



Message from the CEO

Over the past 9 years, FSHD Global has been successful in raising \$8.6 million supporting over 40 medical research and education grants in 9 countries. This has seen both the global and local medical landscape change dramatically, advancing research into Facioscapulohumeral muscular dystrophy (FSHD).

We continue to build on the long-standing success of the Foundation as we strive to reach \$10 million of funding to fast-track medical research improving the quality of life for those living with FSHD. Whilst we have achieved a lot, there is still a long way to go.

We are proud this year to launch a series of educational toolkits for patients, GP's and Allied Health professionals, allowing us to advance awareness along with improving the diagnostics and management of those living with FSHD.

Advancing research remains our primary focus. We are actively seeking donations from both our Community and Corporate Partners. We remain committed to allocating 100% of all tax deductible cash donations towards medical research, medical investment and education. This is a statement we are proud of and has allowed us to advance world-class medical research. This is only possible through the generosity and support of our Corporate Partners who generously donate their services, office space, time and prizes.

We are excited about several flagship initiatives in 2017 including working towards establishing a Centre of Excellence for FSHD at a Sydney Hospital. We look forward to your support in 2017.

Join us in making a difference to the lives of those affected by FSHD. Together we can advance medical research finding treatments and an ultimate cure.



Get Positive

You hear it a lot. “Just stay positive.” It’s definitely good advice but sometimes I feel like yelling in their face, “Well OBVIOUSLY !!!”. If it was easy, if I could just flick a switch and be positive I’d do it!

So how do you get positive when you’re feeling overwhelmed and defeated? At the age of 14 I certainly felt like my world had fallen apart. When I was 13 my father left the family. 6 months later I was diagnosed with FSHD. At that age I didn’t really understand what the diagnosis meant. From the faces of my family and the doctors I knew it was serious and it was bad. The specialist explained the nature of my condition and what it meant for my future. He painted a picture of continuous and inevitable physical decline. On a chart you would see the line always falling, with some plateaus where the condition is dormant, but always falling again as the Dystrophy became active. The future was all downhill. How do you get positive about that?

The short answer is that you can’t. I spent a lot of years being angry and fighting the physical decline. Classic denial I suppose! It was a war I could never win. Eventually I asked myself the question, “Why am I fighting this war?” The answer came, “To find peace.” I worked with a counsellor and did some reading, including a book on Buddhist philosophy. One sentence in this book changed my life. It said **“The causes of suffering contain within them the means of release from suffering.”** Somehow this seemed like the answer I was looking for. I thought about that sentence a lot. What was the secret that lay hidden in those words?

Eventually I decided they were telling me to stop blindly fighting and look inside myself with fresh eyes. To see my life as more than just a physical process. To see my life as a journey of personal evolution where adversity drives healing, learning and growth. With this realisation I saw that as my life progressed over time and the FSHD took its inevitable course the difficult times offered rich opportunities for self evolution. Opportunities to find within myself strength, courage, wisdom and compassion. Now the chart of my life became a continuous and inevitable upward line. Now I had something to really get positive about.

Of course it sounds great in theory but living it in the real world is a different story. I still get frustrated and feel down when things are difficult and seem unfair. I still react to situations and have to catch myself before I spiral down into negativity. But now I’m always looking for the lesson, for the opportunity to be wiser and stronger. To be a bigger person. The opportunity is always there.

So now when someone says to me, “Just stay positive.”, I smile and say, “Yeah, thanks.” and think to myself, “I’m already there buddy, I’m already there!”.

JUSTIN REID

If you would like to share your story, please contact the Foundation at admin@fshdglobal.org



Unite to Find a Cure

World FSHD Day is held on **June 20** and aims to raise public awareness for FSHD. Few members of the public have heard of FSH muscular dystrophy, and many affected individuals don't know that they have the condition. This International day aims to bring awareness of this debilitating disease to the general public and decrease misdiagnoses within the medical industry.

Get Creative

Every dollar counts, so no matter how big or small your event is – we'd love your support. Here are some ideas and inspiration to get you started:

Morning or Afternoon Tea

Morning or afternoon teas are a great idea to get everyone involved. Host it at your home or work and ask your friends and/or colleagues to bring a plate, paying an entry fee to attend.

BBQ/Sausage Sizzle

Everyone loves a sanga! Think about putting on a BBQ at your local Bunnings store, school fête, Saturday sports or shopping centre and help raise funds among your community.

Casual Day or Dress Up Day

Hold a casual or dress up day at your work and ask your colleagues to donate a set amount for being involved.

Dinner Party

Get together with your friends and family and host a dinner party. Seek donations of goods or local vouchers from your community to hold a raffle on the night.

School Mufti Day

Get involved with your local school community by organising a school mufti or orange dress up day for a gold coin donation.

Trivia Night

Create a trivia night with your friends or colleagues. Each guest could pay an entry fee to attend with the winner taking home some donated prizes from your community.

Start Planning for World FSHD Day

1 REGISTER YOUR EVENT

It is important you let us know about your event and become an official FSHD Global fundraiser. This way we can help promote your event and assist in any way we can. Let us know all about your event by completing an 'Authority to Fundraise' form to get started. Simply call our office on 02 8007 7037 or email events@fshdglobal.org.

2 FOCUS ON FUNDRAISING

Although World FSHD Day's main goal is raising awareness for the disease, we still aim to raise funds for medical research and education to ultimately find a cure for FSHD. To achieve the best result, put a fundraising strategy in place – how much do you want to raise from your event? How will you achieve this?? Raffles, gold coin donations, entry fees, guessing competitions and sales are all great ways to raise funds.

3 CONSIDER CREATING A PAGE

Consider using a fundraising platform to assist in your fundraising. Everydayhero.com.au is a fantastic website to help you promote your event, receive and track donations. For more details on using Everyday Hero, contact events@fshdglobal.org

4 PROMOTE YOUR EVENT

Make sure everyone knows about your event by posters, email reminders and social media! We encourage you to use social media platforms; Facebook, Twitter and Instagram to promote your event. Send us your poster, announcement, images and we can help promote your event on our official social media sites. If you haven't already – follow us:
Facebook: /FSHDGlobal
Twitter: @FSHD
Instagram: @fshdglobal
Hashtag: #FSHDGlobal, #WorldFSHD & #CureFSHD

5 UNITE TO FIND A CURE

On June 20th, we encourage every one of our supporters to change their Social Media profile pictures to the World FSHD Day logo and use the official Twitter hashtags #WorldFSHD and #CureFSHD.

6 POST EVENT

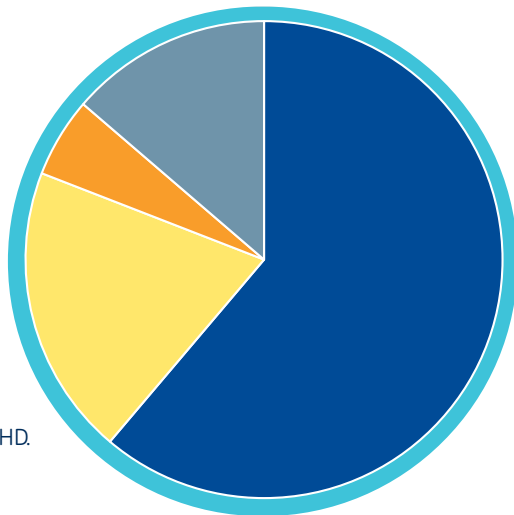
Ensure you take lots of photos of your event and let us know how you went. Don't forget to bank your funds! Ensure you deposit cash donations to your everyday hero page or directly to the Foundation through our website www.fshdglobal.org. We look forward to announcing your achievements and celebrating your success!!

If you have any questions or would like more information about how to get involved in World FSHD Day, please contact us at admin@fshdglobal.org or visit our website www.fshdglobal.org



\$8.6 MILLION FOR RESEARCH

Since 2007, the Foundation has successfully raised over \$8.6 million for medical research aimed at finding treatments and a cure for FSHD.



- \$8,646,869
100% of all tax deductible donations
- \$5,290,380
Medical Research Grants Funded
- \$1,704,643
Medical Research Grants Committed
- \$467,402
Medical Research Education
- \$1,184,444
Funding for pending Medical Research & Medical Education

FSHD Global Research Foundation releases funds to research organisations once a predetermined research milestone has been met.

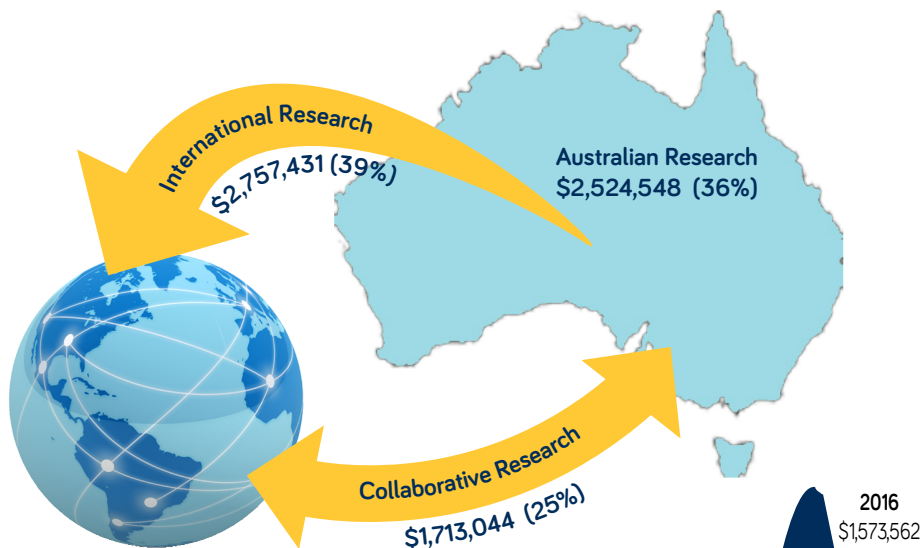
Medical Research Paid to date (75%)

Awaiting research milestone (25%)

41 RESEARCH GRANTS

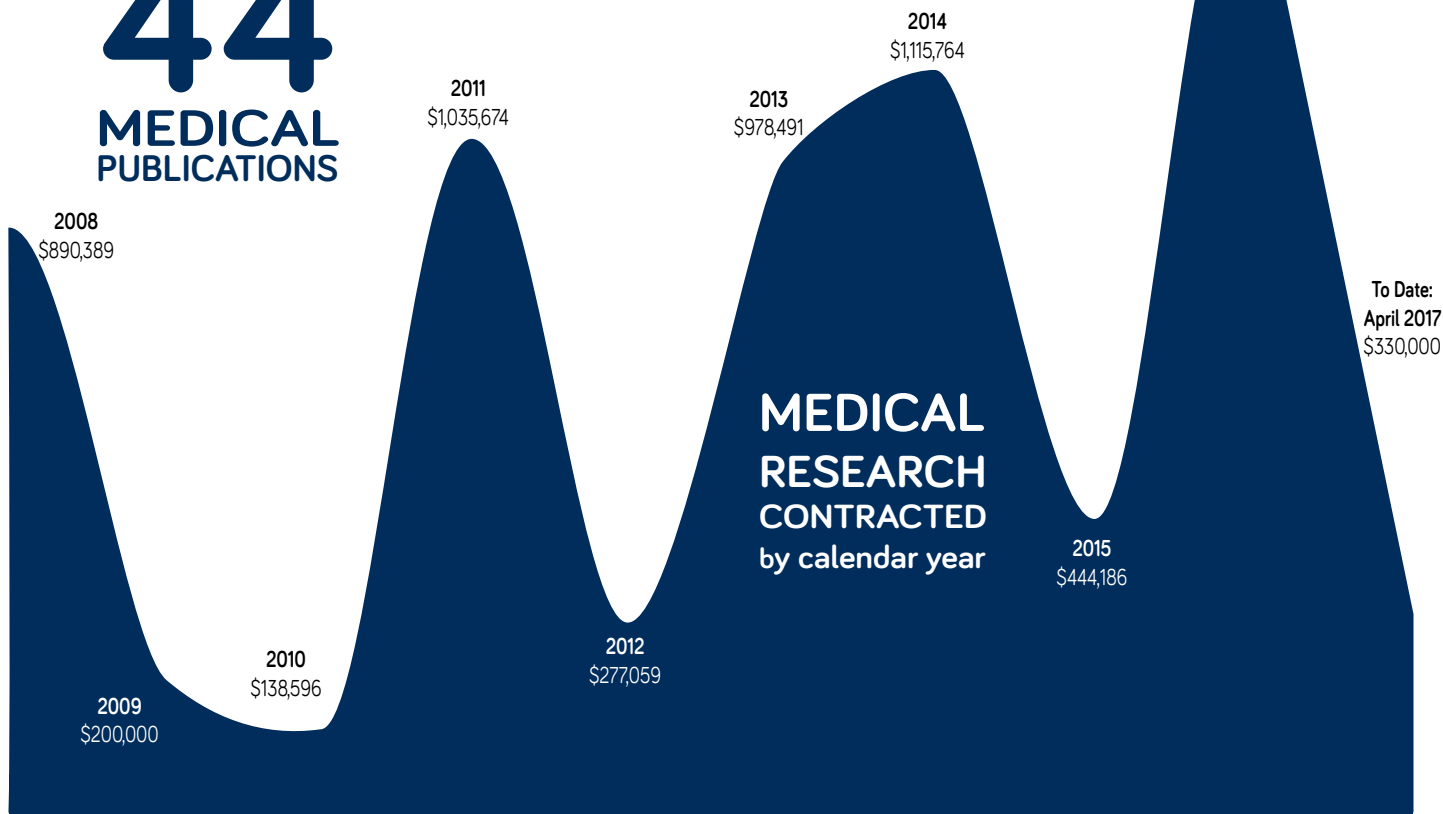
AREAS OF RESEARCH

- Basic 57.5%
- Diagnostic 5%
- Therapeutic 32.5%
- Biotech 2.5%
- Medical Education 2.5%

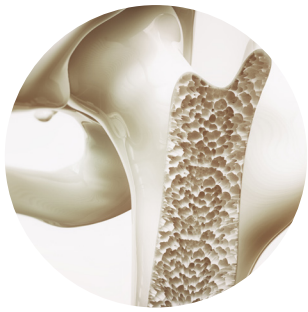


FSHD Global fund research grants in 9 countries: Australia, USA, Germany, Italy, The Netherlands, Canada, France, Spain & New Zealand.

44 MEDICAL PUBLICATIONS



MEDICAL RESEARCH CONTRACTED by calendar year



Bone Health in FSHD

February 18, 2017

In a first for the FSHD field, a study has been published looking closely at the morbidity of low bone density and bone fracture risk in patients suffering from FSHD. The FSHD Global Research Foundation played a particularly proactive role in supporting this study beyond just financial support, including advocating both for not only United States patients but also Australian patients from a Centre of Excellence in FSHD Research at Concord Hospital in NSW to be included in the study. The findings are therefore of International significance. And moreover, a key member of the scientific committee of FSHD Global, Dr Scott Baker, himself an endocrinologist provided expert comments on the design of the study which were acknowledged in the paper.

The study characterised 94 patients with FSHD. One of the key findings of the study was that there is a correlation between strength, physical function and bone density. This could have implications for the clinical management of FSHD patients, for example it may be wise to use lack of strength and physical function to prompt more frequent scans of bone density. It was also noted that there was a high prevalence of abnormal Vitamin D3 levels. Fascinatingly the risk of fractures was 48% in Australia versus 25% in the United States.

Overall the study indicated a high variability in bone density and other characteristics, and having FSHD does not automatically mean a patient has low bone density. However, the prevalence of mostly traumatic fractures in the patient group overall was high. It is the hope of the Foundation that this research paper will draw attention to this important clinical feature of FSHD patients and improve the standards of care not only in Australia but also in other countries. <https://www.ncbi.nlm.nih.gov/pubmed/28214289>

Facio validates over 300 compounds

April 3rd, 2017

Facio Therapies announced today that it has validated more than 300 compounds (so-called "hits") that repress the production of the muscle-toxic DUX4 protein in cultured muscle cells derived from FSHD patient biopsies. Undue production of DUX4 in skeletal muscle is the cause of FSHD.

When produced in muscle tissue, DUX4 is highly toxic due to a cascade of events that eventually result in the devastating effects of FSHD. In people without FSHD, the production of DUX4 is repressed by regulatory mechanisms in the muscle cell. Facio's single goal is to develop a therapy that restores this repression as much as possible.

In December 2016, Facio announced that, together with its drug discovery partner, Evotec, it had tested about 34,000 compounds using the first-ever screening platform that enables reliable quantification of natural DUX4 protein in cultured FSHD-affected muscle cells. The more than 300 hits now identified are labeled "validated" because they came through an extensive battery of tests. For example, their repressive effect on DUX4 in Facio's screening system is reproducible and grows with increasing concentration, going as high as 100% repression. In addition, these compounds are not toxic to the muscle cells in Facio's system. "This is by far the largest recorded hit pool in the FSHD field", commented David Dasberg, Facio's Managing Director. "Even more important, our hits have been thoroughly validated and come out of a broad pool of compounds that we tested in our proprietary screening system uniquely capturing the natural biology of FSHD. We therefore have quantity as well as quality, but there is more. Perhaps the most exciting finding is that our hits span a variety of compound families with different biological modes of action. That raises the possibility of developing a portfolio of DUX4-repressing compounds, giving us multiple shots on goal."

Facio and Evotec have begun work to further characterize the hits, especially with respect to how they work on DUX4. On that basis, selected hits will undergo chemical modification to optimize their safety and efficacy. "Hits are like raw material," David Dasberg noted. "They are not directly therapeutics, and need to be refined in order to become truly viable drug candidates. That will take time, but we are definitely on our way."

Gene discovery could help prevent muscular dystrophy



March 2107

A genetic change discovered in babies born without a nose could help to prevent a debilitating and incurable form of muscular dystrophy.

Institute researchers Dr Kelan Chen, Associate Professor Marnie Blewitt, Dr James Murphy, Ms Tamara Beck and Ms Alexandra Gurzau were part of an International research team that discovered how mutations in a gene called SMCHD1 can cause a rare syndrome called Bosma Arhinia Microphthalmia Syndrome (BAMS), in which the nose fails to form during embryonic development.

SMCHD1 involvement in muscular dystrophy

The researchers also made the connection that the same gene, SMCHD1, is faulty in people with an inherited form of muscular dystrophy called Facioscapulohumeral Muscular Dystrophy Type 2 (FSHD2). This finding brings new hope for the potential prevention of FSHD2, an incurable condition that causes muscle wasting in teenagers and young adults.

Hope for future therapies

Associate Professor Blewitt said the team had already taken the first step towards developing medicines that could halt the progression of FSHD2, with the support of a grant from the FSHD Global Research Foundation.

"There is a very good opportunity to intervene before the muscle wasting commences."

"We hope that this medicine could be used to treat people who know that they carry a defective form of SMCHD1.

"FSHD2 does not commonly cause symptoms until gene carriers are teenagers or young adults, so there is a very good opportunity to intervene before the muscle wasting commences," Associate Professor Blewitt said.

<https://www.wehi.edu.au/news/gene-discovery-could-prevent-onset-muscular-dystrophy>



Where the Science is going now

Israel- Australia Collaborative Tender

FSHD Global Research Foundation has issued a tender for an Israel-Australia Collaborative Medical Research Grant. Both Israel and Australia have strong academic ties and a similar heritage in biomedical research. The Foundation would like to foster research ties between the two countries and leverage respective expertise in order to advance the field of FSHD research. The Foundation has always encouraged scientific collaboration and is excited to see what International connections this tender inspires.

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 Foundation's 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.

Centre of Excellence

We are currently exploring a joint initiative with an Australian Hospital piloting new molecular combing technology for the purpose of advancing research and diagnostics of a wide range of genetic disorders including FSHD, as well as to facilitate FSHD research in Australia. In addition to this we seek to fund a post-doctoral clinical research fellow with the mandate to carry out FSHD research. This project is in the research and development stage as we work towards finding a suitable partner to allow the project to move forward.

FSHD Educational Toolkits

The Foundation is thrilled to release a range of educational toolkits for Patients, GP's and Allied Health Professionals. Our goal is to empower our community when championing for support within the medical world.

In September 2015 FSHD Global convened a workshop of 13 leading International and Australian clinicians to develop a clinical practice guideline on FSHD. The guideline covers diagnosis and management of FSHD and sets out the standard of care that people with FSHD in Australia should expect from their care team.

From this, FSHD Global has created a set of free and accessible educational tool kits for people living with FSHD and health care providers. The "Living with FSHD" booklet covers the care that you should expect from your healthcare team, steps for diagnosis, understanding test results, guidance on communicating with health professionals and some handy tools that may help make appointments more productive. The other booklets are great resources to take to your health care providers to help them better understand the genetics of this disease, symptoms, prognosis and the effective management of FSHD.

Please visit fshdglobal.org/news/fshd-educational-toolkits/ to download your copy today or contact the Foundation at admin@fshdglobal.org to receive a hard copy.

Ways for you to Join our family!



Volunteer

Volunteer your time and skills to the Foundation. Whether it be through our internships, events or advisory boards - any help is hugely appreciated.



Workplace giving

Commit to supporting our Foundation by donating as little as \$2 each month. Simply include FSHD Global as one of your favourite charities for workplace giving. Workplace giving is an easy way for employees to contribute a small portion of their pre-tax salary to charity.



Matching

Rally together some colleagues to participate in corporate giving. Then double your company's social impact by matching their donations!



Donation Boxes

Every dollar counts, and it doesn't always have to be from your own pocket. You can help raise funds by placing a donation box in your local cafe, workplace kitchen or business place. Donation boxes are an easy way to generate awareness within your community.



End of Financial Year Donation

Donate a one-off amount to go towards finding a cure for FSHD. You can elect the particular grant and area of research you wish to support.



Boardroom luncheons

Let us liven up your boardroom! FSHD Global provides engaging and prominent speakers from our networks of scientists, business leaders and people living with FSHD to speak on topics such as the latest FSHD research, philanthropy and the gift of giving. You put on the lunch and we put on the show.



Invoice rounding

Consider appointing FSHD Global as your preferred charity for invoice rounding. When issuing invoices to your clients simply round up the amount and donate the difference to FSHD Global. This small gesture goes a long way in helping us advance treatments and finding a cure for FSHD.



Corporate partnerships

Become a Corporate Partner of our Foundation and be involved in all events throughout the entire year. Let us connect you to pioneers of industry to create prosperous relationships for all parties.



Create your own fundraiser

Host your own fundraising event and raise money on behalf of the Foundation. Whether it be a Christmas party, birthday, ladies lunch, comedy night or dinner, we encourage and appreciate all fundraising activities - no matter how small. We can provide volunteers, collateral and amazing prizes so all you need to do is send out invites.



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