Sanfilippo Children's Foundation

2017 OUR YEAR IN REVIEW

WHAT WE'VE ACHIEVED TOGETHER IN 2017

Our mission is to fund medical research so a cure can be found in time for children battling Sanfilippo today and those born with it tomorrow.

IN MEMORIAM

Zac James 1990-2017

In 2017 we said goodbye to a much-loved member of our Australian community. Amazingly, Zac James was 27 years old when he lost his battle to Sanfilippo Syndrome. It is unusual but not unheard of for Sanfilippo patients to live into early adulthood. Affectionately known as "The Giraffe" due to his height and gentle nature, he is sadly missed by his mum Sharon, dad Andrew and his brothers Richard, Josh and Jordan.

Zac's youngest brother, Jordan "Jordy" James also has Sanfilippo. Zac lived a full life and touched the hearts of everyone he met. Apparently, he particularly loved zombie movies. In a fitting tribute, his family hosted a zombie-themed farewell with guests dressing Halloween style to honour this beautiful young man's cheeky sense of humour and sense of fun. RIP Zac James.

We would like to dedicate this report to Zac and his family.

Cover photography:
Kiki Hopcraft from
Eklektik Photography

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Melissa Histon Photography

A message from our Executive Director

2017 (our fourth year of operation) was an extraordinary 12 months for the *Sanfilippo Children's Foundation* as we celebrated collaboration, advocacy and many a milestone.

It was a year in which, we here at the Foundation and the families and researchers we work alongside, were pushed physically, emotionally and indeed scientifically. In May, we received the exciting news that regulatory approval had been granted for the Abeona Therapeutics gene therapy clinical trial for children with Sanfilippo Type A to commence in Australia – our very first project! The first patient was treated soon afterwards and that alone was cause for huge celebration.

Throughout winter we kept busy (and active) with the SFSuper Series, which combined several physical challenges designed to push people to their "super point". We ended up with 230 participants who collectively raised $245,000 across four key events.

In September, I was joined by a fellow Sanfilippo parent at one of the SFSuper Series' events: the 100km Surf Coast Century trail run in Anglesea, Victoria. Michelle Morrice is mother to eleven-year-old Alec who has Sanfilippo Type C. As a first-time runner, Michelle embodied the spirit of this event, pushing herself to run 24kms. It was an inspiring moment seeing her cross the finish line, arms linked with her fellow team members.

Another highlight (and a surprise) came in October when we were recognised by Research Australia in its annual health and medical research awards. I was honoured to accept the Advocacy Award on behalf of the Foundation. Not only was it humbling to receive such recognition for our young Foundation, but it was a great way to raise more awareness among our scientific community. The Federal Health Minister, Hon. Greg Hunt, certainly went home that night well informed about Sanfilippo!

2017 also saw us wrap up and announce the research projects which we will fund in 2018. These include:

- the next phase (1B) of the Abeona gene therapy trial that will treat a broader group of children;
- the development of a new disease model (a zebrafish) to enable researchers to accelerate drug development;
- drug development for two subtypes of Sanfilippo Types (A & C) ;
- our second PhD student.

These latest initiatives take our project count to 12. Please turn to page 7 for more detail on these exciting projects.

There is certainly a growing sense of hope in our community, but our work in this field is far from done. We have lots to be proud of, but more work to do, and we must also acknowledge the plight of Australian children with Sanfilippo and their families. We sadly remember those children lost to Sanfilippo in 2017 such as Zac James from regional Victoria, a young man who touched so many hearts.

As we know only too well, time is not a friend for a family with Sanfilippo.

While the commencement of the Abeona clinical trial brings hope for those children still battling the disease, we must remember participation is limited and the eligibility criteria are very narrow. It's generally accepted that even if this treatment works, it's the first generation of such a therapy and will require further refinement and other interventions.

It's also important to note that this particular phase of the clinical trial is for just one of the four subtypes of Sanfilippo. So imagine for a moment the families of children with other types as they watch from the sidelines knowing this program is not for them.

And of those who are eligible, imagine for a moment the families as they await the call up. For some that call will never come, as the demand for treatment far outweighs the capacity of the trial and the selection criteria are strict. For those who do get the call and commence screening, it's an anxious time as their child goes through rigorous and highly invasive testing to see if they qualify. Some will make it through and in doing so commit to years of ongoing travel and testing, whether they see improvement in their child or not.

Others will start the screening process but not be selected for the trial because they don't meet the selection criteria for various reasons. Imagine for a moment the family whose hopes are raised only to have them dashed at the last hurdle.

For all of these children, we must continue our mission.
The impact of this first clinical trial could be enormous for the very young and the next generation of children with Sanfilippo, but that's not to say that there aren't other things which might help children live with the condition today. So what are we doing about that?

- We are driving research that will inform the global scientific community more about the disease; how it affects the body; how it progresses; and what the influences are. All of which may open new avenues for research.
- We are funding projects to build infrastructure that will help researchers to collaborate and accelerate development of therapeutics for Sanfilippo.
- We continue to develop resources for our families to inform them about the disease and associated research, and empower them to participate in the search for answers.
- We are working with our sister organisations internationally to ensure we are aligned and striving for the same goals.

Now more than ever we need you – our community – to continue to support us and surround us with energy and encouragement so that we have the strength to fight the fight against this fatal disease.

Words will never be enough to express our gratitude for your support in 2017. I sincerely thank each and every one of you.
We look forward to hearing from you in 2018.

With gratitude and hope,

Megan Donnell

WHAT A YEAR!

COMMENCED AUSTRALIAN ARM OF ABEONA CLINICAL TRIAL

HOPE

Abeona Therapeutic's clinical gene therapy trial started in 2017 when the first child was treated by an Australian clinical team at the Women's and Children's Hospital in Adelaide, South Australia.

$1.12M RAISED IN 2017

The Foundation has built a sustainable fundraising program which has demonstrated year-on-year growth with a revenue of $1.12 million in the most recent financial year.

4 NEW RESEARCH PROJECTS FUNDED

Announced funding for four new research projects including a new disease model (zebrafish) for researchers to accelerate drug development.
TURN TO PAGE 8 TO READ ABOUT THESE PROJECTS.

NEW PATRON & SCIENTIFIC ADVISORY BOARD MEMBER

We announced PROFESSOR JOHN HOPWOOD as our Foundation's official Patron and PROFESSOR IAN ALEXANDER as our Scientific Advisory Board's new Chair. We also introduced a new Scientific Advisory Board Member: DR. KIM HEMSLEY.

HIGHLIGHTS WE ACHIEVED TOGETHER IN 2017!

25 NEWSPAPER ARTICLES PUBLISHED

Continued media coverage including a cover story in the Sydney Morning Herald's Good Weekend magazine (readership of 1.2 million people) and a news item on Channel 7 Sunrise program.

WON 2017 ADVOCACY AWARD

The Foundation took out top gong with the Advocacy Award in the Research Australia annual Health and Medical Research Awards in Melbourne.

GLOBAL CLINICAL GUIDELINES

Launched a project with sister organisation, Cure Sanfilippo Foundation (USA), to develop the first ever global clinical guidelines for the management of Sanfilippo Syndrome.

2 PHD SCHOLARSHIPS

Two Australian students have been awarded supplementary scholarships to undertake ground-breaking projects into Sanfilippo.
TURN TO PAGE 8 TO READ ABOUT THEIR RESEARCH.

SFSUPERSERIES

SFSUPER SERIES RAISES $245,000 ACROSS 4 SUPER EVENTS

A record 230 participants competed (running, cycling, crossfitting and sweating it out) in our flagship fundraising event - the rebranded SFSuper Series.
TURN TO PAGE 12 TO READ ABOUT THIS EPIC EVENT.

SCIENCE UPDATE

Research Focus Areas

OUR RESEARCH PROGRAM WILL CONSIDER FUNDING PROJECTS THAT ADDRESS THE FOLLOWING:

HALT DISEASE PROGRESSION

- Enzyme replacement, gene therapy, cell therapy and other emerging therapies to stop progression of Sanfilippo
- Strategies to enhance the effectiveness of emerging therapies
- Halt disease progression early to optimise brain development

REPAIR DAMAGE

- Repair and reverse cell damage caused by Sanfilippo
- Broad application of neuroregenerative treatments, for example repurposing approved drugs
- Opportunity to collaborate with researchers working on other neurodegenerative diseases

IMPROVE QOL

- Improve quality of life of children/ adults with Sanfilippo
- Palliative care and symptom management specific to Sanfilippo

Research Principles

THE FOLLOWING PRINCIPLES UNDERPIN OUR RESEARCH STRATEGY & APPLY TO EACH FOCUS AREA.

FUND THE BEST RESEARCH
FUND RESEARCH RELEVANT TO OUR OBJECTIVES, REGARDLESS OF GEOGRAPHY, DISCIPLINE OR RESEARCHER.

FOCUS ON TRANSLATIONAL RESEARCH
DRIVE RESEARCH SHOWING TRANSLATIONAL PROMISE, TO GIVE PATIENTS EARLY ACCESS TO EMERGING THERAPIES.

ENCOURAGE INNOVATION & COLLABORATION
ENABLE INTERNATIONAL COLLABORATION. COLLABORATE WITH OTHER FUNDING BODIES TO CO-FUND PROJECTS.

BUILD NEXT GENERATION CAPABILITY
SUPPORT FUTURE RESEARCH LEADERS EARLY IN THEIR CAREERS & ATTRACT RESEARCHERS FROM OTHER DISCIPLINES.

Science Update

Gene therapy news

In May, we heard the thrilling news that the Abeona Therapeutics' gene therapy clinical trial for Sanfilippo Type A had been given the green light to go ahead by the Australian Government's Therapeutic Goods Administration (TGA). The trial site at Adelaide Women's and Children's hospital treated its first patients in 2017. Patients have also been treated in the USA, and Spain.

It is still early days, but preliminary results on a small number of participants are encouraging. Heparan sulphate, the toxic substance that builds up in children with Sanfilippo, has been shown to reduce after treatment in both the cerebrospinal fluid (CSF) and urine. Enlarged livers have reduced in size, brain scans have shown stabilisation of the brain architecture and there are signs that cognitive function is being stabilised. There have been no safety concerns, which has allowed the most recent patients to be treated at a higher dose.

In December, Abeona announced the launch of its gene therapy trial for Sanfilippo Type B with the first patient enrolled at Nationwide Children's Hospital in Ohio in the USA.

Enzyme replacement therapy (ERT) news

In September, BioMarin presented promising preliminary results from its clinical trial of enzyme replacement therapy (ERT) for Sanfilippo Type B, where the missing enzyme is delivered directly into the brain. The preliminary results showed that levels of toxic heparan sulphate, were reduced into the normal range in the three treated children. Their livers, which were enlarged at the start of the study, also decreased in size. Encouragingly, two of the three participants showed improvement or stabilisation in cognitive tests. The treatment was generally well-tolerated but there were some side effects, mostly due to the invasive way that the therapy is delivered.

Part 2 of the study is now commencing which will involve around 30 additional children to give the full picture on the safety and effectiveness of this treatment.

In other positive news, Swedish company Sobi was recently granted Fast Track Status by the FDA and given the green light to start a clinical trial of their enzyme replacement therapy for Sanfilippo Type A. It is anticipated that the trial will start sometime in 2018.

Unfortunately, in 2017 another biopharmaceutical company – Alexion – announced in July that it had cancelled its Sanfilippo Type B enzyme replacement therapy clinical trial despite previously reporting encouraging results. This is a devastating situation for those families in the USA, Spain and UK who were participating in the trial, and for the Sanfilippo community as a whole.

Research funded & co-funded by Sanfilippo Children's Foundation

ABEONA PHASE IB TRIAL OF GENE THERAPY PROGRAM

The Sanfilippo Children's Foundation has joined a collaborative agreement between nine global Foundations to conduct a phase 1b gene therapy trial for Sanfilippo Types A and B. This experimental treatment uses a virus to deliver a healthy copy of the faulty gene to the cells of children with Sanfilippo types A and B. The ongoing phase I/II trial is producing promising preliminary results. (See Gene Therapy News page 6.)

Collectively the foundations will grant USD13.875 million to Abeona with the Sanfilippo Children's Foundation contributing USD1.5 million. An Australian site will have the opportunity to be included in the phase 1b trial which will give more Australian children the chance to access this emerging treatment.

STOPSANFILIPPO FUNDACIÓN · Sanfilippo Children's Foundation · REDSANFILIPPO FUNDACIÓN · ABBY GRACE FOUNDATION Cure Sanfilippo · The Children's Medical Research Foundation, Inc. A Cure for Kirby · FONDATION SANFILIPPO · National MPS Society · Team Sanfilippo Foundation · fundación SANFILIPPO B

Translational | 2 YEAR | USD$1.5 MILLION | A | B | C | D

Chaperone therapy for Sanfilippo Type A

We are funding a one-year project led by Associate Professor Vito Ferro at the University of Queensland. The study is to develop novel, easily manufactured and affordable small molecule drugs that have potential to treat the brains of children with Sanfilippo Type A.

It will look at a type of drug called a "chaperone" which helps a protein to fold correctly and partially restore its function. An advantage of the chaperone approach is that these types of drugs are usually easy and cheap to manufacture and because they are small, can get inside the cells easily.

The research involves computer programs to design molecules with the required characteristics to act as chaperones. The most promising molecules will then be tested on cells from patients with Sanfilippo Type A grown in the lab, to determine whether they can reduce GAG (cellular waste) levels inside the cells.

THE UNIVERSITY OF QUEENSLAND AUSTRALIA

Incubator | 1 YEAR | AUD$90,000 | A | B | C | D

Creating a zebrafish model of Sanfilippo Type A

We have awarded funds to the Australian Regenerative Medicine Institute at Monash University in Melbourne for the creation of a zebrafish model of Sanfilippo Type A. The one-year project, led by zebrafish disease modelling expert Dr. Jan Kaslin, aims to produce a new tool to be used in the fight against Sanfilippo.

Zebrafish are a useful research tool because they allow quick and precise understanding of the mechanisms of disease and they can be used in the search for drugs. Zebrafish have already been used to help unlock a number of biological processes behind diseases such as muscular dystrophy.

Incubator | 1 YEAR | AUD$45,000

A B C D

The Foundation is joined by Cure Sanfilippo (USA) as a funding partner contributing a combined AUD$90,000 to this project.

Chaperone drugs for Sanfilippo Type C

This project is by Professor Alexey Pshezhetsky at the Research Institute of Sainte Justine Hospital Centre in Montreal Canada. The approach is chaperone therapy. The researchers will improve the chemistry of previously discovered chaperones and test if they can reduce the symptoms in mice with Sanfilippo Type C. They will see if the behaviour, memory and lifespan of the mice can be improved.

An advantage of the chaperone approach is that these drugs are much less expensive compared to replacement enzymes or the cost of gene therapy. Also they can often be engineered to pass through the blood brain barrier and are suitable for taking orally.

Incubator | 1 YEAR | AUD$98,000

A B C D

PhD Scholarship

Andrew Shoubridge, University of South Australia, SAHMRI

The Sanfilippo Children's Foundation is providing a supplementary scholarship to Andrew Shoubridge who is undertaking a University of Adelaide PhD project on Sanfilippo at the South Australian Health and Medical Research Institute (SAHMRI). Andrew's study will look at how neurons are damaged in Sanfilippo and how this process can be prevented.

Cognitive decline in children with Sanfilippo is not well understood. We are keen to determine whether more subtle changes in neuron structure affect how brain cells function, and whether this causes the appearance of symptoms. This will help us to understand what we need to target with treatment, and when. Andrew Shoubridge's PhD studies will be carried out under the guidance of Dr. Kim Hemsley in the Childhood Dementia Research Group, as part of the Hopwood Centre for Neurobiology at SAHMRI. The project is co-supervised by Dr. Emma Parkinson-Lawrence University of South Australia.

PhD Scholarship | 3 YEARS | AUD$15,000

A B C D

PhD Scholarship

Laura Hewson, University of Adelaide, SAHMRI

We are proud to be providing a supplementary scholarship to Laura Hewson, who is undertaking a University of Adelaide PhD project on Sanfilippo under the guidance of Dr. Louise O'Keefe. Laura's project is being co-supervised by Dr. Kim Hemsley (SAHMRI) and Prof. Robert Richards (University of Adelaide). The study will look at cellular processes which cause Sanfilippo symptoms, utilising a newly developed fruit fly model of Sanfilippo Types A and C.

These flies develop symptoms of Sanfilippo such as changes to sleep and motor function. They are a useful model because disease processes can be quickly and inexpensively studied. Laura will study the contribution cellular processes, such as inflammation in the brain, make to the symptoms of Sanfilippo.

PhD Scholarship | 2 YEARS | AUD$10,000

A B C D

RESEARCH UPDATE

RESEARCH COMPLETE

Research discovers best way to deliver gene therapy for Sanfilippo Type C

At the end of 2017 a project led by Dr Brian Bigger at the University of Manchester was completed, discovering essential information about the best way to deliver gene therapy to the brain for Sanfilippo Type C.

The researchers compared three different types of a harmless virus called AAV, which is used to deliver the missing gene in gene therapies. In mice they found that a new AAV called AAV-TT appeared to work better than those currently in gene therapy clinical trials. Using artificial brain models in the lab and sheep brains, they were also able to find the best method of injecting the gene therapy for optimal distribution throughout the brain.

The next step will be to use the knowledge gained here to complete preclinical testing of AAV delivery of the Sanfilippo Type C gene (HGSNAT), prior to initiating clinical trial, which could start in the next 2 to 5 years. This research could also help improve gene therapy for other types of Sanfilippo and other genetic diseases that affect the brain. This project was co-funded by the Sanfilippo Children's Foundation, Jonah's Just Begun in the USA and the H.A.N.D.S consortium.

Our contribution was made possible by funds raised for research by the Morrice family. Michelle and family have worked tirelessly via their Hope for Alec campaign to help make this happen. This is people-powered medicine in action and an example of the incredible advocacy and fundraising our community can do!

ONGOING RESEARCH PROJECTS

Four research projects funded by the Sanfilippo Children's Foundation were started in early 2017 and progress so far is described below. More detailed outcomes will become available once these projects are completed.

Associate Professor Andreas Schulze at the Hospital for Sick Children in Toronto is leading a two-year project to screen thousands of chemical compounds for their potential to treat Sanfilippo Types A, C and D. They have created the cells needed to screen for potential drugs and are in the process of testing the cells prior to starting to screen which is on schedule to begin in the second quarter of 2018. In parallel they are using computer modelling to identify potential drugs and this is well underway with targets already identified for further investigation.

Associate Professor Maria Fuller at SA Pathology in Adelaide is leading a project looking into how abnormal levels of lipids (fats) in the brain contribute to the symptoms of Sanfilippo Syndrome. They are also testing in mice whether a certain drug can restore the fat balance in the brain and improve symptoms. This project is running to schedule and generating exciting results.

Dr. Coy Heldermon at the University of Florida was awarded two one-year grants in early 2017 and the projects are underway and making progress towards the aims of:

- Investigating the potential of two different types of stem cells as a therapy for Sanfilippo Type B.
- Testing several different modifications to existing gene therapy technology, along with different ways of administering it, to find the best combination to move forward to clinical trial for Sanfilippo Type B.

OUR FAMILIES

Read more about the experiences of Australian families who are impacted by Sanfilippo Syndrome. Visit our website **sanfilippo.org.au**

Every family has a story. These are just some of the Australian children battling Sanfilippo Syndrome.

LUCAS

Three-year-old Lucas is best buddies with his twin Dominic. Together they play chasies, like to wrestle with Dad and love their Thomas the Tank Engine set. This little "blonde bombshell" loves the outdoors and exploring his world. Lucas keeps his mum and dad and daycare teachers on their toes. Those who know and love this bubbly little man, say he is "brave, courageous and will not be defeated!"

www.hopeforlucas.com.au

KYUSS

Five-year-old Kyuss loves motorbikes, his pet dog named "Hope", and feeding the chooks on his grandparents' farm. He charms people wherever he goes. This little rock star was actually named after the American band Kyuss, who reformed in 2010 as "Kyuss Lives". His family lives in hope. All they want is for Kyuss to live – to live a life full of joy and well beyond his current life expectancy.

www.hopeforkyuss.com.au

ISLA

Isla and Jude are brother and sister. Eight-year-old Isla likes playing with her dolls, dress ups and has a flair for fashion. She is adored by all her classmates. Six-year-old Jude likes his sandpit, playing diggers and riding his bike. Isla and Jude love their therapy dog, a black lab named Remy. Isla and Jude love nothing more than doing "zoomies" up and down the hall with Remy. Isla and Jude's parents are hopeful that one day there might be a treatment available to both their children.

JUDE

www.hopeforislaandjude.com

ALEC

Ten-year-old Alec is "one in a million" or so the saying goes. In fact his subtype of Sanfilippo (Type C) occurs in every 1 in 1.5 million. Alec loves swimming, camping and hanging out with Dad Chris and his sister Sienna. His mum Michelle says a smile from Alec makes the hardships of the day disappear. His family want him to be surrounded by understanding and love.

www.hopeforalec.com.au

SKYE

Eight-year-old Skye's Thai name is "Nongnaphat" which means Angel. This little angel loves to swim and go to the park with her big brother Luke. She enjoys swinging high and isn't afraid of the flying fox. Skye was born on Christmas Day and is a true gift to her family. Her parents worry Christmas Day will not always be a double celebration.

www.hopeforskye.com.au

PETER

Nine-year-old Peter touches the hearts of everyone he meets. Last year he even met Pope Francis in the Vatican City. Sanfilippo has stolen his words and will soon steal his ability to walk. Last year his little sister Natalie was treated for cancer (Wilms Tumour) and has made a full recovery. All Peter's family want is for him to also have a chance of treatment.

www.hopeforpeter.com.au

JACOB

Six-year-old Jacob loves dancing to the Wiggles, splashing in water, and playing with his little sister Alexis. He likes to run and jump and bursts with energy. His grandmother Debbie says Jacob wakes up beaming and brightens every room he enters. His family fears he will one day lose his beautiful spark!

www.hopeforjacob.com.au

MECKENZIE

Ten-year-old Meckenzie pursues life with joyful and reckless abandon. Her favourite thing to do is to meet and greet new people. She is truly gregarious with an infectious personality. Each year since she was diagnosed, her parents and big brother Kyle "celebrate that she hasn't lost the ability to move or talk". "It's a blessing," they say, but one they know will eventually disappear! They adore this little social butterfly beyond measure and treasure every day they have with her.

www.hopeformeckenzie.com.au

MAJOR CAMPAIGNS

Sanfilippo Children's Foundation
SFSUPERSERIES

RAISED $245,000

This year our flagship event, formerly known as the Sanfilippo1000, was rebranded and grew to combine four super events including a run, cycle, crossfit challenge, and family fun run. The SFSuper Series has been a success story for the Foundation, bringing together 230 people and raising $245,000 research dollars in 2017.

The campaign included a dedicated new website, training plans, group training sessions with coaches who donated their time, video updates and fundraising support.

There were plenty of new faces this year with a four-fold growth in participants from 2016. Fundraising included online sponsorship via a new fundraising platform as well as community events ranging from Bunnings BBQs and morning teas to group yoga classes, a trivia night, private dinners and even a barn dance.

The SFSuper Series promises to be bigger and better in 2018. We will be introducing a new event - a swim challenge.

Surf Coast Century Ultra Marathon

We had over 100 competitors at the Surf Coast Century including a record five people who ran the full 100kms (solo), five who ran 50kms and 22 relay teams of four, some of whom were new to running. Thank you to our sponsors and also our incredible corporate team from Westpac!

Big Feet and Little Feet

Bridge Run - Our family event was a hit with over 60 participants taking on Sydney Harbour Bridge in the Blackmores Bridge Run before joining us in the Sydney Botanical Gardens for a well-earned picnic. Thank you to corporate teams from Articulate and Pioneer Credit.

Gears & Beers

With perfect sunny weather in Wagga-Wagga, we had over 40 SFSuper Series cyclists take part in the Gears and Beers – Cycling, Craft Beer & Cider Festival hosted by Wollundry Rotary Club. Planning is already underway for our 2018 pilgrimage to this popular event.

Endure24

Hosted by CrossFit ManlyVale, the concept of the 24-hour CrossFit challenge 'Endure24' was to complete 24 work outs in 24 hours. In total, 32 people took part in the event which took considerable psychological and physical strength The no-sleep aspect is poignant to the parents of children with Sanfilippo, as the neurodegenerative condition causes extreme sleep disturbance. This event replicates the experience faced by families with children who require around the clock care.

There were also three amazing individuals who fundraised under the SFSuper Series banner: Melissa Thomas who did the Coolangatta Gold and Mick Collins and Greg Hawker who paddled in the Myall Classic.

Sponsors

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Service providers

SSRS Sports, Spinal & Rehab. Solutions · TRT RUNNING · tailwind nutrition · GalaBid · ASCENT · MILDREN EVENTS · CrossFit Manly Vale












CAMPAIGNS & FUNDRAISERS

SUMPTUOUS SOUNDS EVENT — $41,000

Our Sumptuous Sounds Garden Party in Sydney attracted 180 guests and generated $41,000 for the Foundation. This was our second Sumptuous Sounds event. Held at the International College of Management Sydney (ICMS), aka the "Castle on the Hill," it combined the sounds of cover band Red Carpet Ride, catering by the ICMS and beverages supplied by 4PinesBeer and BevChain. The event was managed by a committee of passionate volunteers.



A W EDWARDS CORPORATE PARTNERSHIP — $10,000

The Foundation partnered with A W Edwards (a leading, privately-owned construction and fit-out business), as its charity of choice. A W Edwards invited its employees, consultants, subcontractors and suppliers to get onboard and help make a difference with a series of small fundraisers in 2017 including onsite BBQs, networking events and a corporate team entry into the Sydney Tough Mudder.

ASX THOMSON REUTERS CHARITY FOUNDATION — $60,000

In 2017 we were once again a beneficiary of the ASX Thomson Reuters Charity Foundation, which helps disability and medical research charities by organising fundraising. The Foundation again sold art union raffle tickets with proceeds of every ticket sold going to the Foundation. In August 2017 the ASX Thomson Reuters Charity Foundation hosted a Charity Horse Race Day at Rosehill. The Foundation sold several tables to supporters with proceeds from every ticket going to Sanfilippo research.

EOFY TAX CAMPAIGN — $28,000

In 2017 we undertook a direct mail and email campaign for the End of the Financial Year (EOFY) tax appeal. We raised $28,000 in cash gifts generously given by individuals and households.

SECRET SANTA — $60,000

In 2017 our mystery benefactor, "Secret Santa", came bearing a special gift for the families of children battling Sanfilippo. Thanks to this anonymous donor, our supporters could double their donations for the month of December. Our Secret Santa committed to match all donations made as part of our Christmas appeal up to $30,000. We were thrilled to see our community reach this goal, and a total of $60,000 research dollars generated in the spirit of Christmas giving!

COMMUNITY FUNDRAISERS

The Sanfilippo Children's Foundation has continued to grow its grassroots funding model in 2017. We're grateful to our amazing community of supporters who not only fundraise, but beat the drum for the Sanfilippo cause across the country and around the world. In 2017 the Foundation was supported by 80 community fundraisers and generated almost $140,000.

As these figures show, collectively community fundraisers have a great impact and the awareness they raise is immeasurable. Once again a significant proportion of the total money raised was from initiatives run by families with children battling Sanfilippo. Our families continue to inspire their respective communities and networks to join our fundraising efforts.

In 2017 there were BBQs, school discos, trivia nights, birthday donations, workplace and team lunches, cupcake drives, head shaves, online auctions, kick-a-thons, lemonade stands, cake stalls, jars of jam and craft sales, group fun runs and a fitness challenge, and even a sustainability open house day. Pictured are some of the highlights of our community events from 2017.

We must also acknowledge the many volunteers who contribute to community fundraisers. Without the support of this optimistic and energetic group of people, we simply could not do all that we do here at the Sanfilippo Children's Foundation. It is everyday people who are helping to make an enormous difference.

If you can run a small fundraiser within your workplace, school or community, please email us at **info@sanfilippo.org.au**

NIKKI HARRIS RUNS 250KMS IN ARGENTINA

Nichole Harris from Freshwater, Sydney ran 250kms over 7 days in the Patagonia region of Argentina and raised $4,000 for the Hope for Isla and Jude campaign.

SUN RUN, MANLY, SYDNEY

A small group of seven individuals ran in the 2017 Sun Run from Dee Why to Manly, collectively raising $10,000.

CASEY CUPCAKE DRIVE

Relatives of the late Jayda Hannaford, who lost her battle to Sanfilippo in 2016, baked, iced and sold a whopping 1,800 cupcakes – raising $5,000.

TRIVIA NIGHT, BRISBANE

The Superhero-themed trivia night in Ferny Hills, Brisbane, hosted by the Morrice family raised $10,000 for the Hope for Alec campaign.

TRIVIA NIGHT, SYDNEY

The "Back to School" themed trivia night in Freshwater, Sydney raised $18,000 for the Hope for Isla and Jude campaign.

KIDS OT KICK-A-THON

Mini martial arts students from Kids OT kicked for the Sanfilippo cause at Westfield Warringah Mall, raising $5,000 for the Hope for Isla and Jude campaign.

TOUGH MUDDER FOR A TOUGH MOTHER

In Brisbane, Michelle & Chris Morrice and a team of 25 did the Tough Mudder raising $12,500. In Sydney, Fiona Higgins rallied a Tough Mudder team of 12 to raise $6,500 in honour of Megan Donnell.

HOTEL STEYNE FUN DAY

Emily Bold, Claire Harington and Sam Waller hosted a carnival-themed fun day at Manly iconic pub, The Hotel Steyne, in Sydney and raised $5,000 for the Hope for Isla and Jude campaign.

TRUSTS AND FOUNDATIONS

The Sanfilippo Children's Foundation is grateful for the significant support of the following, as well as to those who wish to remain anonymous:

THE HACKET FOUNDATION

ST GEORGE FOUNDATION

PETERSEN FAMILY FOUNDATION

ASX THOMSON REUTERS CHARITY FOUNDATION

MACQUARIE GROUP FOUNDATION

COUNT CHARITABLE FOUNDATION

LADY FAIRFAX CHARITABLE TRUST

KENNEDY CROWLEY LEGACY ENDOWMENT

THE RIPPLE FOUNDATION

THANK YOU TO OUR PRO-BONO SUPPLIERS

These professionals and suppliers have provided much-valued ongoing time, material and resources to our cause free of charge.

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PUBLIC RELATIONS
Megan and Jarrod Rose, Connect PR

SFSUPER SERIES TRAINERS:
Gary and Ana Mullins (TRT Running)
Kylie and Ben Mildren (Mildren Events)

PHOTOGRAPHERS
Ryan Clark, Clark Imagery;
Kiki Hopcraft, Eklektik Photography;
Cris Mahony, Little Love Photography;
Richard McGibbon Photography

PRINTING
Print Junction/Torzyn family;
Geoff Sly, The Printers.

THE TEAM

Sanfilippo Board Members

We thank our Board for their energy, expertise and enthusiasm

Angeline Veeneman
Patient Information & Programs

Daniel Madhavan
Investment & Networks

Mark Arnold
Strategy & Governance

Megan Donnell
Executive Director

Sanfilippo Team Members

From left to right:

Kristina Elvidge, Research & Grants Manager
Jo Bilous, SFSuper Series Event Manager;
Erene Keriakos, Bookkeeper;
Zoe Field, Business Administrator;
Ingrid Maack, Communications & Community Fundraising
Megan Donnell, Executive Director
Remy (dog), our much-loved Director of Team Well-being!

Photograph: Ryan Clark, Clark Imagery.

Our Values

Resilience

We are experts at taking knocks - from families to fundraising to scientific research projects. We dust ourselves off after each fall and continue on our mission.

Differentiation

We believe we need to do things differently - our own way - and that there is always opportunity for success off the beaten track.

Collaboration

Our cause is rare and we need to join forces with others to collectively make a difference and enable our scarce resources to go further.

Energy

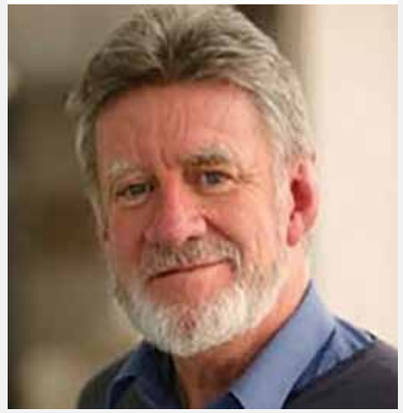
We face a marathon not a sprint - both for research we fund and the families we help. We bring strength and infinite energy to everything we do.

Determination

We are making the impossible happen and understand no matter what obstacles we face there is always a way.

Our Patron

The Sanfilippo Children's Foundation is delighted to have Professor John Hopwood in the much-honoured role of Charity Patron for the Foundation. Until his retirement, John served as Chair of our Scientific Advisory Board (SAB). John is succeeded by existing member Professor Ian Alexander. In 2017 we were thrilled to also introduce a new SAB member, Dr. Kim Hemsley, whose research focus for the past 15 years has been on Sanfilippo Syndrome.



Professor John Hopwood

AM FAA, Patron of the Sanfilippo Children's Foundation, former Head Lysosomal Disease Research Unit, Affiliate Professor at University of Adelaide, officer of the Order of Australia, Australian Academy of Science Fellow and SA Scientist of the Year.

Our SCF Board members and Scientific Advisory Board generously provide all expertise pro-bono and receive no remuneration, for which we are forever grateful.

Our Scientific Advisory Board

Professor Ian Alexander

BMedSci MBSS (Hons), PhD, FRACP (paeds), HGSACG, FAHMS is head of the Gene Therapy Research Unit, a joint initiative of Sydney Children's Hospital Network and the Children's Medical Research Institute in Sydney. He is also the Chair of the Sanfilippo Children's Foundation Scientific Advisory Board.

Dr. Kim Hemsley

BAppSci (Med. Lab. Sci), PhD, is a neuroscientist and Head of the CNS Therapeutics Group, in the Lysosomal Diseases Research Unit at the South Australian Health and Medical Research Institute (SAHMRI). She has affiliate status at the University of Adelaide.

Dr. David Ketteridge

MBBS, FRACP (Paediatrics), is a metabolic physician and General Paediatrician at the Women's and Children's Hospital in Adelaide.

Professor David Ryugo

BA Psych, PhD Psychobiology, is a Professor of Neuroscience at the Garvan Institute of Medical Research in Sydney.

Dr. Nicholas Smith

MBBS (Hon), DCH, FRACP, PhD (Cantab) currently heads the Department of Paediatric Neurology at the Women's and Children's Hospital in Adelaide.

Dr. Michel Tchan

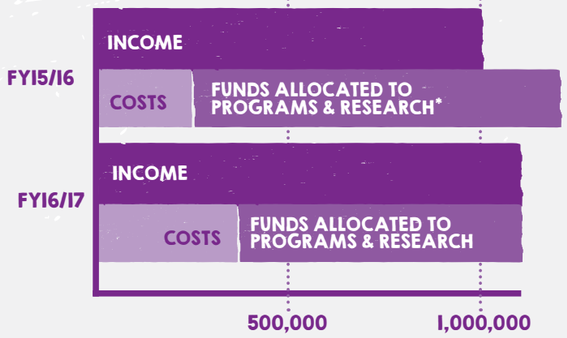
BMedSc, MBSS, PhD, FRACP, is a clinical and metabolic geneticist and Head of Department, Genetic Medicine at Westmead Hospital in Sydney. He is also the Deputy Chair of the Sanfilippo Children's Foundation Scientific Advisory Board.

THE FINANCIALS*

*For a full copy of our audited report, please contact us via email info@sanfilippo.org.au

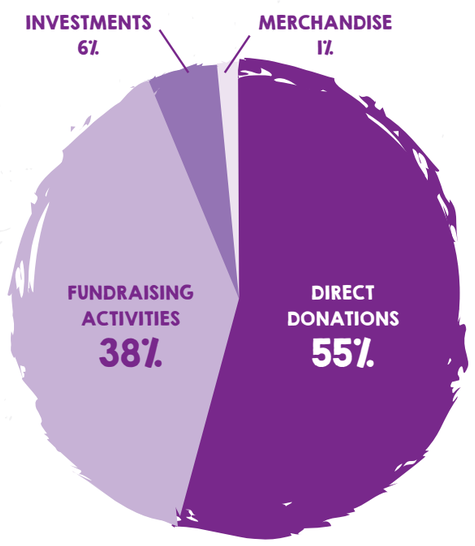
THE BASICS

	FY16/17	FY15/16
INCOME	$1.126m	$1.002m
EXPENDITURE	$394k	$286k
INVESTMENT IN RESEARCH	$732k	$909k



*Research investment in FY15/16 included some funds from previous years surplus

INCOME SOURCES



RESEARCH INVESTMENT SUMMARY

	TOTAL FUNDS INVESTED/COMMITTED
Abeona Therapeutics - Phase I/II Gene Therapy Clinical Trial	$449,330
SA Pathology - Brain lipids in Sanfilippo	$82,360
University of Florida - MPSIIIB Gene Therapy	$45,000
University of Florida - MPSIIIB Stem Cell Therapy	$45,000
University of Manchester - MPSIIIC Gene Therapy	$90,000
Sick Kids Research Inst. - High Throughput	$224,200
SAHMRI PhD - Shoubridge	$15,000
SAHMRI PhD - Hewson	$10,000
Abeona Therapeutics - Phase IB Gene Therapy Clinical Trial	$1,950,000
University of Monash - Zebrafish	$45,000
University of Qld - MPSIIIA Chaperone	$90,000
University of Montreal - MPSIIIC Chaperone	$98,000
TOTAL	$3,143,890

THE DISEASE

The following information provides a better understanding of the disease Sanfilippo (MPSIII).

A rare, life-limiting disease that is part of a wider group of pathologies

LSD — **Lysosomal Storage Disorders**
50 main diseases
1 in 5,000 births

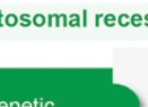
The lysosome is a very small unit in the cell and contains enzymes responsible for recycling cellular materials. A LSD is a disease where those materials don't get recycled correctly and accumulate in the cell, causing dysfunctions.

MPS — **MucoPolySaccharidosis**
7 main diseases
1 in 25,000 births

MPS are complex sugar molecules also called GAGs (GlycoAminoGlycans) naturally produced by the body and used in the building of bones, cartilage, skin, and tissues. The body continuously produces GAGs which need to be recycled.

MPSIII — **MPSIII or Sanfilippo**
4 disease types
1 in 70,000 births

Children with MPSIII lack an enzyme required to recycle the heparan sulfate, one of the many complex sugar molecules (GAGs) the body needs, resulting in extra storage of these molecules in the cells.



MPSIII Type A, B, C, D
A & B most common,
C & D rarer

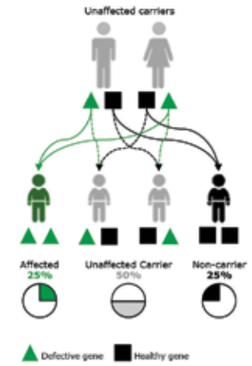
Each Sanfilippo type corresponds to a particular enzyme that is deficient. There is considerable variation in severity and life expectancy within each type.

An autosomal recessive genetic condition

Genetic
It is inherited: children get one defective gene from both their parents. With both parents carriers, there is 1 chance out of 4 for each child to inherit the disease.

Recessive
Each individual has two copies of each gene. In recessive disorders, affected individuals have two defective genes. Individuals who have one defective gene and one healthy copy are called carriers and are unaffected.

Autosomal
The defective gene is on a chromosome that is not a sex chromosome. Both males and females can be affected in autosomal disorders and either males or females can be a carrier for the disease.



A metabolic & neuro-degenerative disorder

Metabolic
Metabolism is the set of life-sustaining chemical transformations within the cells. These transformations are done through the actions of enzymes which act as catalysts; the missing enzyme in Sanfilippo results in a metabolic disorder.

Neuro-degenerative
This disorder affects primarily the cells in the Central Nervous System (CNS), resulting in brain damage. Children experience hyperactivity, sleeplessness, loss of speech and cognitive skills, mental retardation, cardiac issues, seizures, loss of mobility, dementia and finally death.

A progressive and fatal disease

Children at birth appear healthy; the first symptoms are often mild developmental delays. The disease progresses very differently from one child to the next, making it very difficult to predict.

Symptoms spectrum
Brain, sense & nerves
Nose, throat, chest & ear
Mouth & teeth
Heart
Bowel
Bones & joints
Hands & feet
Skin & hair

Thank you!

Thank you to our valued supporters, donors, sponsors, suppliers, volunteers and fundraisers. We couldn't do what we do at the Foundation without you!

THE SANFILIPPO CHILDREN'S FOUNDATION PARTNERS WITH:

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This Year in Review document has been produced by professionals who have discounted and/or donated their time and services. We would like to thank designer Noleen Lance and the team at Lindfield Print and Copy Centre.

Sanfilippo Children's Foundation

RESEARCH AUSTRALIA HEALTH & MEDICAL RESEARCH AWARDS 2017 — 2017 WINNER Advocacy Award

REGISTERED CHARITY acnc.gov.au/charityregister

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