FSHD Global Research Foundation

Annual Report

2016

FSHD
Global Research
Foundation Ltd

Our Mission

"The Foundation is committed to advancing global medical research, education and collaboration to improve quality of life and ultimately find a cure for Facioscapulohumeral Dystrophy. Through transparency, accountability, good governance and pure passion we aim to achieve results as quickly as possible."

FSHD
Global Research
Foundation Ltd

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FSHD Global would like to thank DibbsBarker for their ongoing support in housing our team and volunteeringtheir expertise, time and resources at no cost to the Foundation.

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Chairmans Address

Dear Friends,

On behalf of the Board of Directors, staff and volunteers at the FSHD Global Research Foundation, I would like to extend our appreciation and gratitude to the many people who have generously donated time, services and funds over the past year to assist the Foundation in realising our mission.

Facioscapulohumeral dystrophy (FSHD) is a debilitating muscle wasting disease that impacts both adults and children with devastating physical and psychological effect. The continued support from our community of donors gives a message of hope to families affected by this terrible disease. A message that something is being done to help find treatments and ultimately a cure. The positive effect of this cannot be underestimated.

Since our establishment in 2007, FSHD Global has committed $8.3 million to fund 40 medical research grants and education programs across nine countries, of which 13 are currently underway. Of these active grants; 4 characterise new genes believed to be involved with FSHD, 4 grants focus on drug discovery, 3 grants explore the natural history of FSHD looking at bone health and Infantile FSHD, 1 grant invests in medical biotech for FSHD and another grant looks to dramatically advance diagnostics of FSHD in Australia.

I am proud of the Foundation's continued transparency, accountability and innovation, leading world's best practice in the field. In a short period of time the Foundation has achieved great momentum and incredible advances, funding world leading researchers who work tirelessly to understand FSHD and find treatments and a cure.

Thank you for your ongoing support and determined effort to help us dramatically advance the pace and direction of research into FSHD. I'd like to thank you for helping make the Foundation the success it is.

FSHD is a terrible condition, but it is within our power to change the lives of future generations who are affected by FSHD.

Bill Moss AO
Chairman & Founder
FSHD Global Research Foundation

FSHD is hereditary

One in three people have no family history

It affects all the skeletal muscles in the body

There are no treatments

There is no cure

It affects one in every 7,500 Australians

100% of all tax deductible donations fund research

The symptoms are highly variable

FSHD Global is a multi award winning charity

The age of onset can range from infancy to adulthood

No government funding in Australia

Is a common form of Muscular Dystrophy

A Year in Review

The past year has seen FSHD Global successfully shift the global and now local medical landscape of FSHD, continuing to dramatically advance medical research and education into this rare disease.

In funding world's best research, we are seeing more and more Australian scientists and Research Institutions be awarded medical research grants in areas of Diagnostics and Therapeutics – a monumental achievement.

Our success has relied on; new fundraising initiatives, increased community and corporate engagement, new medical innovations and the launch of our new website, brand and voice. All of which has helped increased our pace and global footprint seeking treatments and an ultimate cure for FSHD.

2016 has been a strong year for the science; seeing a 69% increase in applications for medical research on the past year. With thanks to our communities continued support, commitment and fundraising, The Foundation was able to proudly award $1.4 million funding four new medical research grants;
Australia's first Diagnostics Grant on FSHD, launching the Inaugural Monica Ellis Children's Medical Research Grant (in Australia), a new Therapeutics Grant (in Australia) and a promising Drug Discovery Grant (in America). In addition, 2016 also saw a number of current grants successfully reach medical milestones.

In September 2015, The Foundation established a Consensus Summit welcoming 12 eminent FSHD experts from the field to co-publish Clinical Practise Guidelines on the diagnosis, treatment and clinical management of FSHD. These guidelines have since been transformed into personalised educational tool kits for; patients, GPs and allied health groups, to advance awareness and better understanding of the impact living with FSHD.

2015/2016 has been a phenomenal year for FSHD Global having won multiple awards, winning the Australian Charity Award for Outstanding Achievement and The Australian Business Excellence Award for Best Community Impact.

Remaining innovative and true to our 100% donation model, our passion, drive and recent achievements highlight to the world, FSHD Global is a small organisation doing some very big things!

Natalie Moss
Managing Director
FSHD Global Research Foundation

This award winning app is free to download at GooglePlay or at the Itunes Store. You can also access the app via our website, www.fshdglobal.org
Proudly powered by BlackCitrus

About the Foundation

The FSHD Global Research Foundation focuses on finding treatments and a cure for FSHD. In doing so, we fund world-class medical research, awareness and education. We are also committed to complete transparency and accountability in our operations.

The Foundation was established in 2007 by Bill Moss AO, a well-known Australian businessman and philanthropist who lives with FSHD. Since then, we have been addressing the chronic lack of medical funding and awareness of FSHD, both in Australia and globally.

Since 2007, the Foundation has committed $8.3 million to fund 40 ongoing medical research grants in 9 countries; the USA, Canada, the Netherlands, Italy, France, Belgium, Spain, New Zealand and Australia.

The FSHD Global Research Foundation does not operate like an average not for profit. We allocate 100% of the tax deductible donations we receive to current and future medical research grants. The Foundation’s operations are supported by non-tax deductible sponsorships. This pure charity model offers great transparency and accountability to our mission. Proud of our innovative structure, we offer all donors via the ‘FSHD – Find the Cure’ mobile app the opportunity to track exactly which research programs their money has been allocated to, with updates on the latest milestones of those programs.

With no government support the main sources of our funding for FSHD research are individuals afflicted by FSHD, their friends, supporters, as well as corporate sponsors. All funds donated are invested through careful consideration, guided by our Scientific Advisory Boards, Board of Directors and International Research Committees, ensuring FSHD Global remains a leader in discovering world’s best science.

In its truest sense, the FSHD Global Research Foundation is a small organisation doing some very big things!

What is FSHD

Facioscapulohumeral muscular Dystrophy (FSHD) is a genetic neuromuscular disease characterised by the progressive weakening and loss of skeletal muscles. FSHD places a significant burden on those affected by it and their families.

There is currently no cure and no effective treatments for FSHD.

FSHD is the most common form of muscular dystrophy affecting both adults and children. It is estimated to affect 1 in every 7,500 Australians, however, this number is probably higher as FSHD is commonly misdiagnosed or undiagnosed.

Despite the fact that FSHD affects around 3,200 people in Australia the government has never provided funding for FSHD research. Internationally the level of funding is minimal. As a result, FSHD research is estimated to lag behind research into other forms of muscular dystrophy by about twenty years.

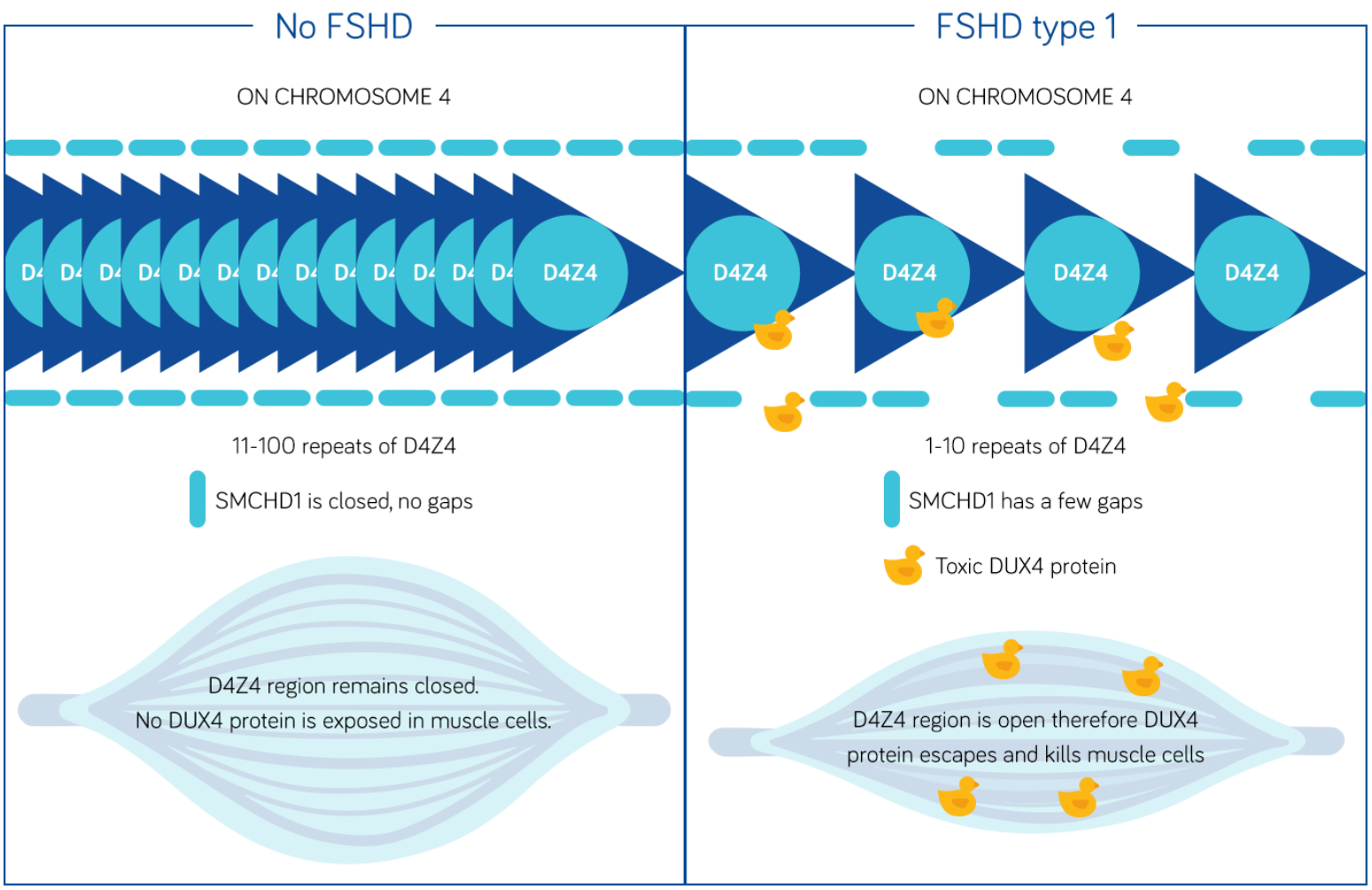
The symptoms of FSHD can manifest at any point from infancy to late adulthood, although the average age of diagnosis is around 30. Infantile FSHD is particularly severe and children affected tend to have more severe symptoms and added health complications.

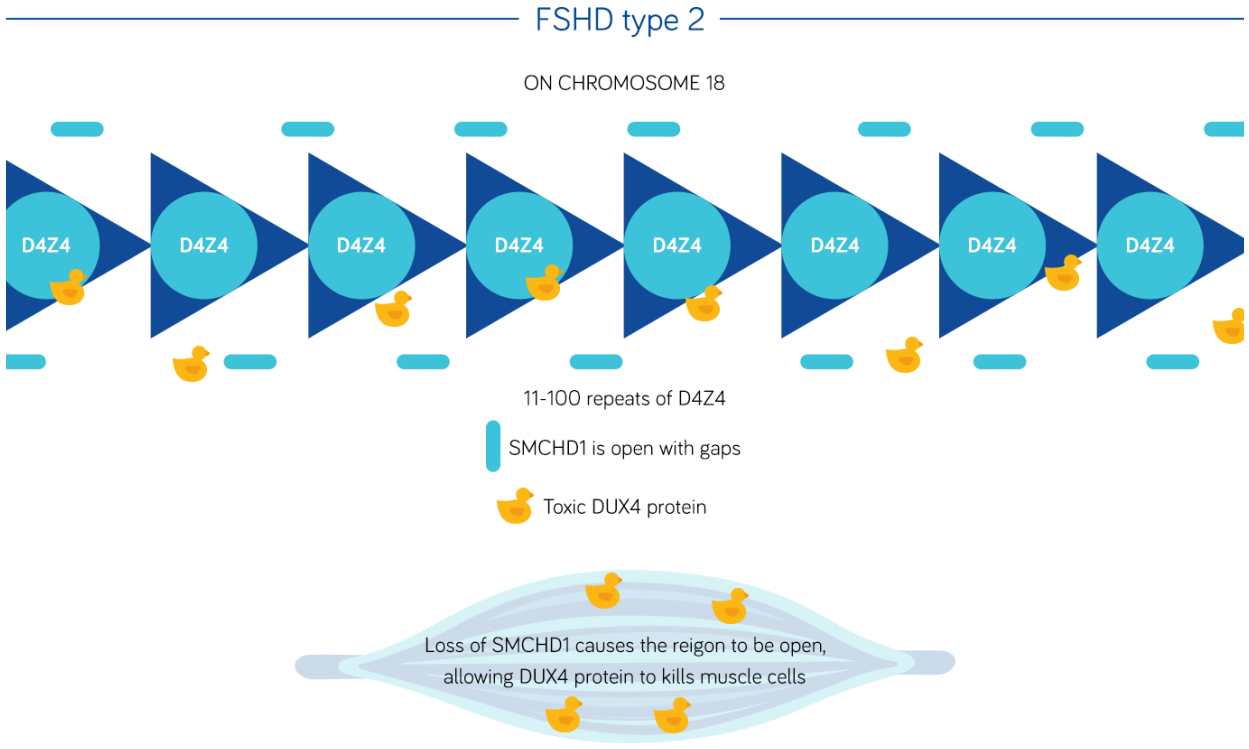
FSHD is commonly associated with progressive weakening of facial (facio), shoulder (scapulo) and upper arm muscles (humeral). However, this explanation does little justice to a disease that can rob people of their ability to walk, talk, smile or even eat. The progression is highly variable and often comes in bursts with sudden deterioration followed by periods of no change. Many people with FSHD may experience serious speech impediments caused to the weakening of facial muscles. 15% of patients have high-frequency hearing abnormalities and one in four also develop abnormalities in the blood vessels at the back of the eye with a small minority leading to vision problems.

The symptoms of FSHD are caused by the production of a protein called DUX4.

This protein plays a normal role in early foetal development, but it is highly toxic when produced in adult muscle tissue. In people without FSHD, the DUX4 gene is repressed and levels of DUX4 protein are low. For people with FSHD, the DUX4 gene is not repressed and the toxic protein damages muscle cells.

While many hereditary diseases are caused by a single genetic defect, the production of DUX4 and the development of FSHD can actually result from defects in two different chromosomes. The majority of sufferers (95%) have what is called FSHD 1 which is caused by a defect on Chromosome 4. The remainder (5%) have FSHD 2 which is caused by a defect on Chromosome 18. This scientific complexity, coupled with the shortage of research funding, makes the search for a cure even more challenging.


No FSHD
ON CHROMOSOME 4
D4Z4
11-100 repeats of D4Z4
SMCHD1 is closed, no gaps
D4Z4 region remains closed.
No DUX4 protein is exposed in muscle cells.
FSHD type 1
ON CHROMOSOME 4
D4Z4
D4Z4
D4Z4
D4Z4
1-10 repeats of D4Z4
SMCHD1 has a few gaps
Toxic DUX4 protein
D4Z4 region is open therefore DUX4
protein escapes and kills muscle cells


FSHD type 2
ON CHROMOSOME 18
D4Z4
D4Z4
D4Z4
D4Z4
D4Z4
D4Z4
D4Z4
D4Z4
11-100 repeats of D4Z4
SMCHD1 is open with gaps
Toxic DUX4 protein
Loss of SMCHD1 causes the reigon to be open,
allowing DUX4 protein to kills muscle cells

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 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 to 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 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.

Emily, Ben, Connor & Zack

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 and his wife Emily wanted to start their family, they were very aware after following Rodney and Anita's journey. The fact that Emily is a Neonatal Intensive Care Nurse 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 we have recently welcomed our 6th Grandchild and second FSHD free granddaughter.

- Marguerite Smith

Kathryn, Ted & Marguerite

Rodney, Olivia & Madeline

Meet the Team

The Foundation has built a strong culture of pure passion and determination to achieve medical advancements for FSHD as quickly as possible. We rely on the generosity, time and expertise of our community to continue to excel. We are fortunate to have an incredible support network made possible by our non-remunerated Board of Directors, Basic Science, Sub-committees, Patrons, Ambassadors, staff and volunteers who each offer vast experience in their respective fields to support our quest for a cure.

Bill Moss AO
Founder & Chairman

Natalie Moss
Managing Director

James Wakim
Deputy Chairman

Alan Watts
Head of Science

Andrew Rigney
Secretary / Director

Barry Robinson
Director

David MacKay
Director

Glenn Willis
Director

Nigel Virgo
Director

Malcolm Beville
Director

Pradnya Dugal
Director

Rohan Hardcastle
Director
(Resigned September 2016)

Beverly Baker
Alternate Director

James Harvey
Alternate Director

Natalie Livet
Acting CEO

Vanessa Kredler
Director of Philanthropy

Danielle Thompson
Director of Events

Stephanie Whittles
Administration Manager

Jason Irvine
Business Development

Olivia Hibbitt
Medical Science Liaison
(Resigned August 2016)

Beverly Baker
Chief Financial Officer
(Resigned May 2016)

Ambassadors

- Rochelle Collis 
- Kerry Johnston 
- Julie Wood 
- Tania Spagnolini 
- Carol Major 

State Branches

- Tania Spagnolini *New South Wales* 
- Les Jones *Victoria* 
- Lucy Burns *Victoria* 
- Leona Luke *Queensland* 
- Claire Anderson *Western Australia* 

Patrons

- Jamie Durie OAM 
- Luke Mangan 
- John Rasko *Patron of Science* 
- Justin Reid 
- Monica Ellis 

In loving memory of Monica Ellis

FSHD
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On March 4 2015, the Foundation lost its Founding Ambassador, enthusiastic supporter and valued friend, Monica Jean Ellis.

To say Monica is missed simply does not do justice to the amazing life she lead, the wonderful example she set and the richness she added to the lives of all who knew her.

Monica was diagnosed with Infantile FSHD when she was only five years old. Everything FSHD stole from her, her dreams and plans, she dreamt new dreams and made new plans.

First came the inability to blink properly and then she lost her smile. Later came the weakening in her shoulders, the inability to walk and then the inability to sit up. She lost the ability to chew and speech became difficult, but Monica refused to let this disease describe her.

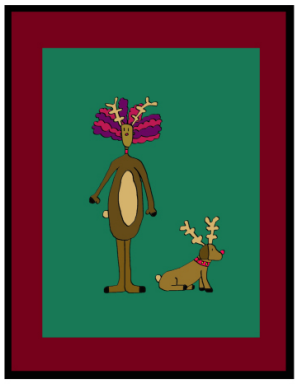
It was difficult not knowing what was wrong or what could be done, and then later, when FSHD was diagnosed, there was so little knowledge regarding how this particular dystrophy progressed or how it could be treated. Lack of knowledge also meant medical professionals sometimes made wrong assumptions because they were unaware of the condition. After all, there had been no focused research on the disease for over twenty years.

She was her own person, she knew what she wanted and she refused to let what she could not do get in the way of what she could do. As she often said "having a disability just throws a spotlight on other choices."
Monica had a wicked sense of humor. Once when being pushed through the surf in her wheelchair and being teased about sharks she quipped "Yeah, meals on wheels."

She was a talented artist and painted from her view of the world. From this perspective she produced the most wondrous fantasy artworks. She created "Silly" who became her alter ego. Silly did and explored the things that Monica simply was not physically able to.

Monica was, and remains an inspiration. FSHD was very cruel to her but she did not complain. She worked beside the Foundation looking for a treatment or a cure. Monica new any cure would be too late for her but she was tireless in her interest and willingness to assist.

Dearest Monica, your slender fingers held so many hearts. We will keep her memory always.



Silly and Molly wanted to help Santa fly the Christmas sleigh.
Even with their red noses they were not able to fly...
Anything is possible at Christmas, even to fly.

Three Wise Silly

Silly and Molly thought the best Christmas present
they could give was themselves.
How the two would get unwrapped?
Molly only had paws and Silly couldn't even reach her nose.
Only time would tell...

Silly angels in a starry Christmas night...

Community Fundraising

Ride to Raise - Sunday August 30th, 2015

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. Having witnessed the effects of FSHD first hand, Justin decided he wanted to do something to help scientists to find treatments and a cure for FSHD. Five of Justin's friends decided to support Justin in this quest as they all planned a 70 km ride from the northern part of the Gold Coast to the New South Wales border and back. The boys raised over $5,000 for medical research for FSHD - well done boys!

Arthur J. Gallagher Golf Tournament - Thursday October 29th, 2015

Arthur J. Gallagher selected FSHD Global as the benefiting charity for their Golf Tournament, November. This event was successful in raising a total of over $44,000 which has supported the Monica Ellis Children's Medical Research Grant. This is an incredible contribution from the staff and supporters of Arthur J. Gallagher.

Queensland Surf Challenge - Friday November 13th, 2015

The Wyndham Corporate Surf Challenge was held in November 2015. Accompanied by talent such as Layne Beachley, this day saw corporates suit up to compete for the win. This great day in the sun saw the Foundation raise over $5,000 from raffle tickets and auctions on the day as well as being gifted a Billboard space in the surrounds of Brisbane Airport.

Motor Traders Association Golf Tournament - Wednesday March 16th, 2016

Motor Traders Association chose FSHD Global to be a benefiting charity in their MTA Masters Golf Day 2016 held at the world class Stonecutters Ridge Golf Course. Although the day started grey, there was no rain to dampen people's spirits throughout the event. Throughout the day, MTA assisted the Foundation in raising a total of $1,000 for research.

CrossFit Muscles for Muscles - Saturday May 21st, 2016

CrossFit Bondi held a 12 hour rowing challenge at the end of May 2016. An arduous task that saw over $7,000 raised and donated to support advancements of FSHD research. They saw 100 strong athletes come together to build their muscles raising funds to help improve quality of life for those living with FSHD.

Crust Pizza

The Cronulla Crust Pizza programme "Sharks Pizzas" saw the Sharks NRL players create their own pizzas to raise funds for their nominated charity. This campaign saw $2 from every "Aussie Albert" pizza - created by Sharks player Mitch Brown - go towards FSHD Global.

Butterfly Effect

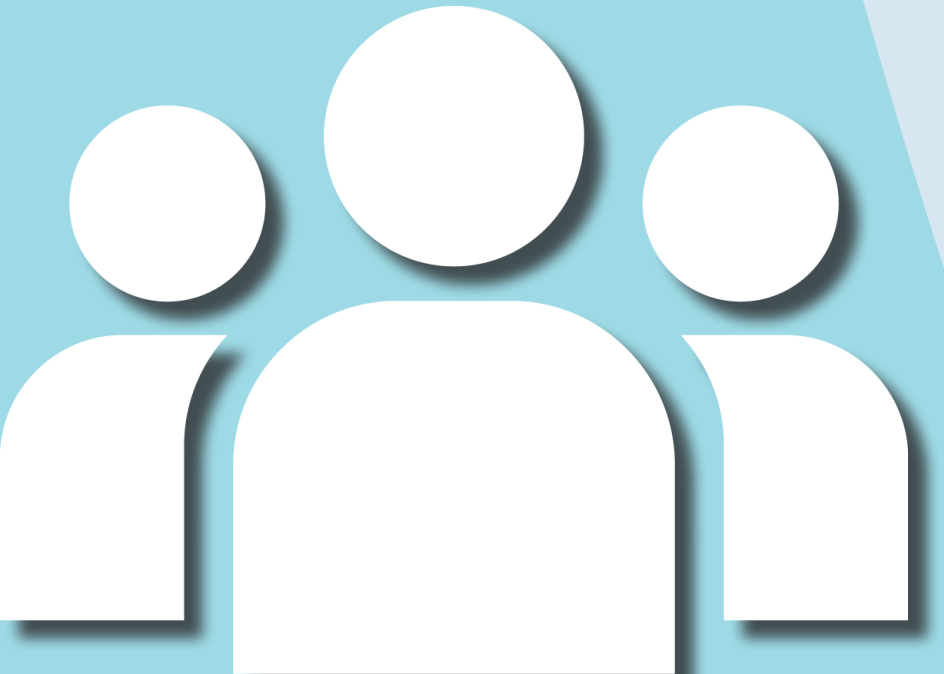
This year we created the Butterfly Effect as an innovative End of Financial Year online campaign. It encouraged our community to challenge friends and family to match their single donation, thus creating a ripple effect advancing medical research. This showed the impact and individual can have with the community raising $24,931 within several weeks.

ASX Thomson Reuters Charity Foundation

This year the Foundation was welcomed back to be a benefiting charity of the ASX Thomson Reuters Charity Foundation. We received a massive $55,838 by partaking in their charitable events and selling raffle tickets into the draw to win a Lexus IS350.

AFL Footy Tipping

Vice president of the FSHD Globals Melbourne State Branch, Les Jones ran our annual AFL Footy Tipping competition again this year raising over $1,400. This years winner will enjoy a 4 night accommodation voucher for the serene Tullah Lakeside Lodge with second and third place winning a $200 and $100 dinning voucher.



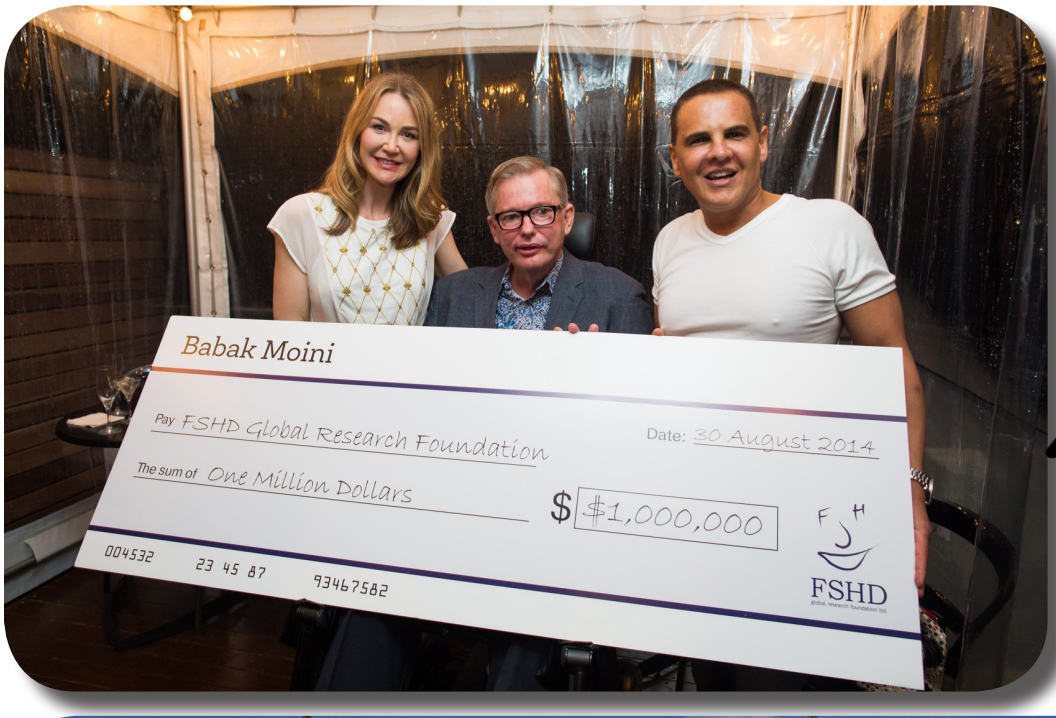
We would like to thank all of our community of supporters for all of their efforts in raising funds and awareness for FSHD. If you would like to run your own fundraiser, please do not hesitate to contact us at admin@fshdglobal.org or on (02) 8007 7037.

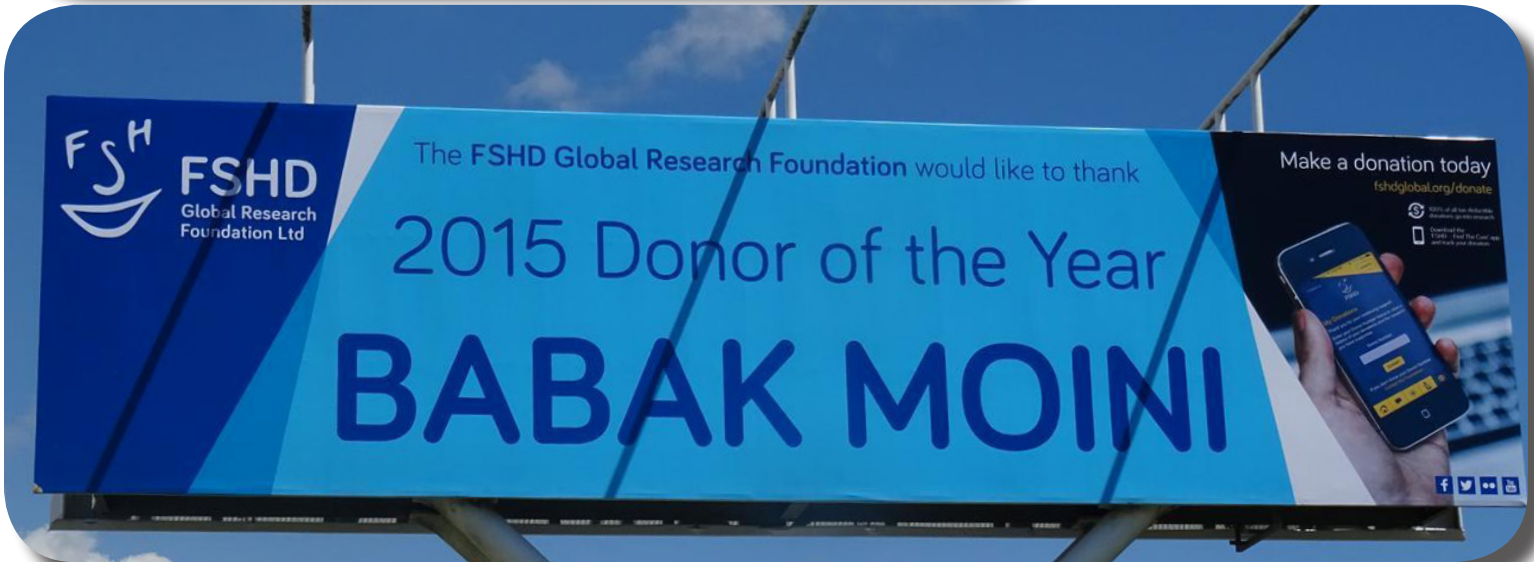
Donor of the Year

Babak Moini
2015 - 2016

Congratulations to businessman and philanthropist Babak Moini for becoming FSHD Global's Donor of the Year 2015 - 2016. We were proud to acknowledge such a remarkable member of our community. Not only has he been an incredible support to our Foundation, he inspired our 2015 EOFY "Butterfly Effect Campaign".

The Foundation worked with Babak encouraging Australians to seek transparent charities to support, challenging the culture of giving by shifting younger generations and new wealth to be creative and active in their moral commitment, giving back to society. In turn, this matching campaign successfully turned $1 million into $2 million within a two month period. This story attracted national media and enabled us to spearhead a 216% increase in donation income on 13/14 FY, raising $2.3 million, with no budget for the campaign. Babak's partner and FSHD Ambassador Rochelle Collis, has also been a huge part of the Foundation this year. She hosted the first Ladies Luxe Luncheon which was a huge success.



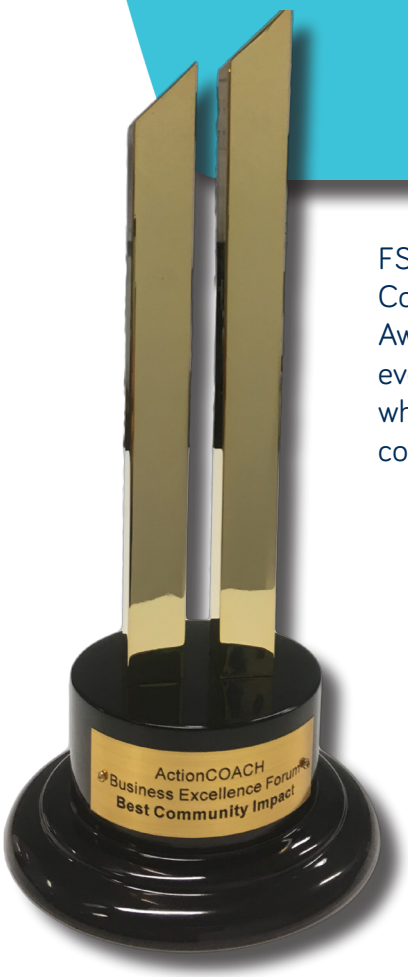


Our Proud Moments

Stem Cell Person of the Year Awards
Finalist

Our Founder and Chairman, Bill Moss AO, was a Finalist in the 'Stem Cell Person of the Year Awards' an international platform acknowledging Bill and the Foundation for establishing the world's first FSHD Human Embryonic Stem Cell Bank. On behalf of the Foundation and everyone in our community we would like to congratulate Bill on becoming one of the finalists in this global competition.

Best Community Impact
Winner



FSHD Global were proud to receive the award for Best Community Impact at the Australasian Business Excellence Awards. We were thrilled to be nominated for this award and even happier to have won. Here at FSHD Global, so much of what we do is about the impact we have both on and for the community. This award is as much for us as it is for you.

The Finances

The FSHD Global Research Foundation is a pure charity allocating 100% of all cash tax-deductible donations to current or future medical research investment, grants and education. As part of this strategy the Foundation's operating expenses are covered by other fundraising activities such as sponsorships and auctions. It is through such transparency, accountability, good governance and pure passion, we seek to find a cure for FSHD as quickly as possible.

As of the 30th of June 2016, the Foundation successfully raised over $8.3 million through the support of our community. 2015 FY saw an incredible major gift of $2 million directly impacting world's best research into FSHD.

The Annual Sydney Chocolate Ball remains to be the largest fundraising event for the disease worldwide. As the 2015 Sydney Chocolate Ball was held on the 4th of July, abnormally 2016 FY saw two Sydney Chocolate Balls.

FSHD Global continues to dramatically advance the global footprint of FSHD by increasing funds distributed to medical research, investment and education. The Foundation remains committed to fund each grant to full term (ranging from 1 – 3 years) with medical research distributions released when pre-determined scientific milestones are reached.

The Foundation has seen exceptional growth over the past two years, with an 81% increase in donations since establishment, a 52% increase in medical research grants awarded, and a 69% increase in applications for research projects. All of which has seen the Foundation successfully shift the global and local landscape of this disease, bring us closer and closer to an ultimate cure

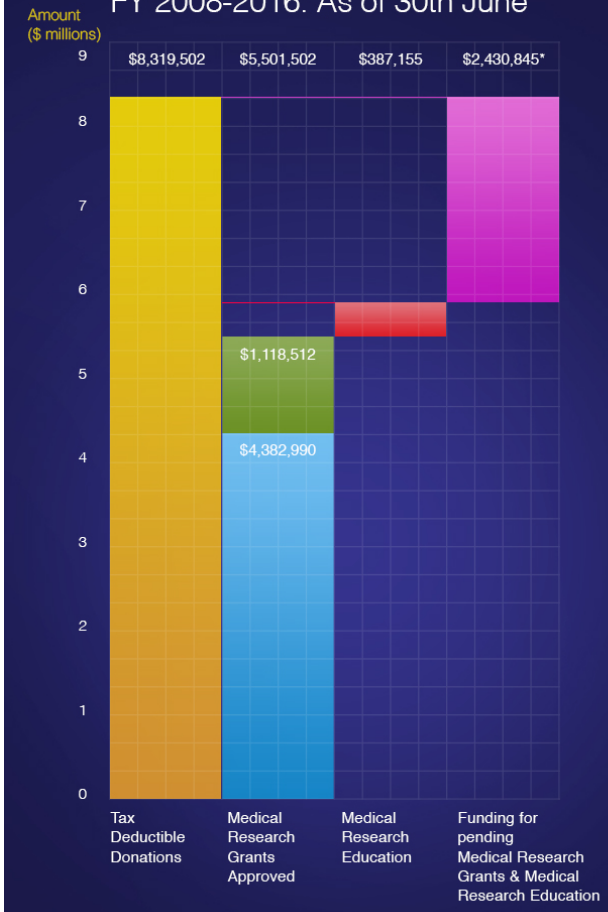
100% of all cash tax deductible donations have been allocated to current or future medical research investment, grants and education.

	2016 FY	2015 FY
INCOME		
Donations		
General Donations	$441,231	$2,481,241
Events Donations	$481,619	$150,358
Corporate Donations	$64,594	$77,900
Campaigns	$27,296	$35,461
Total Donations	$1,014,740	$2,744,960
Other Fundraising Activities*		
Net Fundraising Income	$526,390	$95,504
Other Income	$93,722	$109,852
Total Fundraising	$620,112	$205,356
TOTAL INCOME	$1,634,852	$2,950,316
EXPENDITURE		
Operating Expenses	$464,900	$365,160
MEDICAL RESEARCH		
Distributions		
Medical Research Grants	$850,648	$650,139
Medical Education	$59,724	$110,063
Total Distributions	$910,372	$760,202
MEDICAL RESEARCH INVESTMENT		
Medical Biotech Investment	$333,412	

* Other Fundraising Activities are defined as event sponsorships and ticketing, silent and live auctions, raffles, competitions, staff sponsorship and account interest.

Allocation of Tax Deductible Donations

FY 2008-2016: As of 30th June



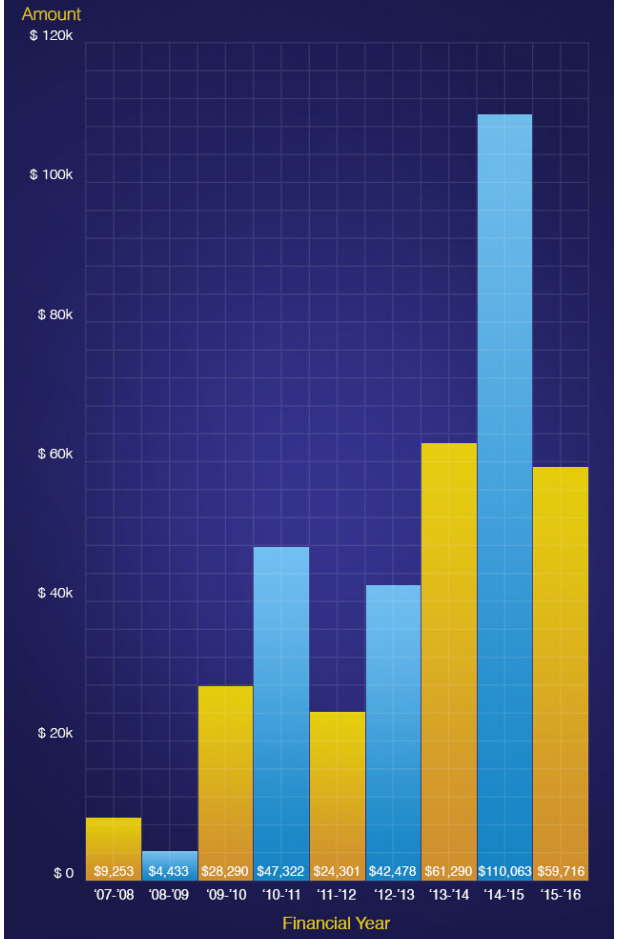
- $8,319,502 Tax Deductible Donations
- $4,382,990 Grants Funded
- $1,118,512 Grants Committed, to be Funded
- $387,155 Medical Research Education
- $2,430,845 Funding future Medical Research Grants & Medical Research Education

*In keeping with the Charter of the Foundation, we aim to invest 100% of all tax deductible donations into Medical Research Grants & Medical Research Education.

*Unaudited figures

Medical Education Funded

FY 2008-2016: As of 30th June



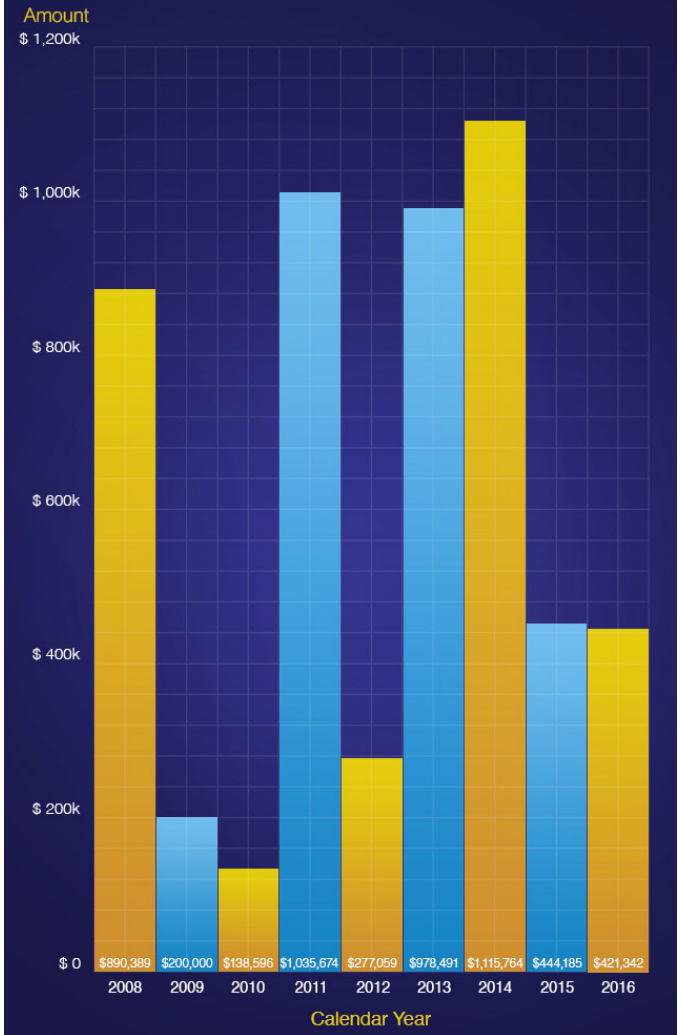
Education includes sending Australian scientists to Domestic and International conferences, sponsoring International researchers to lecture in Australia and sponsorship for worldwide scientific meetings.

$387,146 total cost of education.

*Unaudited figures

Medical Research Contracted

2008-2016: As of 30th June

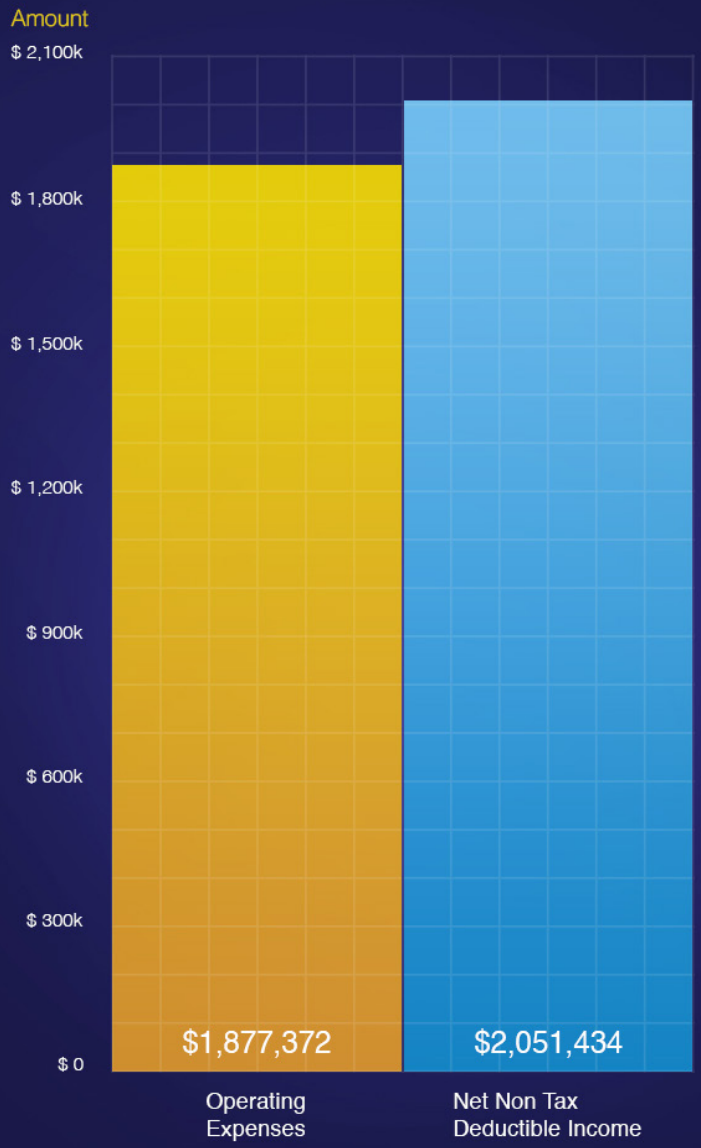


$5,501,502 contracted in total.
$4,32,990 paid as at end of 2016 FY.
$1,118,512 committed for distribution over future financial years.

*Unaudited figures

Net Non Tax Deductible Income vs Operating Expenses

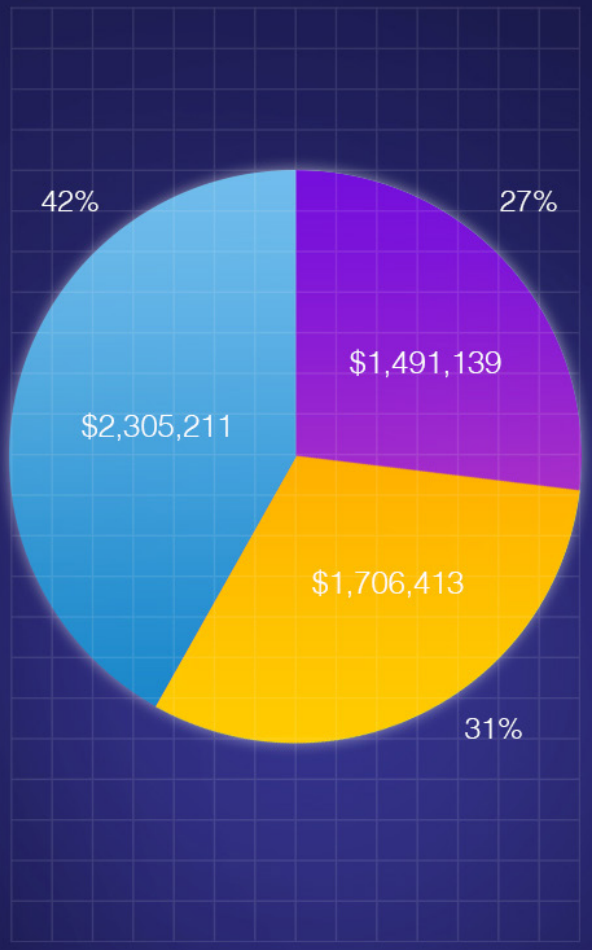
FY 2008-2016: As of 30th June



*Unaudited figures

Medical Research Allocated

FY 2008-2016: As of 30th June



*Unaudited figures

The Science

The Foundation continues to be the world leader in supporting medical research for FSHD.

In the first four months of 2016 we broke records with the Foundation allocating an unprecedented $1.3 million for medical research for diagnostics, therapeutics and Infantile FSHD with the Inaugural Monica Ellis Children's Medical Research Grant. We received a total of 22 applications for this funding, a 69% increase on last years round. All 22 were exceptional projects which would provide invaluable insight into FSHD and bring us ever closer to a cure. Each new project is a new opportunity to bring a cure to people with FSHD.

The Foundation currently has 13 exceptional research projects that are active including drug discovery, gene therapy, new diagnostics, clinical management & trials, explorations into Infantile FSHD and the natural history of FSHD.

Understanding how FSHD affects people

A world first study into bone health

This time last year the bone health of people with FSHD was an unknown. All we had were reports from our community of catastrophic fractures that often lead to people losing their mobility permanently.

Right now we are wrapping up the world's first study into bone health the results from which will help with care planning and with access to medicines that could support healthy bone development.

New insights into infantile FSHD

Research funded by the Foundation has described for the first time the characteristics of children with FSHD. This research has identified important biomarkers and continues to make significant discoveries of disease process.

New forms of FSHD

FSHD is a complex condition. In 2009 a new sub-type was described with a novel genetic cause. FSHD then became FSHD1 and FSHD2. It now appears that there might be an FSHD3. The Foundation is funding research into an entirely new, and previously undiscovered type of FSHD.

Understanding this new FSHD will have benefits for all FSHD research by furthering our understanding of how a person's genetics leads to FSHD-associated muscle wasting.

The search for a cure

The Foundation continues to direct research funds into projects that have a high potential to identify treatments and cure for FSHD.

High throughput screening for possible drugs

Michael Kyba's group have screened 35,000 compounds for agents that will reduce DUX4 (the Toxic protein that causes FSHD) and therefore reduce the effect of FSHD. This year they have progressed from high throughput screens to testing in animal models. This is a very exciting progression for this study and for people with FSHD. Moving into animal models brings this one step closer to humans.

Davide Gabellini has discovered a new genetic switch for the region associated with FSHD. This year, they have commenced a screen of compounds that might keep this switch closed.

Yi-Wen Chen has been testing two potential medicines for FSHD. Our funding has meant her work has moved from cells to animal models bringing her closer to clinical application.

Gene silencing technology

Not content with small molecules, Yi-Wen has also been given funds to investigate the potential for gene silencing oligonucleotides to reduce DUX4. Gene silencing oligonucleotides interrupt the normal process of protein production and have shown promise in other muscular dystrophies.

Yi-Wen was awarded funding to perform pre-clinical studies, with the aim of rapidly moving into clinical work.

New targets for drug discovery

This year Silvere van der Maarel along with colleagues from the USA and Australia completed a three year study into a gene called SMCHD1. Mutations in this gene have been found to cause FSHD2. SMCHD1 is involved in keeping the D4Z4 region closed, mutations

in this gene result in expression of DUX4 regardless of the number of D4Z4 repeats present. This three year research project resulted in six publications in very high impact journals, more than any other project funded by the Foundation.

The discoveries made by this team has resulted in the founding of a biotech company FacioTherapies to find treatments that could reduce the effect of FSHD. This is an unprecedented development for the study of FSHD. This new biotech will be able to drive treatments through all the clinical trial stages getting to people faster. The discoveries made by this team has resulted in the founding of a biotech company to find treatments that could reduce the effect of FSHD. This is an unprecedented development for the study of FSHD. This new biotech will be able to drive treatments through all the clinical trial stages getting to people faster.

New delivery techniques for therapeutic agents
FSHD Global provided funding for a very high performing doctoral student, Alexandra Baroni, to visit Australia from Belgium to work with the Advanced Drug Delivery Group at the University of Sydney. Alexandra has been studying polymers for drug delivery and has developed a novel polymer that is non-toxic and forms spheres around therapeutic agents. This polymer sphere essentially protects the agent from degradation allowing it to work more effectively. Alexandra will be continuing her work with this polymer by using it to encase antisense oligonucleotides for the treatment of FSHD. These agents interrupt the production of DUX4 reducing the amount in muscle cells and hopefully reducing the symptoms of FSHD. These polymers can be used for many other purposes and Alexandra has plans to develop muscle targeting technology to make her drug delivery system even better.

Clinical trial preparedness

Clinical trials are complex investigations into the ability of interventions to elicit a change in a target population. Care needs to be taken when planning clinical trials to avoid situations where the results do not reflect the true efficacy of an intervention such as not measuring the correct outcome or having a poorly specified target population.
The Foundation provided funding for a clinical trial preparedness workshop in Rochester New York in 2015. The report from the workshop was published in February 2016 and details the need for registries to make recruitment easier. The role of recently developed FSHD-specific outcome measures and the use of biomarkers is also discussed.

Building new models

One of the biggest barriers to getting a treatment into the clinic is the lack of robust animal models for FSHD. Without them, finding a cure is very difficult.

This is all about to change. The Foundation continues to fund scientists creating the next generation of animal models for FSHD.

Davide Gabellini is making animal models that contain all the genetic material involved in the human FSHD condition. Paul Gregorevic is making animal models where the expression of DUX4 can be turned off, or on, and even up and down! This fluctuating expression is characteristic of DUX4 in people. Having animal models where expression of DUX4 is as similar to people as possible will mean that results from the animals should be directly applicable to the clinic.

FSHD management statement

As well as funding medical research for future treatments and cures, the Foundation is fighting to improve the clinical care experienced by people with FSHD right now. In September 2015 the Foundation gathered a group of the world's leading FSHD clinicians to develop a consensus statement about diagnosis and management. This statement represents the standard of care that all people with FSHD in Australia should be getting and was published in the July edition of the leading journal in this field Neuromuscular Disorders. The article can be downloaded from here goo.gl/APUkkr or at our website www.fshdglobal.org

The Foundation has now developed comprehensive education materials and an extensive promotion strategy to educate health professionals and advocate for better care for people with FSHD in Australia and the rest of the world. Please contact the Foundation for this toolkit.

Science Blog

Want to know more about the research activities funded by FSHD Global? Check out our new blog with weekly articles on new research, reports on current projects and thoughts about FSHD in Australia. www.fshdglobal.org/blogs

BASIC

Basic research covers the scientific discovery side of research. From understanding what the genes involved in FSHD are doing, to how they interact with the environment to lead to progressive muscle weakness Diagnostics are the tools used to tell if someone has a certain condition. This could be a blood test, an imaging test or a genetic test.

Grant 1: Investigation into the role of FHL1, Calcineurin and NFAT in reducing muscle wasting in FSHD

Grant 2: Derivation of human embryonic stem cells to aid medical research in FSHD

Grant 3: Biomarkers in FSHD, a metabolome study in blood, urine and muscle

Grant 4: Comparing the Dnasel-Hypersensitive Chromatin Landscape at 4q35 of FSHD and Control Cells

Grant 5: Defining the mechanism controlling muscle-specific gene expression in FSHD

Grant 6: Deciphering the longdistance interactions of the D4Z4 array in control and FSHD cells

Grant 7: Molecular Genetic Basis of Facio Scapulo Humeral Dystrophy

Grant 8: Title: Dysregulated Pathways in FSHD: Recreating the FSHD Phenotype

Grant 9: Investigation of the role of FHL1 as a novel therapeutic target to reduce muscle wasting

Grant 10: Title: Study of DUX4 and DUX4c gene expression in human embryonic stem cells

Grant 11: The development of an antiDUX4 therapeutic based on chemical inhibitors of DUX4

Grant 12: Culture and Expansion of DUX4 in Human Embryonic Stem Cells Carrying FSHD

Grant 13: Bill Moss AO Fellowship for Dr Leslie Caron

Grant 14: Tissue-specific silencing of the Planar cell polarity gene FAT1 as a causal mechanism for FSHD

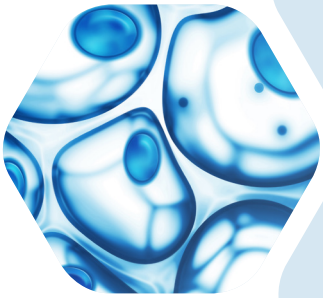
Grant 19: FSHD drug discovery based on chemical inhibitors of DUX4

Grant 20: Identification of drugs for the normalization of aberrant FSHD candidate gene expression

Grant 21: Drugtargeting of myoblast fusion as a treatment for FSHD

Grant 25: Enhancing BMP signaling to treat FSHD

Grant 28: Application of novel isoflavones in an FSHD hESC model system



Grant 30: HDL based therapy is a potential treatment for FSHD

Grant 31: Development and synthesis of AO transporter

Grant 32: A multicenter natural history and biomarkers study of infantile onset FSHD

Grant 35: The Consensus

BIOTECH INVESTMENT

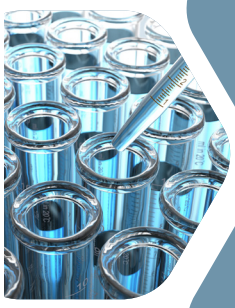
Grant 33:
Facio Therapies Biotech

Grant 29:
Training Agreement to The Netherlands

Grant 37:
The next wave of whole genome sequencing-based FSHD diagnostics, and clinical measures of progression

DIAGNOSTICS

Diagnostics are the tools used to tell if someone has a certain condition. This could be a blood test, an imaging test or a genetic test. Diagnostics are usually built around a particular aspect of a condition that most people have.



Grant 15:
DUX4 inhibition as a therapeutic strategy for FSHD

Grant 16:
Recreating the human chromosomal genetic defect responsible for FSHD in a mouse model.

Grant 17:
Evaluation of antisense strategies to suppress DUX4 expression in FSHD

Grant 22:
Increasing SMCHD1 Levels as a Therapy for FSHD1 & FSHD2

Grant 23:
Clinical Study on Possible Increased Risk of Bone Fracture

Grant 24:
Generation of Drosophila-Based Biomedical Models of FSHD

Grant 26:
Functional study of a novel candidate gene for FSH (LRIF)

Grant 27:
Preclinical Studies of Fisetin and VBP15 in Facioscapulo-humeral Muscular Dystrophy

Grant 28:
Application of novel isoflavones in an FSHD hESC model system

Grant 34:
Targeting DUX4 using gene-silencing oligonucleotides in FSHD models

Grant 36:
Effect of creatine monohydrate on strength and muscle mass in children with FSHD

THERAPEUTICS

Therapeutics is the area of creating treatments for conditions. These can be medicines or physical therapies to help improve quality of life. Research in this area may also include trying to get a better understanding of what people with FSHD go through in their lives to help develop treatments that alleviate these things.

Grant 38:
Small molecule inhibitors of DUX4 as FSHD therapeutics

Grant 39:
High throughput chemical screens for activators of SMCHD1, as potential therapeutics for FSHD

Active Grant Summaries

GRANT 16

Re-creating the human chromosomal genetic defect responsible for FSHD in a mouse model

Principal Investigator: Dr. Davide Gabellini (in collaboration with neurologist Dr Robin Fitzsimons, Sydney Medical School, The University of Sydney)
Research Institution: Division of Regenerative Medicine, San Raffaele Scientific Institute, Italy
Type: Australian and International Research Grant Collaboration
Project Goal: Creation of a mouse model that better reflects FSHD

Facioscapulohumeral muscular dystrophy (FSHD) is one of the most frequent forms of neuromuscular disease. Unfortunately, there is currently no treatment or cure for FSHD. The disease is associated with loss of genetic material (D4Z4) on chromosome 4 that is normally present in many copies toward one end of chromosome 4. FSHD patients have small D4Z4 copy number in their cells. Current animal models in mice are imperfect and the absence of an animal model that faithfully recapitulates key features of a human disease is a key step slowing down the development and test of therapeutic approaches. Unfortunately, D4Z4 repeats are present only in primates (humans, monkeys & apes) and not in laboratory animals such as mice. To solve this problem, Gabellini's group is inserting human D4Z4 and the genes that have been involved in FSHD into mouse stem cells using a technology called 'human artificial chromosomes'. This will generate animals that genetically resemble FSHD patients. This new animal model will allow scientists to study the pathways that are altered in the disease and test possible therapeutic approaches for future human clinical studies.

Dr. Gabellini's group has already transferred engineered genetic material into the intermediate mouse cells needed to finalize the production of the artificial chromosomes. These will then be transferred to mouse stem cells in order to generate "FSHD mice" and control mice which contain the human D4Z4 genetic material without the mutation responsible for FSHD.

Laboratory website: http://www.hsr.it/research/davide-gabellini/

GRANT 19

FSHD drug discovery based on chemical inhibitors of DUX4

Principle Investigator: Dr. Michael Kyba
Research Institution: University of Minnesota, USA
Type: International Research Grant (in Collaboration: Minneapolis & Boston)
Project Goal: To identify novel small molecules that may be developed for the treatment of FSHD

The Kyba group has an ongoing research program to discover chemical inhibitors of the DUX4 protein. This work is directed towards screening and studying more complex compounds than we have evaluated previously, with the objective of discovering a compound that inhibits DUX4 primary activity, as opposed to compounds that inhibit downstream pathways. These are chemically synthesized, but more similar to natural products than

to conventional drugs and have been identified through a screen of a library of complex compounds at the Broad Institute at MIT, and to develop these leads in our laboratories here in Minnesota.

The Kyba laboratory has screened a ~35,000 compound chemical library for inhibitors of DUX4, the protein that is responsible for muscle deterioration in FSHD. They have identified several clusters of related compounds that are suitable for follow up medicinal chemistry to investigate potential as new drugs for FSHD. In the last stage of this research, they have prioritized compounds for follow up, and designed chemical synthesis routes, for the top 3 compounds. In the next phase of this research, they will synthesize one of these compounds, together with a diversity of related compounds, for further study on the inhibition of DUX4, and for testing in animals.

GRANT 20

Identification of drugs for the normalization of aberrant FSHD candidate gene expression

Principle Investigator: Dr. Davide Gabellini
Research Institution: San Raffaele Scientific Institute, Italy
Type: International Research Grant (in Collaboration: Italy, Korea & Holland)
Project Goal: To identify novel drugs that may be used to treat FSHD

The underlying genetic mechanism of FSHD has been known for 20 years. In spite of this there is no effective treatment available. One of the aspects of FSHD that complicates the development of treatments for the condition is the complexity of the issues the mutation causes. The mutation is responsible for the aberrant activity of as many as 18 different genes that code for proteins localized near the FSHD region (locus) on chromosome 4. Because of this FSHD could be caused by the cumulative effects of the combination of proteins being expressed. As a result, targeting the negative effect of just one of the proteins associated with FSHD would be unlikely to address all the symptoms of the condition. This makes the development of a therapeutic approach complicated.

A treatment allowing for a general normalization of the expression of all genes that are effected in FSHD may have a much better chance to ameliorate all the FSHD symptoms.

We have identified a non-protein-coding controlling element (called DBE-T) that behaves as a master regulator of the expression of the FSHD locus. Our results strongly suggest that by controlling the activity of DBE-T it is possible to normalize the aberrant expression of all the candidate genes in FSHD muscle cells.

Our goal is to obtain a drug to block DBE-T activity. To this aim, we are characterizing the fine details of the mechanism of action of DBE-T. In parallel, we are developing high capacity screen to identify molecules that can block the aberrant activity of DBE-T. The combination of these results will allow us to identify effective therapeutics that would prevent the aberrant protein production seen in people with FSHD.

Laboratory website: http://www.hsr.it/research/davide-gabellini/

GRANT 23A & 23B

Clinical Study on Possible Increased Risk of Bone Fracture

Principle Investigator: Dr. Kathryn Wagner & Prof. Alastair Corbett
Research Institution: Kennedy Krieger Institute, Baltimore, MD, USA & Concord Hospital, Sydney, NSW, Australia
Primary Focus: Clinical Study of Bone Health in FSHD
Type: Australian and International Research Grant Collaboration

As of February 24, 2016, enrollment has been completed at the Kennedy Krieger Institute. A total of fifty participants with FSHD (27 females and 23 males) have enrolled in this clinical study at the site.

For the USA site, the age of the participants in this cross-sectional study spans from 18 to 83 years old. The allele size of the D4Z4 portion on the 4q35 chromosome in the participants ranges from 11 to 35 kb, confirming the genetic diagnosis of FSHD.

All enrollees have undergone a DEXA (dual-energy x-ray absorptiometry) scan to assess bone mineral density and lean body mass. In addition, they have had blood collected to assess bone biomarkers, and have had muscle strength and timed function testing performed.

Data from the Concord, Australian site were received on January 31, 2016. Data was collected for 52 participants, 3 of whom were confirmed to not have FSHD through gene testing. These 3 participants will be excluded. Data for the remaining 49 participants will be included in this cross-sectional study.

We have started the process of merging and cleaning the two datasets. Once a final merged dataset is available, statistical analyses will be performed to determine the prevalence of osteoporosis, fractures, abnormal bone turnover, and association with the allele size and strength testing. Descriptive statistics and bivariate associations will be explored to summarize the status of bone health among this sample of 99 participants with FSHD.

GRANT 25

Enhancing BMP signaling to treat FSHD

Principle Investigator: Dr. Paul Gregorevic
Research Institution: Baker IDI Heart and Diabetes Institute, Australia
Type: Australian Research Grant

The Team's first objective has been to develop a new mouse model in which to study how muscles are affected by FSHD, which can be used to test new therapeutic strategies. To achieve this, the Team designed a gene delivery tool that enables expression of the FSHD related gene DUX4 to be controlled in the muscles of mice. Having designed the tunable DUX4 expression system during the initial 6 months of the project, the Team has subsequently focused on defining the conditions required to reproduce intermittent DUX4 expression of varying degrees in the muscles of treated mice. This approach to controlling DUX4 expression is an important feature, as human FSHD muscles express more DUX4 than normal, but intermittently

and still at low levels. Using specific conditions, the Team is now undertaking longer-term studies to a) examine how intermittent, low-level increases in DUX4 expression contribute to the development of FSHD-like symptoms, and b) profile changes in the expression of genes as a consequence of DUX4 regulation, to identify cellular processes that could be targeted by new therapeutics. With this information in hand, the Team intends to progress to the next objective of investigating whether manipulating the activity of specific signalling mechanisms of interest is protective or restorative in mouse muscles that model FSHD.

GRANT 26

Functional study of a novel candidate gene for FSH (LRIF)

Principle Investigator: Prof Rossella Tupler
Research Institution: University of Massachusetts Medical School, USA
Type: International Research Grant

We have recently discovered a novel autosomal recessive form of FSHD, FSHD3, in an Italian family with only two affected sisters born from healthy parents. In autosomal recessive diseases one individual must carry two mutated copies of the same gene to have the FSHD. The genetic defect we found in FSHD3 abolishes a specific function of a protein and correlates with a very severe form of FSHD similar to that observed in rare sporadic cases. The protein we discovered in FSHD3 cooperates with SMCHD1. This is very important because the deficiency of SMCHD1 protein causes FSHD2, a sporadic form of FSHD associated with a mild disease. Interestingly FSHD3 and FSHD2 are clinically very different: FSHD3 display a very severe clinical FSHD, whereas a mild form of disease characterizes FSHD2 patients.

Thus, for the first time, we identify genetic elements that can dissect FSHD pathogenic mechanisms. From a molecular point of view, both proteins participate to the control of the activity of several genes located in various chromosomes, not only on chromosome 4q35, where the FSHD defect resides. It is therefore possible that the mutations in the FSHD3 gene can cause the very severe FSHD3 observed in the two sisters because its deficiency alters the activity of many genes contributing to muscle development and function. On the other hand, SMCHD1 deficiency might influence the activity of fewer genes and cause a milder disease, as observed in FSHD2 cases in comparison with FSHD3.

Overall, our discovery suggests that the basis of the large clinical variability observed in FSHD might lie on the number and type of genes that have an anomalous activity.

To study the effect of the FSHD3 mutation in muscle we have generated a mouse model carrying the mutation in the FSHD3 gene we found in the affected sisters from this new family (Specific Aim 1). We have generated cells with the ability of becoming various types of cells from cells donated by the two FSHD3 sisters and two healthy sibs. These cells are named iPSCs (induced Pluripotent Stem Cells) and we are now in the position of generating skeletal muscle cells in collaboration with Genea Biocells (Specific Aim 2). We have also started identifying genes that are anomalously expressed in muscle cells lacking the FSHD3 protein or SMCHD1 (Specific Aim 3).Our future work will be focused on the identification of the most important ways we should work on to impede the disease appearance and/or progression.

GRANT 32

A multi center natural history and biomarkers study of infantile onset FSHD

Principle Investigator: Dr. Jean Mah (in collaboration with Prof. Monique Ryan, Royal Childrens Hospital, Melbourne, Australia)
Research Institution: University of Calgary, Canada
Type: Australian and International Research Grant Collaboration

The baseline motor performance of 53 study participants with infantile FSHD havebeen studied and documented.

Consistent with previous reports, age, gender, and the size of D4Z4 repeats are associated with disease severity. Earlier onset of facial weakness in the participants was associated with a greater degree of total muscle weakness. The rate at which motor function changes will be determined by follow-up evaluations.

Speech impairment was a very common feature for participants with early onset FSHD. We identified the maximum phonation duration (MPD) as a sensitive tool for identifying speech-related issues in FSHD. We recommend affected individuals to seek professional counseling and to develop strategies to minimize voice strain and fatigue.

Remaining baseline data, including hearing, cognition, and ophthalmologic findings among the Infantile FSHD cohort will be reported once further analysis has been undertaken.

Baseline samples from some participants were found to have a higher degree of hypomethylation which resembles the combined effects of Type 1 and Type 2 mutations. Additional samples should help verify this intriguing observation.
The longitudinal study protocol received ethics approval by the University of Calgary and the Children's National Medical Center and the protocol and operational manual have been disseminated to the twelve participating CINRG sites. Some sites are still undertaking negotiations to begin recruitment however, we anticipate having the first patient enrolled by April 2016.

A potential biomarker has been identified that significantly correlates with disease severity. Results were presented at the 2015 FSHD International Research Consortium Meeting in Boston, MA, and a manuscript is being prepared for publication. The mRNA expression profiling study of blood samples collected from affected individuals with early onset FSHD is underway. Baseline data indicated unique molecular profile that can potentially be useful as biomarkers for disease progression. To extend the expression profiling study to all 48 samples collected, we submitted a grant application to the US FSH society in 2015; however it was not funded. In the longitudinal study, we will include healthy controls in the protocol to serve as comparison for the biomarker studies. We would like to allocate $30,000 CND (out of the initial installment of $45,045.45 CND from FSHDGRF) to Dr. Chen's lab to allow for the collection and storage of blood samples for biomarker discovery. Additional funding (estimated at 120K Australian dollars including microarray profiling for 48 FSHD and 10 control samples and data validation) will be needed to continue the expression profiling and biomarker studies in infantile FSHD.

BIOTECH INVESTMENT GRANT 33

Facio Therapies

FSHD Unlimited, the parent company of both Facio Therapies and Facio Intellectual Property, has published its first audited Annual Report, which covers the period of September 2014 (inception) through December 2015. Having raised €2,475,000 in equity, the Facio group of companies ended the report period with €1,428,593 in cash and cash equivalents. Expenses were mainly directed at Facio's drug discovery program towards a therapy for FSHD. The first result of that program - an initial series of candidate therapeutic compounds - is expected to become available before the end of 2016.

Together with German-based Evotec, and with the support of Leiden University Medical Center (Netherlands) and University of Rochester Medical Center (USA), Facio started its drug discovery program in June 2015. The aim is to identify small-molecule compounds able to take the DUX4 gene, which in FSHD is responsible for production of the toxic DUX4 protein, back to the repressed state seen in people without FSHD. Importantly, measuring DUX4 protein levels in patient-derived muscle cells captures the natural biological complexity of FSHD. In the second half of 2015, Facio fully focused on thoroughly characterizing FSHD-affected muscle cell lines and on defining the conditions that enable testing thousands of compounds. Facio expects to obtain an initial series of compounds with the desired effect before the end of 2016. Immediately thereafter, Facio will investigate their mode of action in FSHD and conduct extensive further testing towards so-called lead compounds suitable for development into a human therapeutic.

"Research has shown that DUX4 expression in FSHD is highly sporadic and limited to a small fraction of muscle cell nuclei", noted Kees van der Graaf, Chairman of FSHD Unlimited. "Therefore, reliably measuring DUX4 protein in FSHD-affected muscle cells to enable testing thousands of compounds in an automated process is a tremendous technical challenge. We believe that meeting this challenge represents a major advancement beyond the state of the art. Moreover, generating the initial series of candidate therapeutics would mean a major step towards a disease-modifying therapy in less than 18 months."

Facio actively pursues alignment with the FSHD community. The €2,475,000 raised in equity during the report period includes €2M from leading community members, Kees van der Graaf and Bill Moss AO, as well as €475,000 from the FSHD Global Research Foundation (Australia) and private investors. In early 2016, the FSHD Stichting (Netherlands) provided a €100,000 convertible loan.

"We are very grateful for the support from the FSHD community", commented Kees van der Graaf. "While our cash position at year-end 2015 is sufficient to fund our operations for at least the year 2016, we are pleased to note growing interest from parties both within and outside the community in financially supporting our program. Full annual report is available from: http://www.facio-therapies.com/news/ar-2014-2015

GRANT 34

Targeting DUX4 using gene-silencing oligonucleotides in FSHD models

Principle Investigator: Dr Yi Wen-Chen
Research Institution: Children's Research Unit, USA
Type: International Research Grant

Gene-silencing oligonucleotides work by preventing the production of the protein product of gene transcription. This is significant in FSHD because they can be used to stop the production of toxic proteins produced through the lack of inhibition in the D4Z4 region.

Gene silencing oligonucleotides have been studied previously in models of FSHD with some promising results. The oligonucleotides that Professor Chen will be using for this newly funded grant represent a significant advancement on these early oligonucleotides. In addition, Idera Pharmaceuticals have identified four potential oligonucleotides that effectively inhibit DUX4 mRNA transcripts in cells.

The aims of this grant are to further develop these potential therapeutics in human FSHD cells and a mouse model of FSHD.

Recent Publications

Over the past 9 years, FSHD Global has successfully seen 41 publications as a result of the world class medical research the Foundation has funded. Below are the most recent publications from 2016FY.

- Tawil R, Mah JK, Baker S, Wagner KR, Ryan MM, The Sydney Workshop Participants. Clinical practice considerations in facioscapulohumeral muscular dystrophy Sydney, Australia, 21 September 2015. Neuromuscular Disorders 26 (2016) 462 - 71

- Caron L, Kher D, Lee KL, McKernan R, Dumevska B, Hidalgo A, Li J, Yang H, Main H, Ferri G, Petek LM, Poellinger L, Miller DG, Gabellini D, Schmidt U A Human Pluripotent Stem Cell Model of Facioscapulohumeral Muscular Dystrophy-Affected Skeletal Muscles.Stem Cells Transl Med. (2016) May 23. [Epub ahead of print]

- van den Boogaard ML, Lemmers RJ, Balog J, Wohlgemuth M, Auranen M, Mitsuhashi S, van der Vliet PJ, Straasheijm KR, van den Akker RF, Kriek M, Laurense-Bik ME, Raz V, van Ostaijen-Ten Dam MM, Hansson KB, van der Kooi EL, Kiuru-Enari S, Udd B, van Tol MJ, Nishino I, Tawil R, Tapscott SJ, van Engelen BG, van der Maarel SM. Mutations in DNMT3B Modify Epigenetic Repression of the D4Z4 Repeat and the Penetrance of Facioscapulohumeral Dystrophy. American J Human Genetics. (2016) May 5;98(5):1020-9.

- Tawil R, Padberg GW, Shaw DW, van der Maarel SM, Tapscott SJ, The FSHD Workshop Participants., Clinical trial preparedness in facioscapulohumeral muscular dystrophy: Clinical, tissue, and imaging outcome measures 29–30 May 2015, Rochester, New York. Neuromuscular disorders 26 (2016) 181-186

- van den Boogaard ML, Lemmers RJ, Camaño P, van der Vliet PJ, Voermans N, van Engelen BG, Lopez de Munain A, Tapscott SJ, van der Stoep N,Tawil R, van der Maarel SM. Double SMCHD1 variants in FSHD2: the synergistic effect of two SMCHD1 variants on D4Z4 hypomethylation and disease penetrance in FSHD2. European J Human Genetics. (2016) Jan;24(1):78-85.

- Genome-wide binding and mechanistic analyses of Smchd1-mediated epigenetic regulation. Chen K, Hu J, Moore DL, Liu R, Kessans SA, Breslin K, Lucet IS, Keniry A, Leong HS, Parish CL, Hilton DJ, Lemmers RJ, van der Maarel SM, Czabotar PE, Dobson RC, Ritchie ME, Kay GF, Murphy JM, Blewitt ME. Proceedings of the National Academy of Sciences U S A. (2015) 112:E3535-44.

- Lim JW, Snider L, Yao Z, Tawil R, Van Der Maarel SM, Rigo F, Bennett CF, Filippova GN, Tapscott SJ. DICER/AGO-dependent epigenetic silencing of D4Z4 repeats enhanced by exogenous siRNA suggests mechanisms and therapies for FSHD. Human Molecular Genetics. (2015) 24(17):4817-28.

Justin's Story

Justin is my big brother.
When we were little we fought all the time, I thought he was bossy & mean, my poor mother.
I didn't really know what it meant that Justin had FSHD, no one seemed to know how it would affect him back when we were 12 & 13. He could still throw a hard punch and run away fast.

Later when I was married with 3 kids and Justin moved to Sydney from Canberra where we grew up, to get to warmer weather, I started to spend more time with him. He was about 28. We had always spent time together growing up, but now he needed me & mum.

He had just stopped driving. Well he hadn't really decided that, but he never drove in Sydney.
It was obvious the muscles in his legs were deteriorating. He showed the determination and drive that he still has today in those early days in Manly. He was still walking, but I picked him up on the days that he needed to go grocery shopping or to attend Hydrotherapy. He has lived independently all his life. I usually had 1 or 2 kids with me and Justin is a very favourite Uncle, always joking. Teaching them all the bad words and planning tricks to play on me.

Justin had dreams, he had been to University, travelled overseas, played in a band, recorded music in his own studio, and dealt every day with the slow cruel wasting of his muscles. Justin had an office chair that he used to scoot around the house from place to place. It gave him the freedom to move without being in the dreaded wheelchair. This he was avoiding at all costs.

We had obtained a manual chair, but Justin would not use it.
Around this time Justin started a counselling diploma at the Catholic University, the biggest challenge was the transport each week for tutorials. Somehow he managed.

He excelled in this course, he wanted to help others less fortunate than himself. He believed he had a natural gift for communicating and a truckload of empathy with young people. He is an incredibly wise and balanced person. He started work experience with phone based counselling, this seemed a good option for someone who could not easily get to an office. He also went and helped out at the MD Association Annual Kids camp. This was a weekend camp for kids with MD 5-14yrs. Justin would prepare video presentations with subtle messages that appealed to the kids, it was always fun. One year he used Kung Fu Panda. It was a great visual parallel for the Kids that proved anyone can achieve great things in life overcome their doubts and fears.

This is what Justin is about, even now as he fights his body, he is not giving up. He is looking at the positive side of everything. It is a spirit I don't think I have. I admire it, but it also confuses me. I am so sad for Justin who was admitted in November 2015 to the Prince of Wales Hospital, first in the Intensive Care High Dependency unit for 8 weeks and then in the infectious diseases Respiratory Ward. There have been some close calls, 3 near deaths in the first 10 days. And one coma that lasted about 5 hrs. Unfortunately Justin still remains living day to day in hospital with hope of leaving soon.

So I love him very much and respect his will to live. But I watch my mum and family visit and sit next his bed and help bath his eyes and use the suction machine to remove the mucus that is filling up his lungs. He has a feeding tube up his nose because the swallowing muscles have deteriorated so much he has been told he won't eat or drink again. To me it is a living hell, but it isn't my life. I am able to walk out of the hospital, down into the car park and drive to my house and breath in the air and thank someone that I am OK.

It takes its toll on the family though. It's just Mum and me, Dad left when the diagnosis came through, Justin aged 12, me 11. It is a tough time for a boy experiencing puberty, worse when your dad bales out and really has not much to do with you going forward.

He is not much more than a skeleton lying under a sheet, with tubes everywhere and the BPap machine and cough assist standing by. But he is thinking.He questions everything; the doctors advice, the medication, his time at the gym in rehab. He wants to go home and live independently again. He has something to say about everything that happens in his day, from the nursing staff to the air conditioning vents. His vocal chords are affected too, so he has a raspy breathless voice, but he still has a lot to say. He has asked to have someone to transcribe his thoughts. He wants to write. The first piece "How to Survive Hospital."

I admire him greatly and will support him whatever he chooses to do. He has always supported me and we are very close. We have spent many many afternoons and nights discussing things that puzzle and intrigue us: the complexities of life, the behaviours of people and what makes life important and meaningful. He always listens and then gives me very simple clear advice that is so wise, I go home feeling better.
Justin is about connection and authenticity. He is an honest man.

Justin's Story by Gaenor Meakes

In the Spotlight

The Foundation continues to achieve recognition in the media in both traditional and social media avenues in Australia and around the world. Below are just a few of this years highlights in the media.

Muscular dystrophy sufferer inspires fellow patients with positivity and sense of humour

Justin Reid shares his health struggles as he and fellow FSHD Patrons Luke Mangan and Jamie Durie OAM talk Chocolate Ball.

Jamie Durie OAM on The Morning Show

FSHD Patron Jamie Durie OAM made a guest appearance on the Morning show with Kylie Gillies and Larry Emdur as he talked about his return to Australia for the FSHD Chocolate Ball.

How one man turned a life sentence into his fortune

Bill Moss AO talks about his journey through life with FSHD and his incredible life successes. Daughter Natalie shares views of how this impacted her life and goals.

2GB 873AM

Former Macquarie Bank Executive and Founder of FSHD Global Research Foundation, Bill Moss AO

Ross Greenwood speaks to Former Macquarie Bank Executive and Founder of FSHD Global Research Foundation, Bill Moss about negative gearing, FSHD and the Parramatta Eels.

Our Achievements

Since establishment in 2007, The Foundation has raised $8.3 million, funding 40 global medical research and education grants across 9 countries.

We proudly allocate 100% of all tax deductible donations to current or future medical research investments, grants and education.

FSHD Global remains one of the largest contributors to FSHD medical research (outside the US Government).

Our Board of Directors, Science Advisory Boards, Patrons & Ambassadors receive $0 remuneration.

2016 saw a 69% increase in applications for research, with the Foundation proudly awarding $1.4 million to new grants.

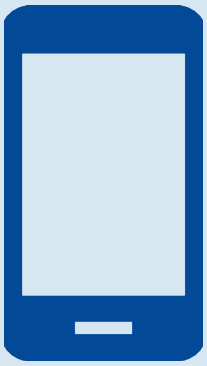
Our independent International Science Advisory Board elected Australian researchers as world's best for Diagnostics in FSHD.

The Foundation continues to actively educate and shift the Australian medical landscape to focus on this orphan disease.

Our active research focuses on; gene discovery, exploring the natural history of FSHD, Infantile FSHD, drug development & discovery, therapeutics and advancing diagnostics.

In a short period of time FSHD Global has helped advanced researchers from understanding the basic mechanisms of the disease, to actively seeking treatments and an ultimate cure - a monumental achievement.

Since publishing Clinical Practice Guidelines on the Diagnosis and Clinical Management of FSHD, The Foundation established an Australian first tool kit for patients, GP's and allied health groups to better understand the impact of living with FSHD.



Having launched a new world-class website, fshdglobal.org offers a comprehensive understanding of; the disease, research, community platforms and offers educational resources - actively engaging a global community.

FSHD Global is a multi award winning Foundation, recently winning the Australian Charity Award for Outstanding Achievement & The Australian Business Excellence Award for Best Community Impact.

The Australian first 'FSHD - Find the Cure' App is an innovative tool allowing donors to track exactly which medical research grant(s) their donation has funded, around the world.

Our Events

6th Annual Sydney Chocolate Ball

Held on the 4th of July, FSHD Global celebrated the fight for 'Independence' against this debilitating disease with an Independence Day themed gala 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. 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. It truly was a spectacular night.

Science Week and Consensus Summit

September 2015, 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 the 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. As a result of the consensus summit, the Foundation will launch an educational and advocasy tool kit for people living with FSHD.

FSHD Global Golf Tournament

Held over two days; 14th of October at St Michael's Golf Club and 3rd December at The Lakes Golf Club, the 7th Annual Golf Tournament saw 125 people come together to support our Foundation and enjoy the sunshine. Accompanied by NRL legends Andrew Johns, George Rose and Solomon Haumono, the Tournament raised a total of over $77,000. Congratulations to the winning team COzero.

HSBC World Rugby Sevens Series

FSHD Global was named an Official Charity Partner of the HSBC World Rugby Sevens Series held in Sydney, February 2016. Blessed by great weather, the weekend was filled with excitement both on and off the field. The two day event saw 73,313 attend, of which many spectators wore wild, fun and outrageous costumes. Our dedicated team of volunteers raised over $10,000 selling event programs for donations and united to create a great presence and voice for FSHD Global around the stadium.

World FSHD Day

FSHD Global initiated World FSHD Day - a day uniting all FSHD organisations around the world to bridge the gap of education across government, families & media on the effects of the disease raising greater awareness and funding opportunities worldwide. This initiative saw FSHD Global host a cocktail party that was deemed a global success. If you would like to host your own event or fundraiser for 2017 World FSHD Day, please do not hesitate to contact us.

WORLD FSHD DAY
Unite to find a cure

7th Annual 2016 Sydney Chocolate Ball

This year's Sydney Chocolate Ball was held on the 28th of May at The Star Events Centre. With fine champagne and decadent Lindt chocolate flowing all night, our guests were treated to contortionists, ballerinas in bubbles, cabaret, champagne aerialists and so so much more. Over 630 guests enjoyed Celebrity Chef and FSHD Patron Luke Mangan's exquisite chocolate inspired menu and stayed entertained by our host and FSHD Patron Jamie Durie OAM. FSHD Patron Justin Reid, commanded a standing ovation as he shared his inspirational personal journey living with FSHD. The audience were again left inspired and moved by Guest Speaker Carol Major - mother of Monica Ellis, who spoke from the heart about her loving daughter who lost her battle last year with FSHD. Among the power, passion, decadence and entertainment it was universally agreed that this was the biggest and best Sydney Chocolate Ball to date! We were successful in raising over $1.15 million dollars helping us dramatically advance our quest to find a cure.

What's Next?

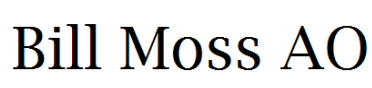
FSHD Global Annual Science Week
September 5th to 8th - Sydney, Melbourne, Brisbane & Perth.

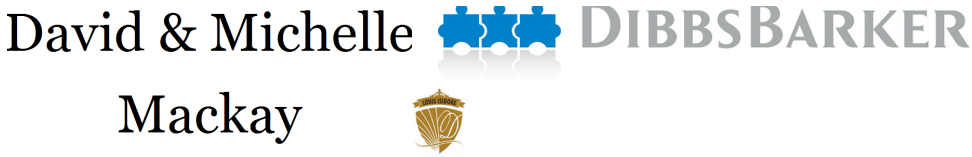
8th Annual FSHD Global Golf Tournament
September 30th, 2016 - St Michaels Golf Club, Little Bay, Sydney

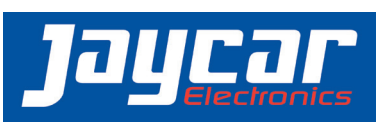
Inaugural FSHD Global Poker Tournament
October 21st, 2016 - Harbour 220, Sydney

8th Annual Sydney Chocolate Ball
June 17th, 2016 - The Star Events Centre, Sydney

A Thank You to our Sponsors





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Help Make a Difference

One-Time Gift

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

Monthly Giving

Commit to supporting our Foundation by donating as little as $2 each month. A small commitment can make a huge difference to advancements in research and takes steps closer to finding a cure for FSHD.

Volunteer

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

Corporate Giving

Rally together some colleagues to participate in Corporate Giving. Challenge your company to match donations that its employees make and make twice the difference!

Bequest

Leave a legacy that will last for generations. Gift a sum or percentage of your estate to FSHD Global to ensure the research to find a cure for FSHD continues beyond your lifetime.

Become a part of the Family

Host your own fundraising event and fundraise on behalf of the Foundation. Whether it be a birthday, ladies lunch, comedy night or dinner, we encourage and appreciate all fundraising attempts – no matter how small.

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 and offer a simple way your friends and family can get involved.

Corporate Partnership

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

For more information please do not hesitate to contact us at admin@fshdglobal.org or visit www.fshdglobal.org

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