A Publication of the Facioscapulohumeral Muscular Dystrophy Society

FALL 2011

FSH Watch

CONNECTING THE COMMUNITY OF PATIENTS, FAMILIES, CLINICIANS AND INVESTIGATORS

CLINICAL MANAGEMENT

FSH Muscular Dystrophy:

Maintaining respiratory health to reduce pneumonia potential

by LOUIE BOITANO, MS, RRT, RPFT
University of Washington Medical Center
Seattle, Washington

Only a small number of the people diagnosed with facioscapulohumerl muscular dystrophy (FSHD) develop symptoms of respiratory muscle weakness and even for these people, such symptoms usually do not appear until later in life. The expiratory muscles, primarily the lower muscles of the abdomen, are often affected. A progressive weakening of the chest and abdominal muscles over time may also cause kyphoscoliosis, a deformity of the spine and chest that can restrict the ability to expand the lungs and chest wall. A small number of people will develop sufficient respiratory muscle weakness to affect their breathing at night. A portion of the population will also develop sleep disordered breathing because of developing weakness in the muscles that control the upper airway.

Breathing muscle weakness and sleep disordered breathing can be supported using bi-level pressure mask ventilation, also called BiPAP. Continuous positive airway pressure (CPAP) may be used if only the upper airway muscle weakness causes sleep disordered breathing. While a majority of people may not develop severe enough breathing muscle weakness to require breathing support, the

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Marshall Wolff/courtesy metrowestdailynews.com

Lexi and a friend during their rotation at the May 2010 sailing competition in the Daniel Green Memorial Regatta at Lake Cochituate in Natick, Massachusetts

LIVING WITH FSHD

From generation to generation

by LEXI PAPPAS

Lexi, now a senior in high school, wrote this paper for her "Memoirs" class. Everyone read his or her memoir aloud. All the students received copies of each other's stories and the teacher asked them to comment. Lexi's classmates were impressed with her story and found it inspiring. They never knew she had any problems and they were glad that she was able to come out and tell everyone about it. It took a lot of courage for her to do that. We thank her for sharing with Watch readers her reflections on her life with FSHD and sports.

September 8th, 2006

I walk over to the sheet of white paper hanging on the wall outside the athletic office. I am filled with anxiety: my hands are clenched, my eyes squeezed shut, afraid to actually look at that piece of paper. I am hoping and praying that my name appears on it.

I open my eyes. Turn around. Walk back to my mom's car.

September 14th, 2007

Last day of Field Hockey tryouts, for a second year in a row. This time, I

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WHAT'S INSIDE

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FALL 2011

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Dear Friends,

I am excited to share with you the date and location of the Society's 2012 International Patient and Researcher Network Meeting: June 30-July 1, in Atlanta, Georgia.

Organized and sponsored by the FSH Society, the meeting is a partnership among patients, families, clinicians and scientists. It will include lectures by and question and answer sessions with leaders in the field of FSHD, reports from major clinical centers and research centers, and educational sessions on clinical advances in FSHD. Popular breakout sessions include topics that participants have requested as well as the interests of teenagers and young adults.

This gathering is held every two years, most recently in Las Vegas in 2010. It is a memorable experience for all who attend. Over the years, patients have formed bonds with each other that endure today. Patients, family members and researchers have gained a better understanding of the challenges each other faces as they navigate FSHD. While the program is formal in the sense that every minute is filled, the two days are relatively informal with sessions provided for individuals to share their experiences living with FSHD.

Atlanta is an accessible city. The hotel is easily reached from the airport by MARTA, the light rail system serving the area. Children and adults enjoy the Georgia Aquarium, Turner Field (home of the Atlanta Braves), the High Museum of Art, the Zoo, the CNN Center, countless other museums and nature venues, shopping and restaurants.

Overnight accommodations will be available at the Atlanta Marriott Marquis. The FSH Society has secured a special conference room rate. The hotel has many wheelchair accessible guestrooms, including some with roll-in showers. Check the FSH Society website in the coming weeks for reservation information. Online meeting registration will also be available in early 2012.

We hope to see you there!

Sincerely,

Nancy Van Zant
Executive Director

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: Nancy Van Zant and Daniel Paul Perez
Editorial assistance: Howard Chabner, Carol Perez and Charles Perez
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DVD still available

Young people living with FSHD—DVD series still available for loan

Many young people shared their experiences of living with FSHD at the Society's 2010 International Patient and Researcher Network Meeting. Three sessions were facilitated and recorded, and they are now available on DVD. The following topics are included in this series which we have named "Living with FSHD."

- Getting to know each other: teenagers and young adults
- Dating and forming lifelong relationships
- How to motivate young people to exercise

The series was funded by the Dorr Foundation and produced by the FSH Society, Inc.; Rodney Fulmer, videographer

If you would like to borrow these DVDs for a period of a few weeks, please e-mail *info@fshsociety.org* with your request.

How You Can Help

by ANNE MARSHALL GILLESPIE

Dear Family and Friends,

In 2005, my daughter, Catherine, was diagnosed with FSHD when she was 25 years old. The diagnosis came after several doctors misdiagnosed the vague symptoms that began when she was in college. This really "knocked the props out from under her" and all of us who love her. No one in our family had FSHD and in fact, none of us had ever even heard of it.

As her mother, I felt so helpless and I wanted desperately to do something. The first step was to learn more about FSHD, so I read everything I could find about the disease. I also attended the July 2006 International Patient/Researcher Network Day for FSHD in Cambridge, Massachusetts, sponsored by the FSH Society. At this conference, I met and networked with many people who are affected by this disease, and with doctors and scientists doing research on it. It became very clear that much is being done all over the world and that there is real hope that a cure, or at least a treatment, is just around the corner if only there is enough funding.

I learned more about the FSH Society at this conference and decided that this organization was the best way for me to channel my energies and my desire to do that something. There are many very worthy causes in our complicated world. However, I always wonder how much goes into the actual research and service to the people who are affected and how much goes to administrative costs, overhead, etc., of the organization. I discovered that less than 20% of the contributions to the FSH Society go toward operations and overhead. Also, I have found that the Society is the best source for up-to-date information and advice.

In the fall of 2006, I sent out a one page letter to all my friends and family telling them about Catherine's diagnosis, some basic facts about FSHD and about the work of the FSH Society. I asked for their help in raising funds for the Society and mentioned that I truly believed in its mission. I worked hard on the letter to make it short and concise, but personal. The majority of the letters were sent via e-mail but for the few family members who do not use e-mail, I just copied the e-mail letter and sent it by regular mail. Essentially, I started with my Christmas card list and my electronic address book. I have continued to do this each October, and I am pleased to say that each year many of my wonderful friends and family regularly contribute. If each FSHD-affected family sent letters just to their closest friends, think what an impact that could make. It really is not much trouble and the response I have had each year has been very rewarding.

Perhaps you will join Marshall and many other Society members, both patients and their family members, by writing to others. Your efforts and these gifts will only accelerate the drive to treatments. For assistance and advice, contact info@fshsociety.org. FSHWatch

Gifts of stock to the FSH Society

As you begin to review your financial affairs as 2011 draws to a close, it will be important for you to consider whether investment market conditions make it advantageous for you to make a gift of stock to the FSH Society at this time.

CONSIDER THE TAX BENEFITS

A gift of appreciated securities held for more than one year may provide significant benefits to you as a contributor, such as:

- **Providing you a charitable income tax deduction for the fair market value of the gifted securities as of the date of gift**
- **Eliminating capital gains tax that would ordinarily become due if you had sold the appreciated securities on the open market and donated the proceeds from the sale to charity**
- **Providing a way to help you to achieve your long-term financial objective of reducing your income and estate taxes**

Caution—Tax benefits are lost if:

- **the stock is sold and then proceeds given**
- **the stock is worth less than you paid for it**
- **the stock has been held for one year or less**

For more information, contact the FSH Society at 617-658-7878, or go to the website, www.fshsociety.org and click on Contribute and Gifts of Stock. You should also consult with your financial advisor before initiating a charitable gift of stock.

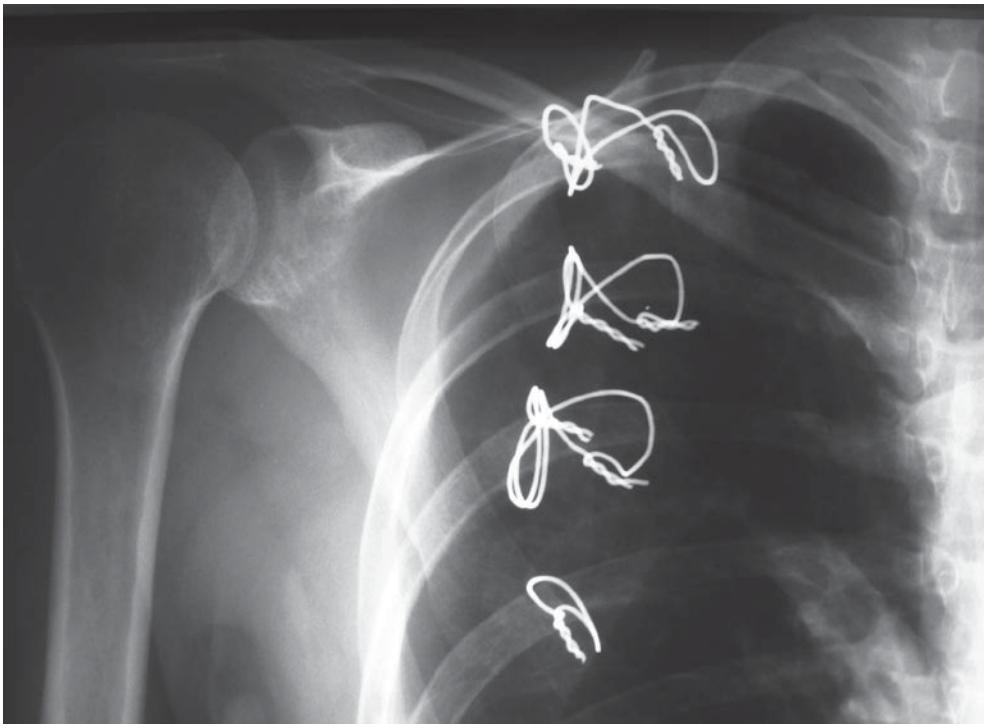
Scapulothoracic fusion in FSHD

by LEIGH ANN CURL, M.D.
Harborview Sports Medicine and Shoulder Surgery
Baltimore, Maryland

Scapulothoracic fusion is an orthopedic operation in which the scapula (shoulder blade) is fused to the underlying adjacent ribs. It has been shown to be an option to improve upper extremity (arm) function in select individuals with FSHD.

Normal shoulder motion and, subsequently, normal upper extremity function occur due to a complex orchestrated interaction between two fundamental muscle groups: (1) the muscles that control motion across the ball and socket joint—primarily the deltoid and rotator cuff, and (2) the muscles that control the motion of the scapula along the chest wall –primarily the rhomboids and trapezius. Normal shoulder function requires integrity of both muscle groups. In many individuals with FSHD the muscles that control the scapula are selectively impaired: the result is a "winging" scapula. This can result in painful irritation of the prominent scapula against hard surfaces such as a chair back, but more significantly leads to loss of the ability to raise the affected arm above shoulder height. As an individual with FSHD tries to raise the arm, the scapula wings uncontrollably relative to the chest wall and the remaining shoulder muscles simply cannot compensate for the lack of normal scapular control. As a result overall motion – and subsequently upper extremity function – is impaired. Achiness or vague discomfort in the shoulder is a common associated complaint and is thought to be due in part to the increased strain on the functioning muscle groups as they attempt to compensate for the lack of proper scapular muscle function.

The goal of scapulothoracic fusion surgery is to achieve stability of the scapula by "fixing" or fusing it to the underlying ribs. This gives the muscles that work across the ball and socket joint a "stable platform" upon which to operate, resulting in improvement in activities that require the hand to be at or above shoulder height. In successful cases overall functional motion is improved, with many patients also noting less fatigue and more sustained strength with overhead activities or reaching. Additionally, a more normal cosmetic posture of the shoulder is achieved.



An x-ray image of a patient following scapulothoracic fusion: a surgical wiring technique has been utilized for fixation of the scapula to the adjacent ribs

A successful outcome requires a careful evaluation of many individual patient factors. Most critical: the remaining muscles of the shoulder (especially the deltoid and rotator cuff) must have normal function. This can be determined by a careful in-office evaluation. A *scapulothoracic compression test* can be done to "simulate" the fusion: the surgeon manually compresses and stabilizes the scapula against the chest wall while the patient raises the arm. In individuals likely to achieve success from a surgical fusion, patients will demonstrate an obvious and often dramatic improvement in their ability to raise the arm.

Other factors, such as the co-existence of lower extremity impairment due to FSHD or the presence of medical conditions such as poor or compromised lung function, can also impact the likelihood of a successful outcome and must be carefully evaluated prior to a recommendation for surgery. Since the post-operative recovery involves significant restrictions on the operated arm (discussed below), individuals who require a cane or walker for assisted walking, or those who are dependent on their upper extremities to assist in transfers out of a bed or chair may not be able to tolerate the post-operative demands of surgery.

The surgical procedure is typically about three hours in length and is done under general anesthesia. An incision is made between the spine and the shoulder blade. The muscle tissue (mostly fibrous and scarred due to the FSHD) around the margin of the scapula and adjacent rib levels is stripped away to allow the bone surfaces to come into contact. The edge of the scapula is then secured to three or four adjacent ribs utilizing metal surgical wires and/or plates (this varies by surgeon). Bone graft from the pelvis is often utilized to facilitate an eventual bone-to-bone fusion in which the scapula and rib bones will permanently grow together. Thus the ability of the scapula to move or wing is eliminated as the scapula is now permanently fixed in place.

Following surgery a four to seven day hospital stay is usually required. Antibiotics are administered for 24 hours to prevent infection and narcotics are utilized

to control post-operative pain. Patients are encouraged to get out of bed with assistance to a bedside chair, on the first or second day after surgery, and should be walking with or without assistance by day three or four. The operated arm is protected in a sling for a total of six to eight weeks. Gentle assisted motion to avoid stiffness may start at three to four weeks, but unprotected motion is typically discouraged until ten to twelve weeks. Heavy lifting and strengthening are discouraged until solid fusion is achieved (typically four to six months). Due to the significant limitations with the operated arm for an extended period of time following surgery, individuals considering this procedure must have a strong support network in place to assist with recovery.

As with any surgical procedure there is the potential for complications. Most common is bleeding that collects between the ribs and the lungs (*hemothorax*). While for most individuals this causes no concern and will resolve on its own, in others it can cause a significant sense of shortness of breath requiring that the blood be drained. This can be done bedside or under ultrasound guidance using a small needle and does not require a return to the operating room. Other potential complications include puncture of the lung (pneumothorax) and infection. Some individuals, especially those who are extremely thin, may complain of sensitivity along the site of the surgical wires or plates, necessitating that they be removed months after the fusion site has matured. Failure of the fusion to occur over time (a non-union) is rare and has been most commonly associated with smoking or patient non-compliance with the postoperative restrictions.

Following successful fusion individuals can return to activities without any additional imposed restrictions. For properly selected patients the resulting improvement in upper extremity function is dramatic. Most individuals can get their hand easily to the top of the head (most times slightly higher) and can sustain a controlled reach with relative ease. It is typically not possible to achieve complete overhead motion (hand directly overhead to ceiling) as this usually requires normal movement of the scapula, which has been restricted by the fusion. One "compromise" of note is that following fusion the ability to reach behind the back and touch along one's spine can be more limited after surgery than before: thus, improvement in overhead function can come at the "cost" of more limited motion reaching behind the back.

Unfortunately, for some patients the overall range of motion and function that they ultimately achieve following surgery can fall short of expectations. This can occur from a number of factors: less than ideal scapular position on the rib cage, stiffness that develops in the ball and socket joint following surgery, unrecognized compromise of the deltoid and cuff muscles prior to surgery. For some individuals, progression of the FSHD over time to involve muscles not previously affected can also compromise long-term function.

Scapulothoracic fusion may be an option to improve upper extremity function in select individuals with FSHD. An office evaluation and comprehensive discussion with an orthopedic surgeon experienced in the care of individuals with FSHD can determine if surgery is an appropriate consideration. Proper patient selection, compliance with post-operative restrictions, and a strong support network during recovery are all critical in achieving a successful outcome following surgery. FSH Watch

Charitable IRA Rollover Opportunity Extended to December 31, 2011

The U.S. compromise tax bill ("Tax Relief, Unemployment Insurance Authorization, and Job Creation Act of 2010") extends until December 31, 2011, a provision known as the "Charitable IRA Rollover" that allows taxpayers age 70 1/2 or older to make tax-free transfers of up to $100,000 per year directly from their Individual Retirement Accounts (IRA) to charities.

U.S. taxpayers can make distributions directly to one or more charities from your traditional or Roth IRA, as long as you are at least 70 1/2 years old when you transfer the gifts. Such gifts can be made without increasing your taxable income or withholding.

In a nutshell:

ELIGIBILITY: Individuals 70 1/2 years and older
AMOUNT: Up to $100,000
METHOD: Check sent from your IRA administrator to the FSH Society
HOW TO INITIATE: Call your IRA administrator for instructions and notify the Society.

Check with your advisors about the best ways to take advantage of this opportunity to give to the Society. As always, the Society will also be pleased to assist you in any way possible.

Pregnancy and birth outcomes in women with FSHD

by EMMA CIAFALONI, M.D.
University of Rochester Medical Center
Rochester, New York

Emma Ciafaloni, M.D.

FSHD occurs equally in men and women, but women tend to be less severely affected than men. Life expectancy is normal and therefore, as with other slowly progressive muscular dystrophies and chronic neuromuscular disorders, pregnancy is an important event for many women living with FSHD. Unfortunately, very little is known about the course and outcome of pregnancy and delivery in women with FSHD and most available studies address pregnancy in myotonic dystrophy, deal with single case reports, or review series biased toward the complicated cases. Conclusions in the literature are based on anecdotal data and are frequently contradictory. The information from these studies is not therefore representative of the whole group of muscular dystrophies and cannot be transferred to FSHD, a dystrophy that typically affects abdominal and trunk muscles.

There has never been a study in the United States looking at the effects of pregnancy on the course of FSHD. The only published series that includes FSHD cases describes the course and outcome of 58 pregnancies in 27 patients (including 11 with FSHD) with muscular dystrophy or congenital myopathy in Germany (Rudnik-Schoneborn, 1997). Of the 11 women with FSHD, four reported exacerbation of weakness during pregnancy. Of the 26 gestations in women with FSHD, three ended as first trimester miscarriages (13%), one had pre-term labor and was complicated by vaginal bleeding, and two resulted in pre-term birth. One delivery was complicated by a grand mal seizure. Fetal outcome was normal in all 23 children. Two children showed early signs of FSHD and a more severe clinical picture than their mothers.

Neurologists and obstetricians have therefore very limited information to offer when it comes to counseling and managing women with FSHD who are pregnant or are considering becoming pregnant. As a result of many questions on the subject received by the FSH Society, conversations between the Society and physicians at the University of Rochester led to the submission of a grant proposal to study the effects of pregnancy on women with FSHD. The grant request was peer-reviewed and initially funded for three years by the Society.

With our study we wanted to gather information about pregnancy and birth outcome in women with FSHD that would help neurologists and obstetricians counseling and managing women before and during pregnancy. We used the National Registry of Myotonic Dystrophy and Fascioscapulohumeral Muscular Dystrophy Patients and their Family Members established at the University of Rochester with the support of the National Institutes of Health (NIH) to identify women with FSHD willing to participate in this study. The study was funded by a grant from the FSH Society. This was a retrospective study, meaning that the information was collected by a combination of self-reported information from the women participating, and review of medical records pertinent to their pregnancies and deliveries by a neurologist and an obstetrician. A questionnaire was sent out to women with FSHD and 48 women responded. Conclusions were based on a total of 105 gestations and 78 live births. Ten women never had a pregnancy and six of them decided so because of having FSHD.

The study showed that pregnancy and birth outcomes are generally good in women with FSHD. The incidence of prematurity did not differ from the general population but a significantly higher incidence of low birth weight infants was noted. The reason for this remains unclear. The rate of forceps deliveries and operative vaginal deliveries was significantly higher than in the general population even when the diagnosis of FSHD was not yet known. This is due to difficulty with the second stage of labor when skeletal muscle effort is required and is likely due to the abdominal and truncal muscle weakness typical of FSHD even in the early stages and mild cases.

One out of four women reported worsening of FSHD symptoms during pregnancy that, for the most part, did not resolve after childbirth. This is similar to what has been reported by others and is consistent with what many women in our neuromuscular clinic have reported to us anecdotally. The most common complaints in order of frequency were: worsening weakness and difficulty carrying the baby due to worsening of shoulder weakness, followed by less frequent problems such as frequent falling, difficulty carrying the baby due to worsening of leg weakness, worsening or new onset pain, and overall difficulty caring for the baby due to FSHD. Nine out of 10 women who become pregnant reported that they would choose pregnancy again and many added comments to this effect on the questionnaire.

Women with FSHD who are planning to become pregnant should know that pregnancy outcome is generally good in the majority of cases and 90% of women have stated they would choose pregnancy again. Establishing care with a neuromuscular specialist and an obstetrician familiar with FSHD early in the decision stages is highly recommended. Access to a physical therapist also familiar with the unique pattern of muscle weakness of FSHD will help coping with some of the new possible challenges arising from worsening weakness during and after pregnancy or from just having to care for an infant. The need for operative vaginal delivery is higher in women with FSHD, even in those with mild to moderate severity disease. Low birth weight is more common than in the general population. There is a possibility of worsening weakness and pain after pregnancy and this should be taken into account when making a decision.

This work was supported by a FSH Society Delta Railroad Construction Grant and a FSH Society Lewis Family Research & Education Grant.

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come more prepared than I was last year, knowing that I definitely have a shot at making the team. All of the girls line up and we count off every other person to make teams for one last scrimmage. I am ready to go, but I can feel butterflies in the pit of my stomach and can feel the beat of my heart pounding against my chest. Although the coach seems to like me and puts me in first as a starter, I am still nervous that I might not make the team.

I was playing for about fifteen minutes when the coach switched me out with another girl. We were all on the sidelines cheering for our teams.

"Lexi, you played really well out there; you were so close to scoring that goal!" one of the girls said to me.

"Thanks, and I know. I just really want to score a goal, but I could not get there fast enough." I was out of breath saying this, more exhausted than I have been this whole week.

"Yeah, you were so close. But it's funny, I have noticed that when you run, it looks like you are running in slow motion because you have such long strides," she said. "I don't mean you run slow, just that you *look* like you're in slow motion." She giggled.

"Yeah." I nervously giggled with her.

I knew that something was wrong with my running because my mom told me a few years earlier that I may have a problem with running someday. At the time, I did not quite understand what she was saying, but now it was beginning to make some sense.
I decided I would ask my mom.

A few hours passed and tryouts were finally over! I was so happy. It was one full week of hell, well, for an eighth grader anyway. I did not realize at that point how horrible tryouts would be at the high school level; this was bad enough. I was excited for the season to start and I found out, just a few minutes earlier, that I had made the team. Thank God for that.

I walked over to my mom's car. She was always there waiting for me, cheering me on. She was on her cell phone when I walked over, but I could tell she wanted desperately to know whether or not I had made the team.

I could not hide my excitement any longer and I shouted, "Mom! I made the team!" A big smile came over her face; she hugged and kissed me and said over and over how happy and proud she was for my accomplishment. I was feeling all right for an eighth grader, just all right.

I could not stop thinking about what my friend had said earlier about my running. It was stuck in my head: *you look like you are running in slow motion; you look like you are running in slow motion....* I could not get it to stop! It was consuming my mind and I could not hold it in any more. Even though I was almost afraid to hear my mom's answer, I had to ask her.

"Mom, why do I look so funny when I run?" *Woops, it slipped out...my mom usually doesn't like talking about this subject....* There was an awkward silence. I looked at my mom and I thought I saw her eyes welling up with tears. Yikes, what did I say?

"Well, Lex, I guess we need to talk about this since it is going to come into play more, now that you have made the team."

I didn't know what she was talking about. All of a sudden, I had this rush of anxiety, like what I felt two years ago when my name wasn't on the team list. My mind started racing again, *is this what she was talking about a few years ago?*

"You have a muscle problem. I have it too and so does grandpa. It's called FSHD; it's a form of muscular dystrophy. Although you don't have many symptoms right now, they will come, eventually. You may not be able to run in a few years and you may not be able to raise your arms up over your head, just like me."

What? What is this? Does this mean I won't be able to play my favorite sports, like field hockey and lacrosse? Will I ever get better? Is there a treatment for this stupid thing? What are my friends going to think when they hear about this? So this is why I didn't make the team last year? Is this why my running time stinks?

January 2009

I never thought I would ever have to stop playing the sports I had dreamed of playing in college, or that I would have to give up running for life. It's not that I was not *allowed* to run, I just can't physically run. My legs just won't go—they won't do what I want them to do. It's like a battle between me and my legs, trying to force them to move faster but to no avail. It was even a struggle for me to do normal things that people do every day, like walk up the stairs, or participate in gym class with a game of tag and other fun games that involved running. I was not ready to give all this up. I was only 15!! I was only a teenager.

How was I supposed to give up sports? I loved them, played them every day since I was little...field hockey, lacrosse, basketball, soccer. Even though I wasn't on a team for all those sports, I still enjoyed them. *Isn't there one sport I can play?*

"Yeah, I didn't even realize that we had a sailing team, until I heard them announce a sign up meeting today in school," my sister said to my mom.

Wait, sailing? Lincoln Sudbury has a sailing team? I've been sailing my whole life. I can do this. Maybe I should try out.

Present Day, 2011

It's funny because I never knew I would actually end up sailing in high school. It was more of just a summer thing in the past, but now I have been sailing on the Varsity team for three years in a row! People say that I am never one to give up or quit; I never really thought about that until now.

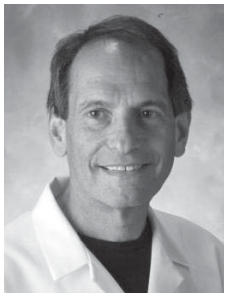
I played two sports for the majority of my childhood, fell in love with them and never quit—until I was forced to quit due to my inability to run. I thought my life was over when I realized I wouldn't be able to play sports like everyone else. How silly to think that way. My life isn't over—it's just beginning. Sailing is perfect for me because I do not have to use my legs and I'm actually really good at it, too. In fact, I am now pursuing sailing in college.

I do not know what the future holds for me. But I've learned one thing—don't give up. Although my body might never be the same, my mind and heart are strong and healthy. I will never give up on my ability to make it in life. I am going to college, I ***will*** sail in college and I will live my life like everyone else.

I'm just a teenager, but I should never have thought my life was over just because I couldn't do the things I missed the most. FSH Watch

Neuromuscular Clinics

Editorial note: FSH Watch will occasionally highlight neuromuscular clinics that we believe might be of interest to patients and their families.



Jerry R. Mendell, M.D.

John T. Kissel, M.D.

FSHD Clinic at Ohio State University/ Nationwide Children's Hospitals

Clinical and Research Programs in FSHD at Ohio State University

by **JERRY R. MENDELL, M.D.,** *Clinic Co-Director* **and** by **JOHN T. KISSEL, M.D.,** *Clinic Co-Director*

FSHD is a top priority disorder for the Ohio State University and Nationwide Children's Hospital (OSU/NCH) muscular dystrophy program. FSHD is a most prevalent and common form of muscular dystrophy; affecting 1 in 14,000 individuals. The OSU/NCH FSHD program encompasses three major areas: 1) a multidisciplinary clinic that provides state-of-the-art care for patients with FSHD, 2) an extensive basic research program to investigate the molecular genetics and pathogenesis of FSHD, and 3) an expanding translational research initiative to apply the knowledge gained from laboratory work to eventual clinical trials in patients.

NCH/OSU FSHD Clinic:

The FSHD Clinic meets in two locations—the Ohio State University Main Campus and Nationwide Children's Hospital—and services both children and adults. The clinics meet weekly at OSU main campus (Monday afternoons) and at NCH (Wednesday mornings). Both clinics are fully affiliated with the Muscular Dystrophy Association and meet for a half day each week. The clinics are true multi-disciplinary clinics staffed by representatives from a wide range of health care specialties, including:

Dr. Jerry R Mendell	muscle specialist
Dr. John T. Kissel	muscle specialist
Dr. Kevin Flanigan	muscle specialist
Dr. Gloria Galloway	muscle specialist
Dr. Albert Tsao	muscle specialist
Dr. Bakri Elsheikh	muscle specialist
Dr. Richard Shell	lung specialist
Wendy King, PT	physical therapist
Linda Lowes, PT	physical therapist
Lindsay Alfano, PT	physical therapist
Theresa Berner, OT	occupational therapist
Jerry Reynolds, Ph.D., RT	respiratory therapist

MDA representative, who provides social services support and assistance.

Additional services for occupational therapy, genetics counseling, social work consultation, and formal medical consultation in the fields of dietetics, orthopedics, gastroenterology, physical medicine and rehabilitation, audiology, ophthalmology, developmental medicine, general pediatrics, and hospice care are available through the OSU/NCH network.

Over the past year, the FSHD Clinic has evaluated patients with FSHD from all parts of Ohio and the eastern United States, including South Carolina, Alabama, and New York, as well as more western states, including California. The clinic is fully affiliated with the FSH Society and the MDA, registers all of its patients with the National FSHD-Myotonic Dystrophy Registry (with the patient's permission), and is a major national referral center for this disease.

Clinical Research Program:

The FSHD Clinic has a history of performing clinical trials in FSHD, including trials involving prednisone, albuterol and the myostatin inhibitor MYO-029. Plans are currently underway for a detailed natural history and clinical outcome study. This outcome study will lay the foundation for further clinical trials that are in the planning stage.

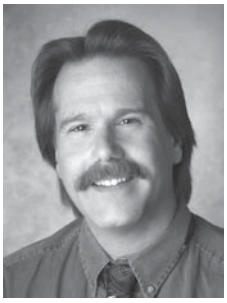
Research Program:

FSHD research advances in this past year have been exceptional. Two groups have contributed significantly to important findings: one group in the Netherlands under the leadership of Dr. Silvère van der Maarel and another group at Nationwide Children's Hospital supervised by Dr. Scott Harper. For the past 20 years

we have known the approximate location of the gene for FSHD. However, confirmatory studies have repeatedly failed to identify the gene with certainty. Research over the past year has changed this picture. We are now virtually certain that the disease is related to a gene called DUX4, found on chromosome 4. Why is this important? Treatment depends entirely on finding the gene, knowing what is wrong with it, and understanding how it causes the disease. We are now armed with information that helps us develop treatment strategies that can be translated into clinical trials. These new findings for the first time put us on the path toward a treatment that will enable us to repress this abnormal gene function. In fact, we are hopeful that many of our patients at OSU and NCH can participate in future clinical trials that will help further understand the abnormal gene function and test agents that can potentially improve this form of muscular dystrophy.

It is a great privilege for us to work with the MDA and the FSH Society and we are committed to doing everything we can to help patients with this disease. Our staff is devoted to this effort and we know we can make a difference!

For a clinic appointment call 614-722-2203.



Gregory T. Carter, M.D., M.S.

▶ MDA-Providence Regional Neuromuscular Center
Olympia, Washington

by **GREGORY T. CARTER, M.D., M.S.**
Medical Director

With all of the recent advances in our understanding of the underlying molecular genetic abnormalities in FSHD, it is hard to believe that we still really do not understand all of the clinical aspects of the disease. This summary will review some of the things we are trying to achieve out here in the Pacific Northwest, as well as outline our future research directions. I am very fortunate to collaborate with a powerful team of brilliant clinicians and researchers. This includes Drs. Tom Bird, George Kraft, and Michael Weiss in our outpatient neuromuscular clinics, as well as Drs. Seth Friedman, Sandy Poliachik, and Dennis Shaw from Radiology, and Drs. Mark Jensen and Ivan Molton from rehabilitation psychology. Together we provide these services for children and adults: diagnostic and genetic counseling, and multi-disciplinary care with muscle expertise: physical and occupational therapy, speech language pathology, respiratory therapy and psychological counseling.

FSHD is a dominantly inherited disorder so it often affects many people within a single family line. While there may be an initial fairly restricted pattern of weakness, this can change quite dramatically as time progresses. Early involvement of the facial and scapular stabilizer muscles results in a distinctive clinical presentation. Typically there will also be foot extensor muscle weakness and pelvic girdle weakness. Atypical patterns of FSHD may include facial muscle sparing and isolated camptocormia, which is an abnormal posture with marked flexion of thoracolumbar spine that resolves in the recumbent position.

Review of research

Available evidence suggests that the disease progresses in a descending fashion, with subsequent involvement of the hip-girdle muscles and then the distal anterior leg. There is wide variability in age at onset, disease severity, and side-to-side symmetry, even among affected members in the same family. Historically FSHD has been described as one of the milder forms of muscular dystrophy yet that is not accurate. Nearly 20% of FSHD patients will eventually use wheelchairs. There can be other associated manifestations, including high-frequency hearing loss, visual loss due to retinal telangiectasias, breathing problems, and chronic pain.1-4 Thus there is still much we do not know about FSHD.

My colleagues and I have been studying muscle anatomy using magnetic resonance imaging (MRI) for some time now. MRI has many advantages, particularly since it is non-invasive yet still reveals a tremendous amount of information about the muscles. MRI could potentially be used as a non-invasive marker to follow the changes in skeletal muscle following an interventional trial with drugs, exercise or gene therapy.

We have recently been able to show that FSHD results in a unique combination of abnormal findings on magnetic resonance imaging (MRI) of the skeletal muscle. This is a big advance because assessment of muscles using limb and whole body MRI is non-invasive and widely available. That paper will be published soon in the journal Muscle and Nerve. 5

We have also been investigating the role of biopsychosocial factors in patients with FSHD and chronic pain. Not surprisingly, FSHD patients report that having a good social support network results in overall better well-being and psychological functioning. Thus it is important for folks with FSHD to build a support network of family and friends to help them deal with this disease.4

On that note, I certainly want to thank the FSH Society for all the help and support they have given me over the years.

For a clinic appointment call Phyllis Fluke, MA, 360-493-4272.

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1 Kilmer DD, Abresch RT, Aitkens SG, Carter GT, Fowler WM, Johnson ER, McDonald CM. Profiles of neuromuscular disease: facioscapulohumeral dystrophy. Am J Phys Med Rehabil 1995; 74(5):S131-139

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4 Miro J, Raichle KA, Carter GT, O'Brien SA, Abresch RT, McDonald CM, Jensen MP. Impact of biopsychosocial factors on chronic pain in persons with myotonic and facioscapulohumeral muscular dystrophy. Am J Hosp Palliat Med 2009; 26(4):308-319

5 Friedman SD, Poliachik SL, Carter GT, Budech C, Bird TA, Shaw DW. The magnetic resononce imaging spectrum of facioscapulohumeral muscular dystrophy. Muscle Nerve 2012; (accepted).

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muscles of breathing may become weak enough to affect respiratory health. If you experience any developing shortness of breath with activity, or poor sleep quality you should ask your rehabilitation medicine physician or neurologist for a referral for a pulmonary evaluation to determine whether you have any breathing muscle weakness. If you do have some weakness this can be monitored over time to determine if it is progressive and whether any therapy should be started to support your respiratory health.

Sighing and coughing are two activities that are required to maintain our respiratory health and are directly dependent upon the strength of our breathing muscles. We all sigh regularly to keep our lungs inflated. If we do not have enough breathing muscle strength to fully breathe in and inflate our lungs we can develop a deflation of the lungs, especially in the lower parts of our lungs. This deflation in the lower lungs can result in a greater risk for developing a respiratory infection. By not taking deep breaths we can also lose the flexibility of our lungs and chest wall, which in turn can also affect our breathing.

If you do not have the ability to keep your lungs inflated a simple therapy called manual hyperinflation or "breath stacking" may be used to help expand the lungs and chest wall. A resuscitator bag with a one-way valve, a flex tube and a mouthpiece at the end (see picture) are required. If you have enough hand strength to pump the resuscitator bag you can do hyperinflation therapy on your own. Otherwise, you will need a caregiver to help you. A hyperinflation maneuver is started by first breathing in as deeply as you can. You, or your caregiver, should then put the mouthpiece in your mouth and repeatedly compress the bag lightly and slowly enough several times to inflate your lungs to a level greater than you can breathe in on your own but that does not cause chest discomfort. You should then hold your breath for five to six seconds allowing the lungs and chest to expand. Repeating this maneuver eight to 10 times while giving yourself time to rest in between maneuvers and doing this therapy two to three times each day can help keep your lungs inflated, and maintain lung and chest wall flexibility. A video clip showing this therapy can be seen at www.irrd.ca/education. Click on the selection "Respiratory Protocols for SCI and Neuromuscular Diseases" and then click on the sub-heading "Interventions." The table of contents has a number of titles with "LVR" (lung volume recruitment). Click on any of the titles for educational information. Clicking on title No. 19 "LVR with resuscitation bag – video" will show you how hyperinflation therapy is done.

Manual Hyper-inflator System including: resuscitator bag, one-way valve, flexes tube length and mouthpiece. (Caution: a second one-way valve housing with the valve leaf removed should be placed between the valve and the mouthpiece to prevent the possibility of inhaling a dislodged valve with hyperinflation maneuvers.)

Having enough strength to cough and clear the mucus we all produce in our lungs is also important in maintaining our lung health. Mucus protects our lungs by removing the foreign particles in the air that we breathe. Mucus is produced by small glands in the walls of the airways of our lungs. The sticky surface of the mucus blanket that forms on the walls of our airways collects the foreign particles that we breathe in. Tiny hairs called cilia that grow from the cells of the airway walls move the mucus and particles up through the airways until we finally cough to clear the mucus from our lungs. Good abdominal muscle strength is necessary to produce enough force to clear the mucus when we cough.

Most people with FSHD have enough cough strength to clear the amount of mucus that we all normally produce. If we develop a respiratory infection we will often produce more mucus than normal, and we need to cough more often to clear the increased amount of mucus. With a need to cough more often already weakened cough muscles can become fatigued and further weaken our cough strength. If we cannot continue to cough and clear the increased amount of mucus it will back up in the airway of our lungs, causing congestion that can affect our breathing. People with neuromuscular weakness that affects their breathing muscles are most often hospitalized for difficulty breathing because of lung congestion that results from a weakened cough during a respiratory infection. For this reason it is important to both self-monitor your respiratory health and to get the support needed to decrease the potential of being hospitalized.

If you do have a degree of breathing muscle weakness, one way of self-monitoring is by using a finger oximeter, a small device that measures your blood oxygen level. This is a relatively inexpensive device that can be purchased without a prescription through a number of Internet medical equipment suppliers. This method of self-monitoring is described by Dr. John Bach on his website www.doctorbach.com. Click on "Outpatient Protocol "on the list of sub-titles to see a description of how to monitor using the oximeter. Monitoring your oxygen saturation can be used to determine whether your breathing is affected if you develop symptoms of a respiratory infection. You should contact your physician if you are developing symptoms of a respiratory infection and cannot maintain an oxygen saturation of 95%. If you develop fever or chills and produce mucus that is thicker and/or yellow or green in color your physician may want to prescribe an antibiotic for respiratory infections.

If you are developing lung congestion that can affect your breathing, you should start therapies that will increase your cough strength to stay ahead of the congestion. This can be done by either manual or mechanical means. Combining manual hyperinflation therapy with an abdominal thrust, also called a "quad cough," can increase cough strength. Cough strength can also be improved by using a therapy called mechanical in-exsufflation using the Cough Assist device. Video clips showing the application of both manual and mechanical cough augmentation therapy can be seen at www.irrd.ca/education. Under Respiratory Protocol for SCI and Neuromuscular Diseases, click on Interventions (LVR with bag, MI-E, ventilator, and GPB), and then click on No. 86, titled "Abdominal Thrust video," to see manual hyperinflation therapy combined with abdominal thrust to improve cough strength. Also at Respiratory Protocol…, click on Cough Assist – New Generation of MI-E, and then click on No. 29, titled "LVR Cough Assist video." There are a number of other subtitles under "Interventions" that provide patient education for cough support therapy at this website. If you have breathing muscle weakness, the key to maintaining your respiratory health is in self-monitoring and starting therapies with the goal of maintaining your respiratory health and avoiding hospitalization. FSH Watch

POOL PARTY RAISES OVER $12,000

Kevin Kirby, left, and friends hosted a great fundraiser for FSHD research in Atlanta, marking summer's end, on September 24. Roman Greene, center, of Memphis, represented the Society and together with many other guests (including host committee member, Host Committee Member Nunzio Lupo, on right) enjoyed hot sun, yummy food, and amazing music selected by Atlanta DJ Mike Pope. Over $12,000 was raised for research and gifts are still coming in!

FAMILY DINNER RAISES $5,000

The Fulmer Family Dinner, June 10, in McDonough, Georgia, raises $5,000 for the Society. Over 125 family members, their friends, and patients attended. Thank you, Rodney and Brenda Fulmer.

UNITED WAY COMMUNITY CAMPAIGNS, FALL 2011

You may have an opportunity to support the FSH Society this fall when you make a United Way pledge for 2011. Check with your human resources department for more information.

COMBINED FEDERAL CAMPAIGN (CFC), 2011 CAMPAIGN

The FSH Society has been approved by the Office of Personnel Management for the 2011 campaign. The CFC is the world's largest and most successful annual workplace charity campaign, with more than 300 CFC campaigns throughout the country and internationally to help to raise millions of dollars each year. Pledges made by Federal civilian, postal and military donors during the campaign season (September 1st to December 15th) support eligible non-profit organizations that provide health and human service benefits throughout the world. The FSH Society's identification number is 10239.

MATCHING GIFTS AND OTHER WORKPLACE GIVING

Many employers offer workers options for directing the company's funds to a charitable organization of their choice. When this opportunity is available to you, please consider how your workplace might make a gift to the FSH Society..

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

Go to www.facebook.com and sign up. It is free and easy. Search on FSH Society and join the discussion. You can also join FSH Society Yahoo! Groups forum, online since the 1990's and with tens of thousands of searchable posts. Bookmark them and come back often. You can also access the FSH Society FaceBook page and Yahoo! Groups 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 vertical navigation menu.

HAVE YOU MADE A GIFT TO THE SOCIETY IN 2011?

Please help now. The FSH Society is a world leader in combating muscular dystrophy. It has provided over $3 million in seed grants for pioneering research worldwide and has developed an international collaborative network of patients and researchers. If you are not already a member, won't you join in this effort? Please return your membership gift, or another gift, in the enclosed envelope. Or contribute online at www.fshsociety.org. Go to Contribute, and select the gift category you wish to make. Thank you.

VOLUNTEERS ARE STILL NEEDED FOR MUSCLE BIOPSY STUDY

To date, approximately 60 individuals in 26 groups of FSHD-affected volunteers and their unaffected relatives have participated in the FSHD-NIH-Wellstone Muscular Dystrophy Cooperative Research Center's research study. Muscle samples are in extremely short supply and tissue donors are most needed. The study is in particular need of patients with suspected FSHD-related hearing and retinal involvement, and from minority races and ethnicities, but all are welcome. In order to determine eligibility, you will need to provide a copy of your gene test result and medical records indicating FSHD diagnosis.
For more information, please contact Doris Walsh at the FSH Society 617-658-7877 or *doris.walsh@fshsociety.org* or Genila Bibat, M.D., Johns Hopkins, 443-923-2778.

DOES THE SOCIETY HAVE YOUR CURRENT E-MAIL ADDRESS?

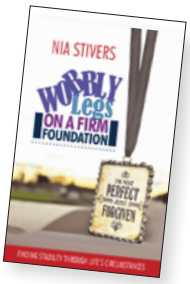
If we do not have your current e-mail address and if you want to be sure of receiving up-to-the-minute information from the Society as news breaks, please send your e-mail address to us at info@fshsociety.org. Thank you.

New books

Defining Moments: What every leader should know about balancing life
By Kees van der Graaf
IMD International, 2011, $40

The author, founder and president of the Dutch FSHD Society, has a 31-year-old son, Bart, who was diagnosed with FSHD in 1992. Since then he and his wife Renée have worked tirelessly to support people with FSHD and to find a solution for FSHD. The book takes you through the life of Kees, his wife Renée, and his sons Bart, Diederik and Michiel. It shows you how they coped with the FSHD of Bart, who also shares his own touching story. As one journalist wrote: "It makes you smile, it makes you cry, and you will also learn something".

To order the book, go to Amazon.com. To learn more about the author, go to *www.imd.org*, and search for Kees van der Graaf. FSH Watch



Wobbly Legs on a Firm Foundation: Finding stability through life's circumstances
By Nia Stivers
Westbow Press, 2011, $13.95

Nia Stivers shares her experience of life with FSHD, her spiritual journey, and the love and support of family and friends: "Being diagnosed with FSH took me from crying on my Disney character pillow each night to thanking God for 'my strongest weakness' with many emotions in between! *Wobbly Legs* takes the reader through that journey from grief to glory. It is my wish to share this book with fellow FSH Muscular Dystrophy comrades in the hope that it will spur them into understanding the gift we have been chosen to live with here on earth. I also believe it is important for family and friends of those with FSHD to read and realize what an important role that they play in that acceptance."

Nia Stivers

Nia will contribute $1 to the FSH Society for every book that you buy as a result of this post. To purchase, go to *www.wobblylegs.com*.

SECOND ANNUAL SOUTHERN CALIFORNIA CELEBRITY WALK 'N' ROLL FOR FSH MUSCULAR DYSTROPHY

Max Adler who stars in the role of "Dave Karofsky" in the mega-hit FOX television series, "Glee," is host of the Second Annual Celebrity Charity Walk 'n' Roll for FSH Muscular Dystrophy, October 9, in Irvine, California.

More than twenty Hollywood celebrities and well known athletes, including Olympic Gold Medal swimmers, Brian Goodell and Kaitlin Sandeno, and winners of Season Number 5 of the Amazing Race, Chip and Kim McAllister, are participating in the event. FSH Watch

KINDERGARTNERS MAKE A QUILT AND RAFFLE IT IN THEIR SCHOOL TO BENEFIT FSHD RESEARCH

The Kindergarten Class at the Hopewell School in Taunton, Massachusetts, has once again made a quilt of the character "Elmer" and held a raffle to benefit the FSH Society. Thank you, Mrs. McKenney and children. We wish you a happy and productive new school year!

Save the Date!

February 11, 2012
Hobe Sound, Florida
Gathering of FSH Society members

Spring 2012
Golf Tournament
Abilene Country Club
Abilene, Texas

June 30-July 1, 2012
FSH Society International Patient and Researcher Network Meeting
Atlanta, Georgia

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