FSHD Global Research Foundation Ltd

Science Consensus Meetings

In a short period of time, the Foundation has achieved great momentum and incredible advances, funding the majority of world leading researchers who work tirelessly on various projects to stop this disease.

Last week, FSHD Global held an Australian first FSHD Consensus Summit to discuss recommendations for the diagnosis and clinical management of FSHD. This forum united 12 scientists, researchers, clinicians and experts in the field for high level discussions enabling FSHD Global to publish a paper aimed at educating the medical community in Australia.

The opportunity to learn from world class leaders in this space, was taken on the road, educating patients, families, donors, and the Australian medical community at large. Our scientists visited New South Wales, Victoria and Queensland enabling us to provide updates on FSHD Global's International medical research grants, sharing the latest knowledge and understanding of this disease.

The feedback and attendance was overwhelming, with everyone involved thankful for Dr Rabi Tawil, Dr Stephen Tapscott, Dr Kathryn Wagner, Dr Jean Mah, Dr Baziel van Engelen and Dr Sabrina Sacconi, presentations, commitment and dedication to the field of FSHD.

We look forward to sharing a video of these presentations with you.



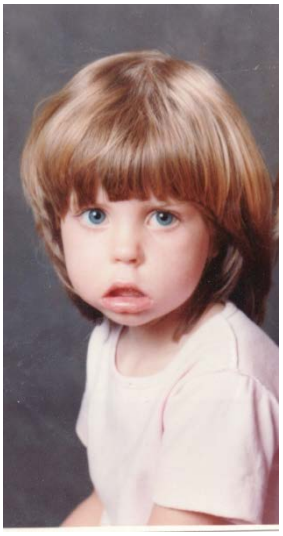

Ride to Raise

We recently shared the story of Justin Parke, a 12 year old who has witnessed the effects of FSHD on his Grandpa and Uncle, and with his friends embarked on a 70 km bicycle ride, raising funds for medical research.

We are proud and ecstatic to report that Justin and his friends successfully raised over $4,000! On behalf of the FSHD Global community, we congratulate you for this achievement and for inspiring others with their own story and fundraising.

This Christmas let the legacy of Monica Ellis live on

As Christmas rapidly approaches, this year we will sell Monica Ellis's Christmas Cards.



Monica was a talented artist, who painted from her view of the world. Diagnosed with infantile FSHD at the age of 5, Monica refused to let this disease describe her. She often said “having a disability just throws a spotlight on other choices."

Monica passed away earlier this year, to say she is missed simply does not do justice to the amazing woman she was and lives she inspired. In honouring our Founding Patron, the Foundation is selling her Silly and Molly Christmas cards, supporting the new Monica Ellis Children's Medical Research Grant, focusing on infantile FSHD.

With several designs, single and mixed packs of 10 are available to order for $30, as are gift tags for $15. Corporate packs are also available should you wish to purchase these for your clients or staff.

This Christmas let the legacy of Monica Ellis live on.

Living with FSHD - Marguerite's Story

My Father Edward fondly known as Ted Carson was the youngest of 5 children born in Hobart Tasmania in 1927.

Dad always had what he called his "wonky shoulder," having no idea of what was actually wrong with him, never having spoken about this with his doctor; he just got on and worked hard at everything that he did, compensating or being inventive to do what he needed to do as his muscles began to let him down.

It was not until my son Ben, at the age of 15 started having problems (he did karate at an elite level, and thought that he had an injury) and was placed under the care of a sports physiotherapist, who suspected that there was more to this supposed injury than met the eye and referred him to a Sports Physician; after some testing the devastating diagnosis of FSHD was found. Of course this opened up a huge can of worms and also shed light on problems other members of the family had or were encountering.

From my father's direct family, it would appear that at least two and possibly three siblings were affected by FSHD to varying degrees, two girls and Dad; one girl (Cecelia) severely disabled, although she did live into her sixties, was affected from her mid-teens.

Cecelia lost the ability to walk unaided when she was quite young but as she never went to a doctor until she was in her late 50 no-one knew what was wrong with her and even when she entered an aged care facility there was still no definitive diagnosis of her condition.

When Dad got married, he and Mum had two daughters, both of us carrying that defective gene. My sister is affected moderately too severely where as I, until recent times have had only minor symptoms. (I am now 60 years of age).

My sister Kathryn had two daughters neither of them having the defective gene.

I have three children a girl and two boys. My daughter is free of the disease but both my sons are affected. My elder son, Rodney, much like me with very minimal symptoms where as my youngest son, Ben who is now 30 has been showing symptoms since his early teens.

Although Dad lived to 87, the last 10-15 years were very difficult for him and the end of his life found him totally incapacitated. It is my hope that some treatment will become available for my sister and son before they find themselves in the same situation as Dad.

Thanks to the fantastic research that is being conducted into FSHD and the resultant genetic testing that is now available, part of our family story with FSHD does have a happy ending

When Rodney (my eldest son) and his wife decided to start their family, little or no thought was given to FSHD and the impact it may have had on their children.

It was only when fertility issues arose and IVF became necessary for them to conceive a child that the option of Genetic Screening was raised.

They were living in New Zealand at the time but travelled to Sydney IVF and started a journey that has been long, with a high cost not just financially but emotionally as well. The result has been a beautiful daughter (Olivia) who is FSHD free and four years after her arrival another daughter, FSHD free, is on the way.

When Ben (5 years younger than Rodney) and his wife Emily wanted to start their family, they were very aware after following Rodney and Anita's journey, and the fact that Emily is a Neonatal Intensive Care Nurse where genetic selection was the only course for them also. Once again the family followed and supported them on this emotional and financially taxing journey, but the joy of them now having two beautiful sons (Connor 3 and Zack 1) free of FSHD makes it all worthwhile.

They have both donated affected embryos for research, which has eased some of the heartache that they have gone through.

It meant so much to Dad to know that finally his family, who he had devoted his life to, was now free of this dreadful disease. It was a burden that weighed heavily on him when he understood that he had passed this disease on to his daughters and thus his grandsons; it has been a heartbreaking journey for us all.

Since putting our journey on paper earlier this year (2015) we have recently welcomed our 6th Grandchild and second FSHD free grand-daughter (Madeline Abigal 03.09.2015).

Marguerite Smith