

Gene discovery could lead to FSHD treatment

The importance of international FSHD research collaborations was again highlighted in the news last month; with a Melbourne research team investigating the gene SMCHD1, which is dysfunctional in people with a form of the disease called FSHD2*.

Australian researchers Dr Marnie Blewitt, Dr James Murphy and Ms Kelan Chen from the Walter and Eliza Hall worked with researchers at the University of Leiden in the Netherlands who studied patients with FSHD2

“FSHD is a progressive disease, and we don’t begin to see symptoms until the affected person is in their teens or early 20s,” Dr Blewitt said. “By understanding the function of Smchd1 and how mutations affects its function, we could in the long term develop drugs that would substitute for its activity and prevent the debilitating muscle wasting which occurs in FSHD.”

<http://www.wehi.edu.au/news/gene-discovery-could-lead-muscular-dystrophy-treatment>



Smchd1 is responsible for 'silencing' other genes, and is dysfunctional in people with a form of muscular dystrophy.

Ride to Raise

We recently received a heart-warming email from one of our younger fundraising champion’s family. We are so proud of Justin as he prepares for his "Ride to Raise"

Justin Parke is a fit and healthy 12 year old. However, his grandpa, his uncle and a number of other members of his extended family suffer from FSHD.

During Justin’s lifetime he has witnessed the effects of FSHD on his grandpa. Over the years his grandpa has gradually become weaker and weaker. These days he struggles to walk and do many of the things most of us can do easily.

Justin's uncle, who is quite young, is a professional jazz drummer. Justin is concerned that his uncle will have to stop playing drums if a cure for FSHD is not found soon.

Justin is aware that some people are much more affected by the disease than Justin’s grandpa and uncle. He knows that some sufferers as young as seven years old are wheel chair bound.

Recently, Justin decided he wanted to do something to help scientists to find a treatment and a cure for FSHD. Five of Justin’s friends have decided to support Justin in his quest. The boys and some of their parents plan to ride 70km, from the northern part of the Gold

Coast to the New South Wales border and back, to raise funds for the FSHD Global Research Foundation. The ride is expected to take place on Sunday, 30 August 2015.

Justin is delighted that his school, Ormiston College, is supporting the cause by actively encouraging students and their parents to sponsor the boys.

The boys have already raised \$700 and are aiming to raise at least \$1500. If you would like to sponsor the boys by making a donation to the Foundation, please do so using the following link:

<https://give.everydayhero.com/au/ride-to-raise-2>



Drug Delivery to Patients Muscles

The Institute of Health Sciences and Technologies at the University of Mons (Belgium) has initiated a project to develop very small transporters (nanocarriers) for drugs that could be injected into the blood stream and specifically target muscles - which should hopefully become drugs to fight FSHD.

This \$50,000 collaborative grant funded by FSHD Global Foundation allows to initiate this large interdisciplinary project of drug nano-formation will be shared between chemists (Ir. Alexandra Baroni and Dr. Laetitia Mespouille PhD associated with Prof. Ph. Dubois), pharmacist (Prof. Bertrand Blankert) and molecular biologists (Aline Derenne and Prof. Alexandra Belayew) at the Universities of Mons and University of Sydney (Prof. Richard Payne, School of Chemistry, and Prof. Kim Chan, Faculty of Pharmacy).



Another part of this funding provides a fellowship to Aline Derenne: her task is to identify ligands that specifically recognize proteins present at the surface of muscle cells, and these will be used to decorate the nanocarriers in a later stage of the project, assisting drug delivery to patient's muscles.

Sydney Chocolate Ball

Described as “the Logies of Sydney”, the 2015 Sydney Chocolate Ball was a grand success, having raised one million dollars net to support the Foundation's quest to find a cure for Facioscapulohumeral muscular dystrophy.

It was big...It was bold... and simply the best Sydney Chocolate Ball to date, with 650 guests celebrating the fight for 'Independence' against this debilitating disease.

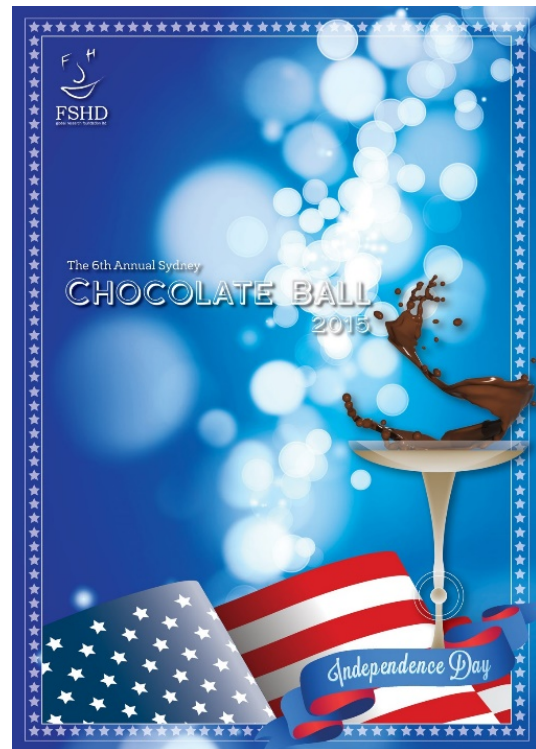
FSHD Patron and Chef Luke Mangan exceeded all expectations with his Americana chocolate inspired menu. This Lindt chocolate feast was coupled with fine Champagnes Krug, Dom Pérignon and Moët & Chandon.

Emcee Jamie Durie OAM kept everyone captivated during what was a jam packed evening of entertainment.

Within the next six months we aim to commit \$2 million towards new medical research and education, focusing on improving Australia's Diagnostics as well as launching a world first human clinical trial, building muscle strength in the body.

We must thank our ongoing supporters and sponsors in our community whose determined efforts, generosity and donated items helped to ensure this sold out event was a grand success. Auction items such as a private lunch with Socceroos legend Tim Cahill, an exhilarating 10 day adventure to the Galapagos Islands, and an absolutely exquisite Chocolate Diamond, all contributed to the excitement and success that is the Sydney Chocolate Ball. We could not produce such a memorable night were it not for the generosity of all those involved.

Be sure to follow us on Instagram (@fshdglobal) to view live photos from the night



Building muscles for those who can't

CrossFit Bondi held a 12 hour rowing challenge at the end of June. An arduous task that saw almost \$5,000 raised and donated to support advancements of FSHD research. It was through the great friendship between CrossFit Bondi's co-owner Glenn Ryan and Justin Reid, which saw 100 strong athletes come together to build their muscles for people like

Justin who have this severe muscle wasting disease, FSHD. Click [here](#) to view their fundraising page.

You can also commence your own fundraising campaign through Everyday Hero.

