Sanfilippo Children's Foundation

ANNUAL UPDATE 2019 - 2020

What We've Achieved Together

This photo of twins, Jobe and Tate Koistinen, was taken by Jenny Gibbs from Sugar Snaps Photography. Jobe (left) is battling Sanfilippo.

Driving research for a world without Sanfilippo Syndrome

IN MEMORIAM

Noah Bevan 2004-2019

In October 2019, we tragically lost Noah Bevan, aka 'Noah B', to Sanfilippo. He was 15 years old. Noah passed away peacefully in Brisbane surrounded by family. He was known for his megawatt smile that would light up the room. He is sadly missed by Mum Julie, Dad Craig and his adoring brothers and sisters, Justin, Adam, Nicole, Mitchell, Joshua, Ella and Jack.

We would like to dedicate this *Annual Update* to Noah and the Bevan family.

No family should face this future. We will fight on in your honour dear Noah.

OUR EXECUTIVE DIRECTOR'S MESSAGE

Horror bushfires, Coronavirus, economic turmoil... 2020 has begun with more than a few challenges. No-one could have predicted the current climate of uncertainty and anxiety we face about the state of the world. Nor could they have predicted the outpouring of generosity and philanthropy seen here in Australia. Throughout this – despite the chaos – our work here at the Sanfilippo Children's Foundation goes on and our commitment to our vision has never been stronger.

We are thrilled to have signed contracts for this year's four research projects outlined in this *Annual Update*. As always, none of these projects would have been possible without you – our incredible network of researchers, clinicians, supporters, donors, sponsors, families, volunteers and fundraisers.

Our project on the cellular process of autophagy is already underway at the University of Adelaide using the world's first fruit fly model of Sanfilippo. Autophagy is being explored for other neurodegenerative diseases such as Alzheimer's and Parkinson's. Imagine if we could identify how autophagy could be targeted to develop therapies for children with Sanfilippo.

Also underway is our project at the University of South Australia exploring the effects of Sanfilippo on the lungs. The respiratory complications of the disease are of great interest as lung infections are the leading cause of death in children with Sanfilippo, and little is known about why these complications develop.

We are particularly proud of our two-year translational project which will test several potential drug candidates in a Sanfilippo Type A mouse model. This collaboration between Flinders University and the University of Queensland aims to develop new therapies and rapidly translate them into the clinic.

While many of our projects are at Australian Institutions, this year we are also funding research in Portugal at the National Institute of Health Dr. Ricardo Jorge (INSA). This project looks toward the development of nanoparticle drug delivery with an emphasis on delivery to the brain.

We are delighted that not only are our researchers collaborating but Foundations are too! In 2020 two projects are being co-funded by our friends at Fondation Sanfilippo Suisse in Switzerland. We are so grateful.

Now more than ever we need to join forces with others to collectively make a difference, enable our scarce resources to go further and maximise our impact.

This was the intention of our first Solving Sanfilippo Symposium, in which we recently brought 30 researchers together to help. Read more about it on page 8.

To urgently solve a childhood dementia like Sanfilippo, it's going to take commitment from a range of people. It's going to take collaboration, trust, openness and innovation, the like of which we've not seen before. The strides the Sanfilippo Children's Foundation has already made won't be enough to solve Sanfilippo... new thinking, new ideas, new partnerships will be needed if we are to fulfil our purpose to drive research for a world without Sanfilippo Syndrome.

Photography: Cristina Mahony

We, of course, acknowledge the plight of Australian children with Sanfilippo and their families within this *Annual Update* and we sadly remember those children lost in 2019 such as Noah Bevan from Brisbane, a young man who touched many hearts.

Words will never be enough to express our immense gratitude for your ongoing support. I sincerely thank each and every one of you.

Megan Donnell

OUR HIGHLIGHTS

Highlights You Helped Us Achieve!

FUNDED 5 NEW RESEARCH PROJECTS

In 2019 we funded 5 research projects at institutions including the South Australian Health & Medical Research Institute (SAHMRI), Adelaide's Women's and Children's Hospital, the University of Adelaide, and at the Telethon Institute of Genetics and Medicine in Italy.

READ MORE ABOUT OUR EXCITING 2020 PROJECTS ON PAGE 4.

THANKS A MILLION

$1 MILLION MILESTONE

In 2019 we hit $1 million in funds raised via our SF Super Series, since it formally began in 2015. The Series has grown to include running, cycling, swimming and crossfitting events. Our flagship event in 2019 was a run from Newcastle to Sydney that finished on the Opera House steps. Thanks a million to all our supporters! Read more on PAGE 8 and learn what's next.

NATIONAL MEDIA COVERAGE

The Foundation and our families continue to make headlines in metro and regional news media.

Highlights included a TEDx Talk about Childhood Dementia by Megan Donnell, an article in *The Monthly* journal, an ABC Radio Focus interview and the launch of the Hope for Jobe video on Rare Disease Day with an FB reach of 81,000+ people.

GRASSROOTS EVENTS UNITE COMMUNITY

Our Sanfilippo families and supporters continue to raise research dollars across their workplaces, gyms, schools and communities with fundraisers including golf days, plant sales, cocktail parties, sock sales, a pet photoshoot, drag bingo and trivia nights, a pop-up wedding and even a marathon in Alaska! This small army of enthusiastic and dedicated people are key to helping us make an impact.

OUR RESEARCH

2020 RESEARCH PROJECTS FUNDED WITH YOUR SUPPORT

The Sanfilippo Children's Foundation has selected the following projects to fund in the year ahead. We are excited by the potential of these projects and what we might achieve together.

Targeting Autophagy in Sanfilippo

Studying Sanfilippo in fruit flies will help to increase our understanding of the disease and quickly identify potential therapeutic options.

Australian geneticist Dr. Louise O'Keefe from The University of Adelaide and the SAHMRI (South Australian Health and Medical Research Institute) will lead a team to investigate autophagy in Sanfilippo Syndrome Types A and C. Autophagy is a natural process inside cells involved in removing and recycling cell components that are not required or no longer working.

In 2018, Dr. O'Keefe and her team developed the world's first fruit fly model of Sanfilippo Type A. The team will use this model, along with a Type C fly model, to determine whether increased autophagy levels can improve Sanfilippo symptoms in flies. Individual steps in the autophagy process will also be investigated, to see which steps could be targeted therapeutically.

This project is jointly funded by the Sanfilippo Children's Foundation and the Swiss organisation, Fondation Sanfilippo Suisse.

Incubator | 1 YEAR | AUD $92,000 | A B C D

Investigating the effects of Sanfilippo on the lungs

Knowing more about how Sanfilippo damages the lungs can help to guide patient care and prevent long and distressing hospital stays due to pneumonia.

University of South Australia researchers Prof. Sandra Orgeig and Dr. Emma Parkinson-Lawrence have been awarded funds from the Sanfilippo Children's Foundation.

In this project, samples of fluid from the lungs of children with Sanfilippo and mouse models will be used to investigate how the disorder affects the lungs. Lung infections are the leading cause of death in children with Sanfilippo, and little is known about why these complications develop.

By analysing components such as heparan sulfate and immune cells in the lung fluid, this project will provide information on how lung infections arise, which can help to inform therapeutic options and clinical practice.

Incubator | 1 YEAR | AUD $33,000 | A B C D *

** Sanfilippo Type A will be studied in this project but the findings could be applied to all types of Sanfilippo.*

Evaluating potential drug therapies for Sanfilippo

Evaluating the safety and effectiveness of drug candidates in mice is an essential step before progressing to a clinical trial in children with Sanfilippo.

Dr. Adeline Lau and Assoc. Prof. Kim Hemsley from Flinders University in collaboration with Assoc. Prof. Vito Ferro from the University of Queensland have been awarded a translational grant to study potential drug candidates in a Sanfilippo Type A mouse model. The research will help to determine the safety and effectiveness of a number of key molecules, some of which have been developed using funds from the Sanfilippo Children's Foundation.

Drugs in this study aim to help stabilise the faulty Type A enzyme, or to reduce the production of heparan sulfate. Drugs will be tested individually and then the best candidate will be investigated further to see whether a combination of these drugs can have a greater effect on improving symptoms.

This project is jointly funded by the Sanfilippo Children's Foundation and the Swiss organisation, Fondation Sanfilippo Suisse.

Translational | 2 YEARS | AUD $483,000 | A B C D *

** Sanfilippo Type A will be studied in this project but the findings could be applied to all types of Sanfilippo, as well as other mucopolysaccharidoses.*

Genetic Substrate Reduction Therapy for Sanfilippo

Reducing heparan sulfate accumulation inside the brain could help to reduce symptoms and slow disease progression.

Dr. Maria Francisca Coutinho from Portugal's National Institute of Health Dr. Ricardo Jorge (INSA) has received funds from the Sanfilippo Children's Foundation to develop nanoparticles to deliver substrate reduction drugs to the brain.

Substrate Reduction Therapy (SRT) aims to decrease the production of heparan sulfate so that there is less to build up inside the body. The project will develop cutting-edge nanoparticles to deliver SRT drugs through the blood-brain barrier and into the brain.

The team will test this approach in neurons in the lab and if successful, future projects will test them in animal models.

Incubator | 1 YEAR | AUD $25,000 | A B C D

OUR RESEARCH

AN UPDATE:

Ongoing research projects

Amazing progress is being made – results from several studies are bringing us closer to finding effective treatments

Brain in a Dish update

In mid-2019 a collaborative 'Brain in a Dish' project was started to search for drugs with the potential to treat Sanfilippo Syndrome.

Skin samples from three children with Sanfilippo and three healthy children have now been donated to the study. These cells are now on their way to being transformed into brain cells in the lab.

This is done by taking skin cells from patients and reverse-engineering them into stem cells and then into neural cells, creating a 'brain in a dish' – an individualised representation of a person's brain.

Extensive work will then be carried out to understand how the Sanfilippo cells differ from their healthy counterparts before testing hundreds of drugs on them to find any that could be repurposed as a treatment.

"Testing using a patient's own cells fast-tracks the research because it enables multiple drug combinations to be trialled rapidly and without risks to the children themselves," says Assoc. Prof. Kim Hemsley who is one of the project's Chief Investigators.

The team will recruit a further two children to the study.

We thank the study participants and their families for taking part and the researchers for their hard work so far, including Assoc. Prof. Kim Hemsley (Flinders University), Dr. Nick Smith (Women's and Children's Hospital, Adelaide), Dr. Cedric Bardy (SAHMRI) and Prof. Mark Hutchinson (University of Adelaide) and their respective teams.

The project is funded with a $2million grant from the Federal Government's Medical Research Future Fund (MRFF) with a further $500,000 contribution from the Sanfilippo Children's Foundation.

"This work can not only improve the lives of those suffering with Sanfilippo but has the potential to yield findings for many more common neurological diseases." Dr. Nick Smith

"We hope this ground-breaking method of drug screening will lead to treatments." Megan Donnell

Two new PhD students

The Sanfilippo Children's Foundation has awarded top-up scholarships to Nazzmer Nazri at the University of Adelaide and Leanne Winner at Flinders University.

Nazzmer, under the supervision of Assoc. Prof. Kim Hemsley (Flinders University) and Dr. Nick Smith (Women's and Children's Hospital, Adelaide), will be looking at whether carriers of Sanfilippo are at risk of neurological problems later in life, as is the case with some other related disorders. He will study the brains of mice that are carriers of Sanfilippo Type A.

Leanne, supervised by Assoc. Prof. Kim Hemsley and Dr. Mary-Louise Rogers (Flinders University) and Dr. Marten Snel (SAHMRI), will be searching for non-invasively sourced biomarkers in the blood that could be used to measure disease progression and success of treatments. She will initially use samples from the Sanfilippo mouse model but will later verify any potential biomarkers in patient samples.

Progress for Sanfilippo Type B gene therapy

In 2017 the Sanfilippo Children's Foundation – co-funded by the Fundacja Sanfilippo (Poland) and Sanfilippo Initaitive (Germany) – awarded an incubator grant to Dr. Coy Heldermon at the University of Florida to optimise gene therapy for Sanfilippo Type B. Promising results were presented at the February 2020 WORLD conference in Florida, and the potential therapy will move forward towards clinical trial. The researchers tested different types of virus (adeno-associated virus - AAV) to deliver a healthy copy of the gene that is faulty in Sanfilippo Type B (NAGLU).

The virus had its protein shell altered to improve its ability to deliver the gene and reduce the chance of the immune system attacking it. Researchers also compared delivery of the gene therapy by injection directly into the brain and into the fluid around the brain.

With treatment at three days of age, the mice with Sanfilippo Type B had reduced storage of heparan sulfate in the brain and their hearing and survival was similar to mice without Sanfilippo. This technology has now been licensed to a gene therapy company – Lacerta Therapeutics – and they will perform toxicology studies and prepare for clinical trial. Further information on this clinical trial timeline and location(s) are not available as yet, but we will keep our community informed on updates as they are released. Meanwhile, Dr. Heldermon's group will complete studies to help decide which is the best route of delivery.

Substrate reduction therapy drug discovery update

A two-year translational project in Toronto led by Prof. Andreas Schulze was co-funded with Cure Sanfilippo Foundation (USA) in 2017. The researchers screened thousands of chemical compounds for those with the potential to reduce the production of heparan sulfate, the complex sugar that accumulates in the cells of children with Sanfilippo.

Four promising candidate drugs have been identified, all of which have a history of use for other conditions. Hopefully, after further testing, one or more of these drugs will move forward to a clinical trial in Sanfilippo patients. Another shortlist of at least 10 possible drugs is also undergoing further testing.

Continue on Page 8

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

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

LUCAS

Lucas is best buddies with his twin, Dominic. This little "blonde bombshell" loves the outdoors and keeps his mum and dad on their toes. Lucas has hearing loss but his family know he will lose much more than his hearing.

www.hopeforlucas.com.au

JOBE

Jobe is a fighter and has been since he started life prematurely. Luckily, Jobe has his fraternal twin, Tate (unaffected), by his side. Our hope is the adorable brothers have a future together.

www.hopeforjobe.com.au

KYUSS

Kyuss loves his pet dog named "Hope" and visiting his grandparents' farm. Kyuss no longer talks but this little rock star still charms everyone he meets. His family lives in hope. All they want is for Kyuss to keep smiling and live beyond his current life expectancy.

www.hopeforkyuss.com.au

MATILDA

Matilda's parents learnt her fate when she was just three months old. They are doing everything in their power to stop Sanfilippo before it robs Matilda of every precious milestone.

www.hopeformatilda.com.au

PETER

Sanfilippo may have stolen Peter's words, his ability to walk and even to eat without a tube, but it's yet to take away his smile or shining eyes. His mother, Nawal, watches old videos remembering the way he was.

www.hopeforpeter.com.au

JACOB

Jacob likes to run, jump and bursts with energy. His nan 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

SKYE

Skye's Thai name is "*Nongnaphat*" which means Angel. This little angel loves to swim, play dress-ups and go to the park with her big brother Luke. She has a vibrant personality and is always up to mischief. Her family fears one day she will lose her sense of adventure.

www.hopeforskye.com.au

ISLA & JUDE

Isla and Jude are brother and sister. They adore each other and are rarely apart. Isla and Jude love their therapy dog, a black lab named Remy. They 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.

www.hopeforislaandjude.com.au

MECKENZIE

Meckenzie is full of personality and giggles. Her favourite thing to do is to meet and greet new people. Each year since diagnosis, 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.

www.hopeformeckenzie.com.au

ALEC

Alec loves camping with his family. He loves to run but these days is unsteady on his feet. His mum, Michelle, says a hug from Alec makes the hardships of the day disappear. If only Sanfilippo would disappear.

www.hopeforalec.com.au

OUR RESEARCH (CONT.)

Zebrafish success

Researchers led by Dr. Jan Kaslin at the Australian Regenerative Medicine Institute at Monash University in Melbourne have successfully created a zebrafish model of Sanfilippo Type A, following an incubator grant awarded in 2018. Although full characterisation of this new model is ongoing, initial tests indicate that these fish will be an invaluable tool in the fight against Sanfilippo Syndrome. This zebrafish model will help to understand the disease process and to test future therapeutics.

Solving Sanfilippo Symposium

On March 3, 2020 more than 30 Australian researchers gathered in one room in Adelaide with a common goal – to come up with solutions for Sanfilippo. The day was filled with presentations of new results, ideas and plans. Lively discussion and brainstorming sessions were had, and new collaborations were formed across the seven organisations and multiple disciplines represented.

This was the first meeting of its kind the Foundation has hosted, and everybody left informed and inspired, ready to innovate and find solutions. Sharing of knowledge and collaboration on this scale is key to fast-tracking research, and this symposium is an initiative worth growing in the future.

OUR EVENTS

EXCITING NEXT STEPS AS SF SUPER SERIES HITS $1 MILLION

We are very proud to say that the SF Super Series has raised just over $1,000,000. Last year, 176 people participated across five events run by the Foundation. The total funds raised in 2019 was over $180,000.

Over five years the Series included multiple running, cycling, swimming and cross-fit events, capped off by John Burgman and his crew running from Newcastle to Sydney.

In 2020 we are building on this success and launching a range of event options for our fundraisers to stay active and continue to support Sanfilippo research.

Sanfilippo Children's Foundation SFSUPERSERIES supported by pioneer credit

50 TCS NEW YORK CITY MARATHON
TATA CONSULTANCY SERVICES

NEW EVENT: NEW YORK THIS NOVEMBER

In 2020 we continue to provide amazing bucket list experiences for our supporters who do an incredible job raising awareness and funds for research.

In November, 12 of our supporters are off to New York to run in the 50th anniversary of the New York marathon. Our very own Executive Director, Megan Donnell, is amongst the runners, as is Scientific Advisory Board member, Assoc. Prof. Kim Hemsley.

More information is on our website sanfilippo.org.au about great events you can participate in and other ways you can get involved.

OUR TEAM

SCF BOARD MEMBERS

Thank you for your energy, expertise and enthusiasm

MEGAN DONNELL
Executive Director

ANGELINE VEENEMAN

MARK ARNOLD

PAUL SCHOFF

PROFESSOR IAN ALEXANDER
(Chair of our Scientific Advisory Board)

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.

ASSOCIATE PROFESSOR KIM HEMSLEY

Assoc. Prof Kim Hemsley is a neuroscientist who is the Head, Childhood Dementia Research Group, College of Medicine and Public Health, at Flinders University, South Australia. 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.

OUR SCIENTIFIC ADVISORY BOARD

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

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.

THANK YOU TO ALL OUR FINANCIAL AND IN-KIND SPONSORS

pioneer credit · HUMPHREYS NEWSAGENCY & OFFICE SUPPLIES · THEMA · PURE RUNNING · Manly Beach Running Club · SHELLY SWIM CLUB · TRT RUNNING · TRAILBLAZERS RUN COACHING

OUR FINANCIALS

FINANCIALS

RESEARCH INVESTMENT SUMMARY

PROJECT	COMMITTED	FY
Abeona Therapeutics - Phase I/II Gene Therapy Clinical Trial	449,330	15/16
SA Pathology - Brain lipids in Sanfilippo	82,363	16/17
University of Florida - MPSIIIB Gene Therapy	45,000	16/17
University of Florida - MPSIIIB Stem Cell Therapy	45,000	16/17
University of Manchester - MPSIIIC Gene Therapy	90,000	16/17
Sick Kids Research Institute - High Throughput Screening	224,200	16/17
University of Adelaide/SAHMRI - PhD Shoubridge	15,000	16/17
University of Monash - Zebrafish	50,450	17/18
University of Qld - MPSIIIA Chaperone	90,000	17/18
University of Montreal - MPSIIIC Chaperone	98,000	17/18
University of Adelaide - PhD Hewson	10,000	17/18
University of Qld - SWIFT	10,000	18/19
Telethon Institue of Genetics & Medicine - Autophagy	45,000	18/19
Telethon Institue of Genetics & Medicine - Super Active Enzymes	90,500	18/19
Telethon Institue of Genetics & Medicine - Behavioural issues	90,000	18/19
Brain in a Dish Project	2,500,000	18/19
Flinders University - Immune System	355,000	19/20/21
Flinders University/University of Queensland - Small molecules	241,500	19/20/21
University of Adelaide - Autophagy in fruit fly model	45,786	19/20
University of South Australia - Pulmonary	32,862	19/20
INSA Portugal - SRT nanoparticles	25,000	19/20
Flinders University/University of Sydney - Biophotomodulation	11,000	19/20
University of Adelaide - PhD Nazri	10,000	19/20/21
Flinders University - PhD Winner	15,000	19/20/21/22
TOTAL	$4,670,991	

THE BASICS

	FY18/19	FY17/18
INCOME	$814.5k	$3.26m*
EXPENDITURE	$539k	$394k
INVESTMENT IN RESEARCH & PROGRAMS	$382k	$732k
FUNDS ALLOCATED TO PROGRAMS & RESEARCH	$2.8m	$1.5m

*Income in FY17/18 included extraordinary investment return of $2.72m

INCOME SOURCES

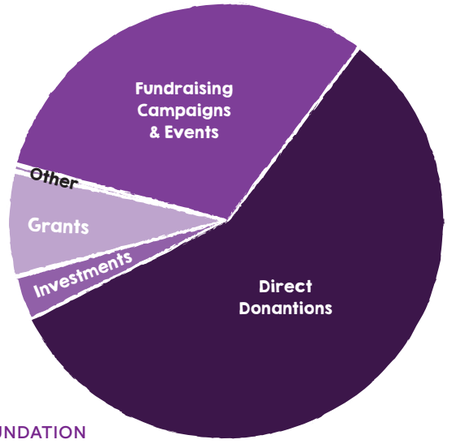
DIRECT DONATIONS $253,856

FUNDRAISING CAMPAIGNS & EVENTS $466,693

GRANTS $27,045

INVESTMENTS $63,613

OTHER $3,291



THANK YOU

TRUSTS & 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:

- **ASX REFINITIV CHARITY FOUNDATION**
- **PETERSEN FAMILY FOUNDATION**
- **ST GEORGE FOUNDATION**
- **PERPETUAL IMPACT PHILANTHROPY PROGRAM**
- **LADY FAIRFAX CHARITABLE TRUST**
- **RIPPLE FOUNDATION**

SUPPLIERS

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

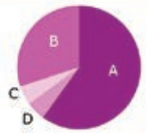
- **LEGAL SERVICES:** Jane Ann Gray, PWC.
- **AUDITING:** Bentleys
- **SF SUPER SERIES TRAINERS:** Gary and Ana Mullins (TRT Running), Pure Running, Trailblazers Run Coaching
- **STYLISTS:** Donna Lee, Bella Donna Events
- **PHOTOGRAPHERS:** Cris Mahony, Little Love Photography; Jasmine Skye Photography; Caroline Bowen, Beloved Photography; Jenny Gibbs, Sugar Snaps Photography.

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

THE DISEASE

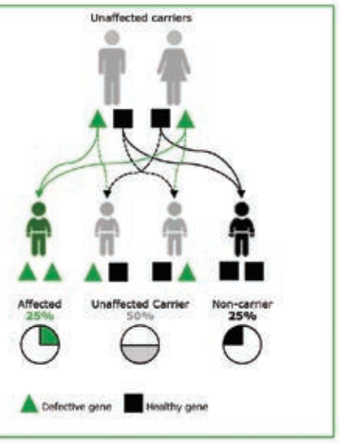
The following information provides a better understanding of the childhood dementia Sanfilippo (MPSIII).

A Rare, Life-Limiting Disease That Is Part Of A Wider Group Of Disorders

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 Form Of Childhood Dementia

Metabolic

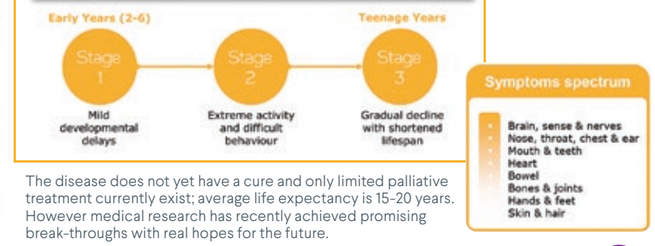
Metabolism is the set of life-sustaining chemical transformations within the cells. These transformations are done through the actions of enzymes which act as catalysts; the missing enzyme in Sanfilippo results in a metabolic disorder.

Dementia

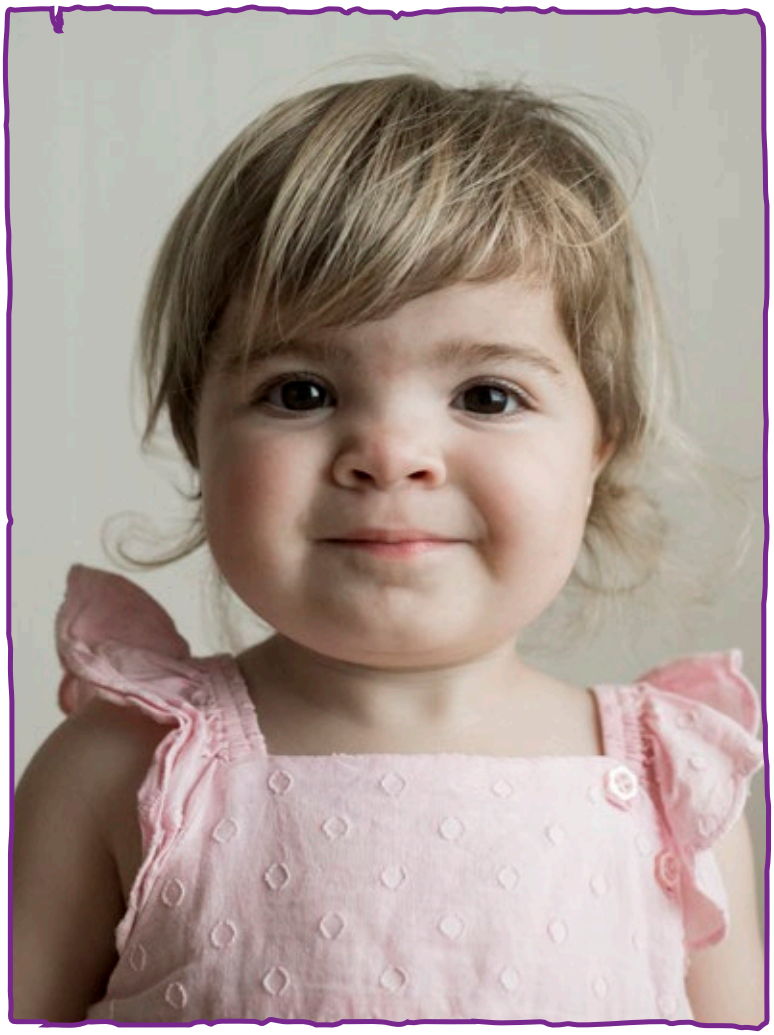
The disorder affects primarily the cells in the brain and is considered a childhood dementia. Children experience hyperactivity, sleeplessness, loss of speech and cognitive skills, mental retardation. cardiac issues, seizures, loss of mobility and finally death.

A Progressive & 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.



The disease does not yet have a cure and only limited palliative treatment currently exist; average life expectancy is 15-20 years. However medical research has recently achieved promising break-throughs with real hopes for the future.



THANK YOU

Thank you to our valued supporters, donors, sponsors, suppliers, families, volunteers and our incredible network of researchers and clinicians. We couldn't do what we do at the SCF without you!

FURTHER TOGETHER

The Sanfilippo Children's Foundation acknowledges the mutual support and respect amongst many organisations within the rare disease community.

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Sanfilippo Children's Foundation