



### *A Word from the CEO*

#### Dear FSHD Global Supporters,

The last six months have proven to be full of achievements in both research and fundraising for the Foundation. There have been a number of highlights with Chocolate Balls in both Sydney and Melbourne raising over \$230,000 for research programs and the Scientific Grants Review Committee (SGRC) reviewing nearly \$3 million worth of funding applications from around the world.

FSHD Global funded researcher Dr. Davide Gabellini recently published his work on the first potential treatment for human patients with FSHD and other frequently inherited muscular degenerative syndromes. Dr. Gabellini emphasised that the \$120,000 in research funding he received from FSHD Global was key to him being able to obtain these novel results.

I would like to congratulate James Wakim and Natalie Moss who have both been appointed to the FSHD Global Board. James is the CEO of the Beirut Hellenic Bank and has been a supporter of the Foundation now for a number of years, while Natalie is a Director of ZMM, a PR and events management company which is also a major supporter of the Foundation. The Foundation is lucky to have two more committed board members who have much to offer and I look forward to working with them.

In May, the Foundation was the major sponsor of the Australasian Gene Therapy Conference. FSHD Global supported two of its international funded researchers, Professor Melanie Ehrlich from the USA and Professor

Alexandra Belayew from Belgium to visit Australia and address the delegates regarding their FSHD research. Professors Belayew and Ehrlich also found time to present their research to both the NSW and Victorian FSHD State Branches.

While developments are fast being made and fundraising has increased, we have not raised enough to fund all the grant applications we would ideally have liked to support.

If you would like to continue your support on an ongoing basis, I invite you to join our Monthly Giving supporter program - by doing so you can make a big difference and ensure future research into FSHD. You choose the amount and can modify or cancel your membership at any time. Small amounts of money over time make a huge difference and the program is a simple way to make a difference on a regular basis.

By filling in the enclosed form and joining the FSHD Global supporter program you are helping to provide sustainable support to FSHD Global's long term commitment to finding a treatment and cure for FSHD and to make up for lost time.

Finally, the annual FSHD Global Golf tournament will be held again this year following the success of previous years. The two day tournament will be held on September 27 at the Terrey Hills Golf and Country Club and November 30 at St. Michael's Golf Club. For more information about registering a team please visit [www.fshdglobal.org](http://www.fshdglobal.org) or contact the FSHD Global office.

Patrick Cameron  
Chief Executive Officer  
[patrick.cameron@fshdglobal.org](mailto:patrick.cameron@fshdglobal.org)

## FSHD SCIENTIFIC RESEARCH UPDATE

### Dr. Gabellini's Research Published

The results of Dr. Davide Gabellini's work on a treatment for FSHD were released this August in an Advanced Online Publication from the highly regarded scientific journal *Molecular Therapy*. His results are extremely important for several reasons.

This is the first time a treatment has tested well for FSHD. Dr. Gabellini's group worked with mice with a similar disease to human FSHD, where a protein called FRG1 is overproduced. They treated the mice with synthetic genetic material to block formation of the FRG1 protein. After a single injection the protein FRG1 was blocked in all of the muscles of their bodies for an extended period of time.

This reduction in FRG1 protein was associated with a real increase in normal muscle appearance, the presence of normal DNA and muscle proteins and an increase in muscle strength. Importantly, there were no adverse side effects of the treatment in these mice that had developed obvious signs of muscular dystrophy before the treatment.

Dr. Gabellini expects that similar types of treatments could also be used to correct other faulty genes thought to be causes of FSHD such as a gene called DUX4. He believes this approach can be used generally in other frequently inherited muscular degenerative syndromes. He also said that scientists know that there are at least 29 faulty genes associated inherited muscle diseases.

Dr. Gabellini stated that overall, these diseases affect as many as one individual in 2,400, making them the most common muscle disorders and that of the three most prevalent muscle diseases (Duchenne, Myotonic and FSHD), Myotonic and FSHD are frequently inherited disorders. He pointed out that research into these diseases has been largely neglected.

## FSHD SCIENTIFIC RESEARCH UPDATE

### Inaugural Bill Moss AM Fellowship

Professor John Rasko, Chairman of the FSHD Global Scientific Grants Review Committee, presented the inaugural Bill Moss AM Fellowship to Dr. Leslie Caron at the FSHD Lindt Chocolate Ball held recently at the Four Seasons Hotel. Dr. Caron is a scientist with five years postdoctoral experience at Harvard Medical School, Boston and is working at Sydney IVF.



Dr. Leslie Caron

*"To date Sydney IVF has derived three stem cell lines carrying the chromosomal deletion which is the molecular cause of FSHD. The availability of disease-affected stem cells is the first step towards a disease model which will help researchers to better understand the disease mechanism and ultimately to assist in the development of effective FSHD treatments,"* said Dr. Caron.

*"The critical next step towards these aims is to differentiate stem cells into muscle cells, the cell type most affected by FSHD and that is what I will be working towards,"* concluded Dr. Caron.

### Finding New FSHD Treatments: Laboratory testing in muscle precursor cells

Professor Melanie Ehrlich, Tulane Medical School, New Orleans, USA

There is no effective treatment for FSHD, a painful, debilitating, progressive disease. Professor Ehrlich and her team recently studied proteins from 17,000 genes in FSHD muscle precursor cells to identify genes that are affected in this disease and they hope their results will suggest novel treatments to make FSHD muscle precursor cells more normal.

Their major findings agree with other laboratories, that abnormalities in FSHD interfere with normal muscle regeneration in patients; that muscle cells are affected by harmful by products of oxygen use; and that inflammation is involved. They have also discovered new proteins and genes that function abnormally in FSHD cells, which were not previously linked to the disease. Prof. Ehrlich's group will use this new knowledge on FSHD to select and test drugs in their laboratory, that are already approved for human use in other diseases, in an attempt to fast track their work to the treatment of human patients.

Further research funded by FSHD Global is about to be published in two international scientific journals (BMC Medical Genomics & PLOS one), by Professor Melanie Ehrlich, Tulane Medical School, USA.

### Evaluation of Synthetic Genes for Inhibition of Faulty Genes in FSHD

Professor Alexandra Belayew, University of Mons, Mons, Belgium

FSHD is a common form of muscular dystrophy in adults, affecting about seven in 100,000 people, causing a progressive wasting of muscles in the upper body. The cause is thought to be the activation of a normally 'silent gene', called DUX4. This group of researchers propose to block the faulty gene, DUX4, in FSHD cells by using a synthetic form of genetic material. They have successfully used this technique in Duchenne Muscular Dystrophy, a similar disease. They plan to observe the effects of blocking the faulty gene DUX4 on muscle development and so identify the cause of the FSHD effects on muscle. This study may thereby identify drugs to treat FSHD.

### Specific Gene Therapy to Correct the Gene Defects in FSHD Embryonic Stem Cells

Associate Professor Michael Kyba, Ph.D., University of Minnesota, USA

Professor Kyba's research plan is aimed at implementing a genetic cure for FSHD by modifying the FSHD cells themselves. Embryonic stem (ES) cells are currently being used to replace defective body cells in a number of different diseases since ES cells themselves can mature into many different types of body cells.

Prof. Kyba's group plans to generate and study the traits in different ES cell lines of the same origin, both the original FSHD cells carrying the defective FSHD associated genes and cells derived from these in which the defective FSHD associated genes have been either corrected to normal or eliminated from the cells genetic material (DNA) completely.

They plan to correct the DNA in two FSHD ES cell lines and then allow the gene corrected and the original FSHD ES cell lines to develop side-by-side into muscle cell precursors or other cell types. They want to do this in order to determine where and when the DUX4 gene protein is abnormally expressed in FSHD.

Finally, the group plans to determine whether cells carrying the FSHD associated gene defects have any observable muscle repair impairment when transplanted into injured muscle of dystrophic mice. These mice have deficient immune responses that allow the transplanted cells to take hold and grow. Their studies should create important new resources for the study of FSHD by themselves and other researchers in the FSHD field and will establish in vitro and in vivo animal disease models of FSHD for study of the effects of the FSHD gene defects on muscle development and regeneration.

Glenn Pilkington, Manager, Grants and Research Development  
glenn.pilkington@fshdglobal.org

For more information on these and other research projects, please visit [www.fshdglobal.org](http://www.fshdglobal.org).

## RECENT APPOINTMENTS

Glenn Pilkington recently joined the Foundation and will take up the newly created position of Manager, Grants and Research Development.  
glenn.pilkington@fshdglobal.org

Komal Jagad has also joined the Foundation as Office Manager.  
komal@fshdglobal.org

## SAVE THE DATE!

Mark these dates in your diary now:

- Tuesday 27 September 2011, FSHD Global Golf Tournament, Terrey Hills Golf and Country Club
- Wednesday 30 November 2011, FSHD Global Golf Tournament, St Michael's Golf Club

## STATE BRANCH NEWS

### FSHD Queensland Group Forms

Recently four FSHD sufferers and a carer met in Brisbane to share their stories about life with FSHD. The meeting provided a fantastic forum for sufferers in Queensland to come together and learn more information about FSHD and its effects. The group would like to continue to meet and recruit new members. If you are interested in attending the next meeting please contact Leona Kelly on 0402 771 109.  
leona.kelly@banorapt.com.au

### Victoria and NSW

The Genetic Support Network of Victoria kindly sponsored a meeting for the Victorian FSHD Branch where international FSHD researchers

Professor Melanie Ehrlich and Professor Alexandra Belayew updated members on their FSHD research.



Victorian FSHD State Branch Members

Melanie and Alexandra also presented to the NSW State Branch members. Both meetings proved to be great forums and allowed FSHD sufferers to ask many questions about where FSHD researching was heading.

## LIVING WITH FSHD

### FSHD sufferer Triecia and mother of three children with FSHD shares her story.

#### FSHD means we live with uncertainty

My son was 13 the day he came home from school and told me he couldn't hold his arms in the air. Then seeing the words facioscapulohumeral dystrophy written on an MRI request shocked my husband and I out of our blissful ignorance as a search of Google saw us recognise the symptoms in our son. We then read of people in wheelchairs and people who needed assistance with eating, of people who could not smile and we were devastated. Surely this could not be happening to our funny, creative and happy go lucky teenager. Our biggest worry for our son was not whether he might end up in a wheelchair, it was how he would cope with this emotionally.



Triecia

#### FSHD means we live with acceptance

I was diagnosed with FSHD six months after our son and it certainly explained a few things. I thought a lot about acceptance when my son and I were diagnosed, I figured I had a choice to either keep mourning the losses associated with FSHD, worry myself sick over the what-ifs or I could accept it and get on with living my life. Acceptance made a lot of sense. This doesn't mean I don't have still the occasional battle between acceptance and a longing for FSHD to just go away – I do, particularly when it makes its presence felt in one of my children.

#### FSHD means we live with those out of the blue sinking feelings every now and then

A few months ago my daughter came home with the observation that she supports her arm when holding her hand

up in tutorials. Two weeks ago her genetic test came back positive. Our beautiful daughter is lucky to have been born with a sunny disposition and she is taking this in her stride. She sent me a text the day after we first saw her geneticist - "Mum I'm noticing things today that I cannot do, but being with the prep kids makes me happy."

#### FSHD means we live with sadness sometimes

When you have a diagnosis like FSHD in the family you automatically look to see who else may be affected. We tried to dismiss the winging going on in the scapulae of our vibrant little eight year old boy, as something that is present in a lot of young children. But every night when I would go in and check on my little boy I was tortured by the sight of him sleeping so peacefully with his eyes open and I knew. We wrestled with whether to have our little boy tested over a long period. The day I heard the news that our little boy had tested positive my heart broke and over the next few days I watched my husband go through grief that felt even harder than my own.

#### FSHD means we live with heartbreak and then we find joy in the present moment

My husband Bill has had to endure the heartbreak of seeing his family members diagnosed with this disease four times. With our heartbreak though has come exhilarating feelings of joy. I am so grateful to have this wonderful man by my side as we watch our little boy's glee on a trampoline, we hear our daughter's passion as she talks of her day in the classroom and we witness our son's excitement at capturing an awesome photograph. Simple things but such precious moments.

#### FSHD means we live with hope

My husband Bill and I also have hope that one day the research that is funded by FSHD Global will lead to a treatment in time to help our children and all those other people living with FSHD who have waited so patiently for hope.

#### FSHD means we need to ask for help sometimes

Your support will hopefully make the road a little easier for our family and for the many other families who are living with FSHD.

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# Fundraising updates

## Melbourne

Thanks to the Victorian State Branch volunteers led by Lucy Burns and Les Jones over 300 people attended and enjoyed the FSHD Lindt Chocolate Ball and raised \$35,000 for research. A great effort by a dedicated group of supporters.

Thank you to everyone who contributed to the night including FSHD sufferer Triecia Gibney who gave a personal account of living with FSHD in a family. Triecia and three of her children have FSHD. Dr. Paul Gregorevic gave an insight into what FSHD is and how the Foundation is approaching research into finding a treatment and cure.



Bill Moss AM (left) with Dr. Lucy Burns and parents

## Sponsors

**Platinum Sponsors** Lindt, Luke Mangan and Moss Capital  
**Gold Sponsors** Astra Mining, David Brown Graphics, Maikhao Dream Resort, The Karma Royal Group, Wyndham Vacation Resorts Asia Pacific and Mulpha  
**Silver sponsors** Intramar, KPMG, Pacific Shopping Centres, ZMM, Beirut Hellenic Bank and Mandarin Oriental

Thank you to everyone who purchased tables and tickets and supplied prizes.

## Sydney

Our thanks goes to the many corporate and individuals that supported the inaugural Sydney FSHD Lindt Chocolate Ball which raised over \$200,000. 450 guests attended the event held at the Four Seasons Hotel and enjoyed a splendid three course meal designed and prepared by celebrity chef Luke Mangan.



The night ran smoothly thanks to MC, Jamie Durie, who kept the night's busy schedule on track. The entertainment was well received with GPO performers and the Undercover Tennors leaving the stage to a standing ovation.

MC Jamie Durie with boxer Danny Green

The inaugural Australian Chocolate Couture Awards were also a highlight. Up and coming designers from across Australia had the opportunity to showcase their chocolate inspired outfits. Thank you to the celebrities involved in the judging – Justine Cullen, Peter Morrissey and Sue Ellen Mackintosh-Dixon, as well as all of the volunteers, who generously gave of their time.

Also a special thanks to a courageous Peter Howes who gave a personal insight into what it is like living with FSHD. Peter's wife and three daughters have FSHD.

## Sponsors

**Diamond sponsors** Moss Capital, Sydney IVF and Bill Moss AM  
**Platinum Sponsors** Lindt, Jamie Durie, Luke Mangan, Mulpha, The Karma Royal Group, Wyndham Vacation Resorts Asia Pacific, ZMM and BBX  
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**FSHD**  
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...making up for lost time

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