with FSHD face in their educational settings. We anticipate that it will cover the potential effects of the disease on the student’s performance, the types of school environmental modifications that may be needed,

the subtleties of emotional expression in children with FSHD, issues surrounding communication, and recommendations for social support. Families and patients will be engaged to help identify challenges they have faced, and suggest topics for inclusion in the booklet. The publication will likely be available in hard copy and digitally, in early 2012.

The FSHD clinical team at the University of Iowa Neuromuscular Clinics will

contribute to the publication, with Katherine Mathews, M.D., as lead consultant and Shelley Mockler, physical therapist, serving as editor. Other contributors will include speech pathologists, hearing and language specialists, social workers, parents, educators and others.

If you want to suggest a day-to-day challenge for inclusion, please write to info@fshsociety.org, and we will forward it to the editor. FSH Watch

Annual Donor Report

Gifts to Membership, Research and Education

Gifts received January 1, 2010 through December 31, 2010



The 2010 Fundraising Challenge for FSHD exceeds its goal!

FROM THE EXECUTIVE DIRECTOR, FSH SOCIETY



Dear Friends,

In November, Duncan and William R. Lewis, Sr., M.D., Chairman of the Board of Directors, together with David and Michelle Mackay, member of the Board of Directors, and Corinne Bronfman, Ph.D., a major donor to the Society for many years, joined together to offer a challenge

to the Society's members and friends to support promising new work that may lead to a treatment for FSHD. They asked you to make gifts to the 2010 Fundraising Challenge for FSHD, the most prevalent form of muscular dystrophy, and they agreed to match your gifts up to a total of \$202,000, on a dollar-for-dollar basis, if you sent them by December 31, 2010.

I am delighted to report that hundreds of individuals, foundations and businesses in the FSHD community contributed \$221,000 to the Challenge. The Challenge Team has made their matching gifts in recognition of all of you who have made gifts large and small. Together, you have all contributed \$423,000.

Notwithstanding the extraordinary economic environment in which we find ourselves, the FSH Society has concluded our most successful fundraising year ever, raising over \$1,000,000 from generous friends like you. You help the Society make important progress. Thank you for supporting advances in research, education, outreach and advocacy.

The list that follows includes donors to the FSH Society in 2010. We hope you will continue to support this work by joining the Society again in 2011. Looking out over 2011, both Dan Perez and I are extremely optimistic for more rapid developments in mechanistic, translational and therapeutic efforts in FSHD. More than ever, our efforts need to continue and your continued and increased support is vital. Thank you and all good wishes to you and your families for the New Year.

Sincerely,

Nancy Van Zant
Executive Director

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Family and friends of these individuals made gifts in their memory in 2010

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Members of the FSHD Future Fund are individuals who have included the FSH Society in their estate planning

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 Herzberg Family
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 Kelly Family (Lucero, Wing, Paladino, Cuoco, Gregorio, Owens, MacDonald)
 Lee Family
 Tim Passon

We deeply regret any errors or omissions

Gatherings of Members and Fundraising Events, 2010

JANUARY
The Roaring 20s: A Wonderful Night of Nonsense
 Hyde Park, NY
 Arlene and Gary Cohen, MD, Chairs

MARCH
Dodgeball Tournament
 Eastern Senior High School
 Voorhees, NJ
 Jeremy van de Rijn, Leader

APRIL
12th Annual After Tax Season Celebration
 Dorchester, MA
 Ellen & Chris Stenmon, Chairs

2010 Venture Out Project
 Abilene Christian University
 College of Business
 Administration
 Abilene, TX

MAY
Frederick County, MD, Gathering
 Hosted by Doris and Henry Walter

Festive Evening of Music and Song
 New York, NY
 Judy Seslowe and Beth Johnston, Chairs

JUNE
Fulmer Family Dinner
 McDonough, GA
 Rod and Brenda Fulmer, Chairs

Quilt Raffle
 Hopewell School Kindergarten
 Class
 Taunton, MA
 Ann Marie McKenney, Teacher

Aussie Barbie
 Hickory Corners, Michigan
 Hosted by Michelle and Dave Mackay

JULY
Yard Sale and Bake Sale
 Fire Island, NY
 Grace Corradino & Brian Kerr, Chairs

FSH Society International Patient/Researcher Network Meeting
 Las Vegas, NV

SEPTEMBER
60th Birthday Party
 Mobile-Tensaw Delta, AL
 Hosted by Ann Biggs-Williams

OCTOBER
1st Annual Walk 'n' Roll
 Irvine, CA
 Amy Bekier and Mimi Garcia, Chairs

2nd Annual Walk 'n' Roll
 Cape Cod, MA
 Rich Holmes, Chair

Masquerade Ball
 Lambertville, NJ
 Hosted by Josh Bobrovcan and Jen Hunter

San Francisco, CA, Gathering
 Hosted by Howard Chabner and Michele DeSha

NOVEMBER
ING New York City Marathon
 Geoff Bello, Runner
 Jeff Johnston, Coach



Bay Area members meet on October 31, 2010



Michele DeSha and Howard Chabner, hosts, with guest Helen Klaben Kahn



Howard Chabner, right, with guests

UNCOVERING THE CAUSE OF A COMMON FORM OF MUSCULAR DYSTROPHY: RESEARCH TEAM MAKES SECOND CRITICAL ADVANCE

... from page 1

development. Normally, the DUX4 gene is suppressed in all other cells of the body. However, the mutation that causes FSHD makes this suppression less efficient.

“The result is that the DUX4 gene occasionally escapes the inefficient suppression and is expressed in some muscle cells, similar to the Old Faithfull geyser that is usually off but occasionally releases a burst of water,” said corresponding author Stephen Tapscott, M.D., Ph.D., a member of the Hutchinson Center’s Human Biology Division. “The occasional ‘bursts’ of DUX4 are thought to be toxic to the muscle cells, which leads to muscle cell death and the muscular dystrophy.”

Tapscott led the study in collaboration with Daniel Miller, M.D., Ph.D., at the University of Washington, and co-authors Silvère van der Maarel, Ph.D., and Rabi Tawil, M.D., at Leiden University Medical Center and the Fields Center for FSHD and Neuromuscular Research at the University of Rochester, respectively.

Previously, these same investigators had shown that the reason some people are protected from getting FSHD is that they have mutations in a region of DNA that is necessary to stabilize the DUX4 gene product. These new findings confirm the role of the DUX4 protein in FSHD and reveal a new

mechanism of human disease caused by the inefficient suppression of a retrogene that has a role in early development. These findings will provide a focus for future development of therapies for FSHD.

There are broader implications of the new research for understanding human evolution as well. Maintenance of a functional retrogene in humans indicates that it provided some selective advantage during evolution.


“Since FSHD is characterized by excessively weak upper extremity muscles and facial muscles, we speculate that the DUX4 retrogene might have a normal role in causing the weaker and more expressive facial muscles in humans compared to non-human primates,” Tapscott said. “If this suggestion is correct, it means that FSHD is caused by increasing the normal role of DUX4 and causing a more extreme weakness of facial and upper extremity muscles. It also means that all humans have a little bit of FSHD and that this contributes to the evolution of these muscles.”

“The progress was made possible by an unusual degree of collaboration and data-sharing among the individual groups,” Tapscott said.

Grants from the NIAMS and NINDS sections of the National Institutes of Health,

the Friends of FSH Research, the Shaw Family Foundation and the Muscular Dystrophy Association also supported the work of Tapscott and colleagues at the Hutchinson Center.

Other funding for this study came from the Fields Center, the Netherlands Organization for Scientific Research, the Netherlands Genomic Initiative, a Marjorie Bronfman Fellowship grant from the FSH Society, the Centro Investigacion Biomedica en Red para Enfermedades Neurodegenerativas, the Basque Government and the Instituto Carlos III, ILUNDAIN Fundazioa.

To read the complete “Faciocapulo-humeral Dystrophy: Incomplete Suppression of a Retrotransposed Gene,” October 28, 2010, by Lauren Snider, Linda N. Geng, Richard J. L. F. Lemmers, Michael Kyba, Carol B. Ware, Angelique M. Nelson, Rabi Tawil, Galina N. Filippova, Silvère M. van der Maarel, Stephen J. Tapscott and Daniel G. Miller, please visit the open access Public Library of Science (PLOS) Journal on-line at: <http://www.plosgenetics.org/article/info%3Adoi%2F10.1371%2Fjournal.pgen.1001181> 

This work has been supported by a Marjorie Bronfman Fellowship grant from the FSH Society, among other funding.



Ann Biggs-Williams 60th birthday



Ann Biggs-Williams celebrates her 60th birthday AND the breakthrough research in FSHD with a riverboat trip on the Mobile-Tensaw Delta, Alabama, in September. Thank you to Ann's friends and family for gifts in honor of these great occasions!



New DVD series available for distribution

Many young people shared their experiences of living with FSHD at the Society's 2010 International Patient and Researcher Network Meeting held in Las Vegas, Nevada. Three sessions were facilitated and recorded, and they are now available on DVD. The following topics are included in this series titled "Living with FSHD." We hope that other productions will follow.

Getting to know each other: teenagers and young adults


Facilitators: Kevin Bache and Stephanie O'Meara; patients

Dating and forming lifelong relationships

Facilitators: Stephanie O'Meara and David Younger, Ph.D., Licensed Psychologist; patients

How to motivate young people to exercise

Facilitators: Mallory Murdoch, patient; Craig McDonald, M.D., University of California, Davis

The series was funded by the Dorr Foundation and produced by the FSH Society. Special thanks to Rodney Fulmer, videographer. If you would like to borrow these DVDs for a period of two weeks, please e-mail info@fshsociety.org with your request. 

STATINS AND FSHD

... from page 1

cholesterol (LDL) and reduce the progression of atherosclerosis, heart attack and death. This effect is most profound in higher risk populations, such as those with known heart disease or with high risk features such as multiple risk factors or complicated diabetes. While patients with FSHD have not been specifically studied with this class of drugs, there is no reason to believe that they would be any less effective for FSHD patients than for the general population.

Almost all drugs are poisons by design. They poison an enzyme or a receptor and alter normal body metabolism and function. As such, all drugs can have side effects ranging from simple nuisance to potentially lethal effects. The side effects of greatest concern in patients with FSHD are myalgia, myositis, and rhabdomyolysis. Myalgia is muscle ache without blood test evidence of muscle damage. Myositis is muscle ache with blood test showing elevation of creatine kinase, suggesting inflammation of the muscle. Rhabdomyolysis is severe muscle damage and can be life threatening.

All statins can cause these side effects. The incidence of myopathy, defined as muscle ache with elevated creatine kinase, is between 1 and 2 persons per 10,000 treated. The mechanism is unknown but it is dose

related. Some statins, such as simvastatin, atorvastatin, and lovastatin are fat soluble (lipophilic) and can get into muscle more easily than the water soluble (hydrophilic) statins, such as pravastatin, rosuvastatin, and fluvastatin. Experiments have shown that the hydrophilic compounds are less likely to cause myopathy; in patients with pre-existing muscle disease, such as FSHD, the hydrophilic statins are probably safer.

Another explanation for why the hydrophilic statins may be safer has to do with how the drugs are cleared from the body. The lipophilic statins are predominately metabolized by cytochrome P450 in the liver. Many other drugs are also metabolized by this enzyme. If a patient is taking multiple medications, there may be competition for metabolism or inhibition of the enzyme, leading to elevated drug levels and increased side effects. Compounds that inhibit the metabolism of statins include the antibiotic erythromycin, the antihypertensive agents verapamil and diltiazem, and even large quantities of grapefruit juice. The hydrophilic compounds have other routes of elimination and are less likely to have drug-drug interactions.


In summary, lifestyle changes are the best way to avoid heart disease. When these

changes are not enough, then hydrophilic statins, such as pravastatin can be used to lower the LDL cholesterol. Because of the risk of myopathy in patients with FSHD, statins should be used reluctantly and at low dose with careful monitoring by the patient and physician for side effects. It is important to remember that statins do save lives. If a patient has significant heart disease, the benefits will generally outweigh the risks. In a patient with simple cholesterol elevation and no known heart disease, the risk may outweigh the benefit.

Drug Class Review: Hydroxymethylglutaryl-coenzyme A Reductase Inhibitors (statins) VHA Pharmacy Benefits Management Strategic Healthcare Group and the Medical Advisory Panel <http://www.pbm.va.gov/reviews/HMGStatins04-09-03.pdf>

VHA Pharmacy Benefits Management Strategic Healthcare Group and the Medical Advisory Panel <http://www.pbm.va.gov/reviews/HMGStatins04-09-03.pdf>


Rosenson, R.S. Current overview of statin-induced myopathy. *Am J Med* 2004;116:408-16

Dr. Lewis is a member of the FSH Society Board of Directors and Scientific Advisory Board. 

Marjorie & Gerald Bronfman Foundation renews pledge for FSHD research for 2011

The generosity and commitment of Mrs. Marjorie Bronfman to FSHD research began in 1997 when she began to make research grants to the FSH Society, and she has renewed her commitment for 2011 with a new contribution of \$50,000. Mrs. Bronfman, along with her brother and one of her daughters, is affected with FSHD. Through a process of review and recommendation by the Society's Scientific Advisory Board, grants are awarded for research fellowships (US\$30,000-US\$35,000/year) for research projects that show extraordinary promise to find the cause of FSHD.

The 2011 contribution will generate important progress in FSHD research, as


have the many grants that have preceded it. The FSH Society is deeply indebted to Mrs. Bronfman, to the Marjorie and Gerald Bronfman Foundation, and to other members of her family for the advances that have been made possible worldwide over these years and for the opportunity to continue advances in 2011. Mr. Edward Schechter, brother of Mrs. Bronfman, has been a careful, insightful and active steward of foundation resources, and he deserves much gratitude for these research grants and for the recent research breakthroughs. For more information about research fellowships, please contact Daniel Paul Perez at the FSH Society or at daniel.perez@fshsociety.org. 




NEWS AND EVENTS

Walk 'n' Rolls raise funds on both coasts, October 9, 2010

On Cape Cod (Brewster, Massachusetts), the Walk 'n' Roll for FSH Muscular Dystrophy was a great event and a good day for the FSH Society and FSHD patients. Thank you Rich Holmes, Martha Knapp, and Bob Smith and many other volunteers for helping to raise important funds for FSHD research!

In Southern California at the beautiful Heritage Park in Irvine, the Walk 'n' Roll for FSH Muscular Dystrophy was also a great day. Thank you Amy Bekier and Mimi Garcia for leading others along the walk and raising funds for research. 



Amy Bekier, Walk chair, thanks the City of Irvine, physicians and investigators at the University of California—Irvine, and participants for their support. Co-chair Mimi Garcia, second from right



Damien Garcia on a roll with balloons



Sandra Byrd with grandson Justin and daughter Jessica



Friends enjoy a beautiful fall day on Cape Cod



ING New York City Marathon, November 7, 2010



Geoff Bello runs ING New York City Marathon in 3 hours and 58 minutes AND raises \$11,000 for the FSH Society in honor of Jeff Johnston, friend and colleague at Detwiler Fenton! Thank you, Geoff and Jeff!

DO YOU FOLLOW THE SOCIETY'S FACEBOOK AND YAHOO! GROUPS PAGES?



Since its launch in early 2008, the Society's Facebook page has attracted hundreds of viewers and fans. Go to www.facebook.com and sign up. It is free and easy. Search on FSH Society and join the discussion. Bookmark it and come back often. You can also access the Facebook page and FSH Society Yahoo! Groups forum by going to www.fshsociety.org clicking on the "Community & Reference" menu tab at top of page and then selecting "Online Community" in the left hand vertical navigation menu.

HAVE YOU INCLUDED THE FSH SOCIETY IN YOUR ESTATE PLANNING?

We can all help the Society and its future work by including a bequest to the Society in our will or other estate planning documents, and becoming members of the FSHD Future Fund. If you have already included the Society in your will, we hope you will let us know. If you will allow us to recognize your dedication in the next Annual Donor Report, your example might inspire others. If you have questions about your planning and/or if you want to let us know that you have included the Society in your will, please contact us at info@fshsociety.org. Thank you.

VOLUNTEERS ARE STILL NEEDED FOR MUSCLE BIOPSY STUDY

Go to www.fshsociety.org for more information, or contact Daniel Perez at the FSH Society at 617-658-7811 or biopsy@fshsociety.org or Genila Bibat, M.D., at Johns Hopkins, 443-923-2778.

DOES THE SOCIETY HAVE YOUR CURRENT E-MAIL ADDRESS ?


If we do not have your current e-mail address and you want to be sure of receiving up-to-the-minute information from the Society as news breaks, please send your e-mail address to us at info@fshsociety.org.

Save the Date!


- April 16**
13th Annual End of Tax Season Fundraiser
Florian Hall, Dorchester, Massachusetts
- May 19**
A Festive Evening of Music and Song
New York Botanical Garden, Bronx, New York
- June 11**
Fulmer Family Dinner
Atlanta, Georgia
- October 1**
Annual Walk 'n' Roll
Cape Cod, Massachusetts
- October 9**
Annual Walk 'n' Roll
Irvine, California



SAVE THE DATE



A FESTIVE EVENING OF MUSIC AND SONG

Thursday May 19 

New York Botanical Garden, Bronx, NY
A vaudeville entertainment with Steven Blier and friends
INVITATION TO FOLLOW
To benefit the FSH Society, Inc. and FSH Muscular Dystrophy
For more information, visit www.FSHsociety.org

Interested in participating? Want more information? Go to www.fshsociety.org, or e-mail info@fshsociety.org