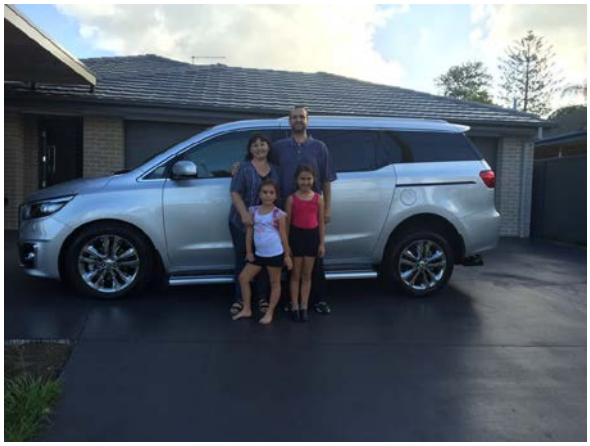
Progress

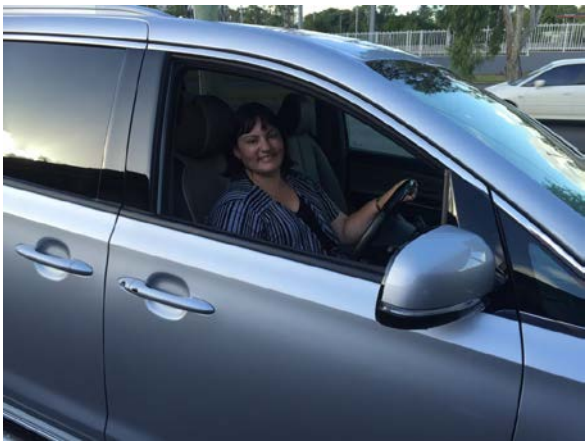
FSHD Global Research Foundation Ltd

Emma's Chocolate Ball car arrives

On 4th July 2015, FSHD Global Research Foundation held their annual Chocolate Ball, with the theme of "Independence Day". I was given the honour of speaking at the Chocolate Ball, sharing my story on living with FSHD. In keeping with the theme, I spoke about how I use health aids and other tools and equipment to maintain as much independence as possible. In my speech, I talked about this and about how an injury I sustained on my shoulder from lifting my wheelchair in and out of the car has meant I now have to purchase an electric wheelchair and a new car that will accommodate it. Of course, the ultimate goal towards independence is a cure for FSHD. While we wait for that though, I would still love to be able to work, take my kids to school excursions and family outings, go shopping, go out with friends and live life to the fullest so the chair and car are both very important.

After my speech had finished, I was back at my table enjoying one or perhaps two glasses of Moet (just to relieve the nervous tension of course!), when our gorgeous host, Jamie Durie, made an announcement. A generous donor, had pledged $150,000 to the Foundation and had also decided to donate a car for me! I was absolutely speechless. In complete disbelief. I may have cried. Not only did this generous man purchase a car suitable for wheelchair conversion, he has organised for Automobility to convert my car so that it will have an electric ramp and I can drive right into the back seat and either remain in my chair for the drive, or exit the car and drive from the driver's seat.





At the ball, I spoke about what an amazing support I have in my husband, Robert, who has been by my side through my falls and surgeries and supported me through my diagnosis and in my life every day. Robert has recently been diagnosed with Lymphoma so is facing a battle of his own. This new car will give me the ability to take on my new role, as Robert's carer and support because it will mean I am independent and able to get to hospital appointments, and require less help from him to get places. I can't imagine how I would have managed if I could not get my wheelchair to the hospital to be there for him. And receiving this amazing gift has helped us financially when we expect to be reduced to one income for at least the next 12 months and would have otherwise been in a position right now that we also had to take out a loan to buy a new car. I feel it is amazing how this has happened to us just at the time when we need it most.

I am planning on attending the Sydney Chocolate Ball again this year, as long as Robert is well enough, so I look forward to seeing many of you there and telling you all about it. It is an amazing, inspiring and hope filled night of spectacular proportions, and for me, it has been life changing. How do you thank someone for a gift of this magnitude? I don't think I can thank Campbell enough, I just hope he really knows the life that he has opened up to my family and I, because he has given me freedom and the ability to be the mother and wife, accountant, and friend that I want to be. Thank you also, FSHD Global Research Foundation, for allowing me the privilege of sharing my story and for working so tirelessly towards a cure that will give us the absolute independence and freedom that is worth waiting for.
Emma Weatherley

Don't forget to join the FSHD AFL tipping competition

The FSHD AFL footy tipping comp has started, but it is still not too late to sign up and be a part of this.

First prize: Signed Collingwood jumper or Mystery accommodation package for 2
Second Prize $200 dinner voucher in CBD
Third Prize: $100 dinner voucher in CBD

Entry fee is $30 with all funds being donated to FSHD Global Research Foundation.

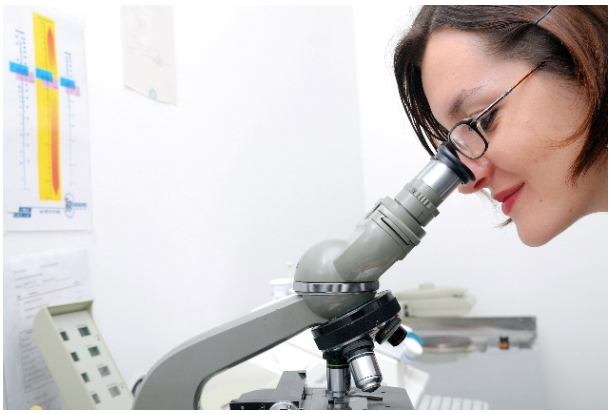
To enter simply follow the details below.
1. Go to http://footytips.com.au
2. Register or login if you are already a member
3. Select "I want to join a competition"
4. Enter FSHD in the search bar. This will take you to the FSHD AFL comp allowing you to join.

Details of fee payment are included once you join the competition. Please be sure to include your name when you deposit money.

If you have any questions at all please contact Les Jones at lesandclaire@bigpond.com

Good Luck!

Small Molecules: Offering hope for people with FSHD



A treatment or cure for FSHD could come from many avenues, gene therapy, stem cell therapy, the development of physical therapies. One potential that many researchers across the globe are exploring is small molecules. These are molecules that are small in size and are biologically active, that is, they influence processes that happen in the body. Most medicines that are available are small molecules.

The creation of large molecule libraries, and the development of high throughput screening methods means that researchers can screen thousands of molecules looking for the ones that produce the effect they want. Small molecule screens offer a powerful tool for researchers to discover not only new medicines, but also uncover new processes that might explain the biology of conditions. Which, completes the circle, by providing new targets for drug-discovery. The screening of small molecule libraries is used routinely in the pharmaceutical industry to find new medicines to treat a diverse range of conditions.

Small molecule screens usually come in two forms. The first is taking a library of biologically active molecules, adding them to cells and seeing if they produce changes in particular proteins or cellular processes. The second is to take your library, add it to a cell system and see whether any cause desired changes in the way the cells behave. (1)

FSHD Global have funded research that uses both these types of approaches. These research projects have the potential to discover new medicines for the treatment of FSHD. The approaches are diverse, one project led by Australian researchers Stephen Palmer and Leslie Caron is screening a set of molecules to see how they affect the development of muscle cells. Professor Michael Kyba from the USA has screened over 35,000 compounds to see if any alter the activity of DUX4 – one of the main genes thought to be responsible for the development of FSHD. Davide Gabellini from Italy has discovered an entirely new mechanism for controlling the genetic area where the mutations involved in FSHD are located. The team are screening molecules that will keep this genetic switch turned off, the effect of this could be to switch off the mutations that cause FSHD.

FSHD Global will continue to work with researchers from around the world and, through the continued support from our community, we will be able to offer funding to help them develop actual treatments for FSHD from promising small molecules.

Seeking 2016 FSHD Global Grant Applications

FHSD Global Research Foundation's mission is to improve the lives of people with FSHD through the advancement of research and development of new treatments. We are therefore committed to funding quality research from around the world.

We have recently opened up applications for 3 Medical Research Grants valued at $1.3 million.

The Monica Ellis Children's Medical Research Fund

In 2015 the Foundation lost a founding Ambassador, enthusiastic supporter and valued friend, Monica Jean Ellis.

Monica was diagnosed with paediatric FSHD when she was just 5 years old. Confined to a wheelchair, unable to sit upright, chew or speak without difficulty, Monica worked tirelessly for the foundation working on finding cures she knew would be too late to assist her. FSHD did not define her, and for every dream FSHD stole from her she dreamt new dreams and made new plans. To Monica "having a disability just throws a spotlight on other choices".

FSHD Global are delighted to announce a medical research grant in honour of Monica's contribution to the Foundation. This grant is made possible by generous charitable donations.

The grant is to fund research into paediatric FSHD.

Diagnostics tender

Diagnostics for FSHD are limited with only a few centres of excellence internationally offering differential diagnosis of FSHD and the sub-types. People with FSHD often have long waits for definitive diagnoses and face many years of symptoms that are not attributed to a specific cause.

The Diagnostics tender is for research that aims to address this issue. This could include, but is not limited to:

- The establishment of patient registries
- Clinical education fellowships
- Infrastructure for testing facilities
- Development of new tests

Therapeutics Tender

Development of new therapies for people with FSHD is the principle aim of FSHD Global Research Foundation. Currently there are no effective treatments and no cure.

As part of the foundations commitment to finding a cure for FSHD we are calling for high quality research projects that address this issue. Projects may be basic science, clinical or translational. However, all must demonstrate the potential for the discoveries to be developed into treatments for people with FSHD. In particular, we will be looking for projects where consideration about the path to commercialisation has been undertaken.