Syndromes Without A Name

2017 Annual Report

SYNDROMES WITHOUT A NAME (SWAN) AUSTRALIA

SWAN Australia is a not-for-profit charity and a peak advocacy and support organisation for undiagnosed and rare genetic conditions in Australia. We provide information and support to families caring for a child with an undiagnosed or rare genetic condition. We also provide opportunities for families to assist them with establishing enduring, mutually supportive relationships. To do this, we offer information seminars, support services and a chance for families and parents to have fun at social events. These events give opportunities for parents to network with like-minded peers, helping combat some of the isolation and the negative social, emotional and financial impacts experienced by families whose child has an undiagnosed or rare condition. We advocate for better resources and more funding into research to ensure more children obtain a genetic diagnosis.

SWAN aims to increase awareness and understanding of the impact and prevalence of undiagnosed and rare genetic conditions within the wider community. In Britain, SWAN UK estimate that 6000 children are born with an undiagnosed genetic condition each year. Geneticists believe Australia would have similar statistics based on population. This means approximately 2650 children are born each year with an undiagnosed genetic condition.

"SWAN families experience isolation, frustration, anxiety, confusion. The hardest part is not knowing what the future holds".

OUR TEAM

Office Bearers

President – Heather Renton
Vice President – Tim McMahon
Treasurer – Alice Marshall
Secretary – Ed Atwell

Committee Members

Dani McLennan,

Cody Oliveira & Rhiana Spinoso

SWAN Ambassador

Dr Sue White

"Approximately 60% of children, who geneticists review with genetic disorders, never receive a diagnosis. This equates to around 20 children per week who could benefit by having an exome to unravel their diagnostic odyssey."

PRESIDENTS REPORT

SWAN has achieved a lot in the last 12 months. Our membership has grown to over 160 members and we have provided many hours of support, focusing on genetic testing and the NDIS. We have run sessions to help carers understand the NDIS options available to them, and attended information and advocacy sessions, so we can better support our members. Our SWAN QLD support group has taken off with thanks to Megan Firsten and Rebecca Glover. We look forward to SWAN QLD holding many more events in the coming 12 months.

We are proud to have launched our new website and SWAN brochure. Many thanks to our SWAN volunteers: Rhiana Spinoso for designing our new brochure, Leigh Eriksen for his assistance with our website glitches and Rebecca Conci for once again being our amazing SWAN photographer.

This last financial year we have focused on more SWAN events. We offered a range of different events:

- **Zebra Fish tour** – SWAN members attended a personalised tour of the Australian Regenerative Medicine Institute (ARMI) FishCore at Monash University
- **Social park gathering** – casual gathering of SWAN families
- **Rare Disease Day** – SWAN Mum Rebecca Conci shared her heartwarming story of her families' courage, and the positives of parenting SWAN children
- **Undiagnosed Children's Awareness Day (UCAD)** – this gets bigger and better every year. Melbourne's fete day had more than 300 people attend. This year, Dani McLennan hosted the NSW event, and we tried for an event in QLD, though the weather was not on our side. Many thanks to Kate McMahon for her enthusiasm and support and great publicity around our VIC UCAD event.
- **Parent Dinner** – parents gathered at Hophaus Restaurant for our first SWAN parent dinner
- **Post Mother's Day Mingle** – A pampering treat for SWAN mums at Madame Brussels
- **SWAN Dad's Group** – A variety of activities for SWAN Dads kindly organised with thanks to our Vice President, Tim McMahon

One of the goals of SWAN is to offer peer-to-peer support, and provide an environment and opportunity for members to be able to form friendships. We have started to achieve this goal, with members forming their own relationships and supporting one another through Facebook, in person and on the phone. SWAN events enable members to have a break, do something different, get some respite and spend time with others who understand them. It is lovely to see parents exchanging contact details with one another at events. It also makes me proud when parents exchange support ideas with one another on social media. I hope the year to come will bring more families together which will not only allow them to support each other but achieve one of our goals which is to look after our SWAN parents mental health by limiting the high rates of depression and anxiety amongst our members. Many thanks to Cody Oliveira for assisting with our SWAN events.

On the political front, I have been fortunate enough to share my personal story and SWAN's story with the Victorian Health Minister, Jill Hennessy MP. Also, Colleen Hartland MP has once again proved a great supporter of SWAN, speaking at UCAD.

SWAN provided feedback on the *General Health Care for Victoria – A Discussion Paper* and wrote a submission on the *Draft National Health Genomics Policy Framework*. We also made a submission on the Federal Government's *NDIS Code of Conduct Discussion Paper.*

We were successful in applying for a Darebin City Council Community Grant, which we used to host UCAD and provide activities for our families on the day. The Herald Sun continue to buy Cadbury Chocolates to support SWAN with our ongoing thanks to Kate McMahon for organising this. We once again participated People's Choice Lottery as a fundraiser.

Many thanks to our SWAN committee for their enthusiasm and support throughout the last 12 months. Our committee and volunteers have enabled SWAN to continue to be the peak body that supports families with undiagnosed and rare genetic conditions. We look forward to expanding our support network and membership over the next 12 months.

"Living without a diagnosis for your child creates uncertainty, anxiety, isolation and frustration".

Heather Renton
President - SWAN Australia

YEAR TO COME

SWAN has a number of events organised for next year. We will be recognising Rare Disease Day in February with a SWAN parent dinner in Melbourne. All of our SWAN children have a rare disease, many of them not diagnosed yet, so Rare Disease Day gives us a chance to come together and celebrate them.

In March, Undiagnosed Children's Awareness Day will once again be celebrated. The day creates awareness of the large prevalence of undiagnosed genetic conditions within the wider community and gives our families an excuse to come together and enjoy fun activities together.

All our events aim to offer opportunities for connection among our members. In 2018, SWAN will aim to increase connections among our rural members. To do this, we have a number of morning teas planned for different rural areas. We will also build SWAN's social media presence, produce informative videos and continue to build the website.

We will also offer SWAN families more opportunities for education and support. To do this, we have a number of workshops and seminars planned to help educate our members and encourage resilience through processing grief and shame.

SWAN will also focus on educating the wider support community such as maternal health nurses and allied health professionals, so they can learn more about our group and how we can assist potential SWAN families in supporting them through their unknown journey.

5 Year Plan and Projects

Currently SWAN is not in a financial position to employ an executive officer, but we hope to be able to do so as part of our five year plan. For now, SWAN is focused on advocating at a policy level to have genome sequencing included under Medicare, so all SWAN children can access it, without the financial burden. Currently genome sequencing is only provided on a research basis and not on a clinical basis. Every SWAN child should have the chance to obtain a diagnosis. A diagnosis leads to better treatment plans for our SWAN children and better social outcomes. For some members, receiving answers through genomic sequencing will alleviate some of the anxiety, uncertainty, confusion that comes with not knowing what the future holds for your child. We will continue to work in partnership with Murdoch Children's Research Institute, The Walter and Eliza Hall Institute and the Royal Children's Hospital to help get the answers our SWAN families are desperately hoping for.

BALANCE SHEET

Syndromes Without A Name (SWAN) – Australia
As at 30 June 2017

	30 Jun 2017	30 Jun 2016
Assets		
Bank		
Community Solutions One	3,656	1,811
Westpac Business Cash Reserve	50	50
Westpac Community Solutions Cash Reserve	14,094	10,022
Total Bank	**17,800**	**11,883**
Fixed Assets		
Office Equipment	671	-
Total Fixed Assets	671	-
Total Assets	**18,471**	**11,883**
Net Assets	**18,471**	**11,883**
Equity		
Current Year Earnings	6,588	8,905
Retained Earnings	11,883	2,978
Total Equity	**18,471**	**11,883**

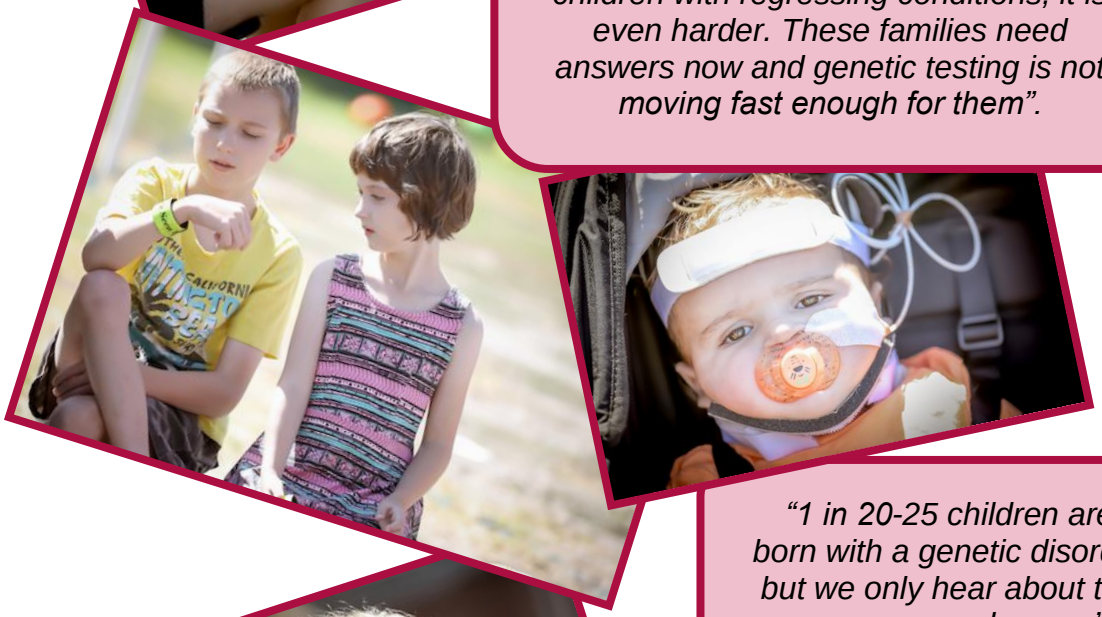
PROFIT & LOSS

Syndromes Without A Name (SWAN) – Australia
1 July 2016 to 30 June 2017

	30 Jun 2017	30 Jun 2016
Income		
Donations	515	-
Fundraising & Events	3,941	2,741
Grants	8,100	8,700
Interest Income	83	828
Other Revenue	5088	28
Total Income	**17,727**	**12,297**
Gross Profit	**17,727**	**12,297**
Less Operating Expenses		
Advertising	2,261	-
Event Expenses	5,357	825
General Expenses	75	-
Gifts	230	-
Governance Expenses	56	54
Insurance	1,755	1,755
Printing & Stationary	839	497
Telephone & Internet	40	40
Website Expenses	304	221
Total Operating Expenses	**10,917**	**3,392**
Net Profit	**6,810**	**8,905**

"A secure diagnosis leads to optimal medical management" – Dr Sue White.

"For families undiagnosed children, life is difficult enough but when you have children with regressing conditions, it is even harder. These families need answers now and genetic testing is not moving fast enough for them".



"1 in 20-25 children are born with a genetic disorder but we only hear about the common syndromes."

Photos from Undiagnosed Children's Awareness Day 2017
Courtesy of Rebecca Conci Photography

PARENT STORY

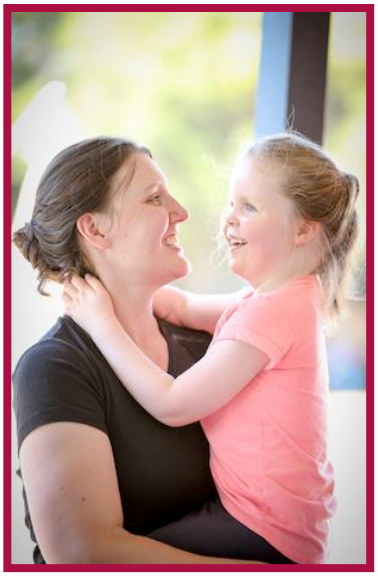


Photo courtesy of Rebecca Conci Photography

Kate McMahon, shares her story about what it is like to receive a diagnosis and how they cope when the diagnosis is so rare.

I used to joke that my daughter had "Olivia Syndrome" when people asked me what was wrong with her. It seemed a better thing to say than "actually, we really don't know". For two and a half years, my husband Tim and I had no idea why our beautiful little ray of sunshine had so many issues. It is a huge relief to be able to now say to people "she has Kleefstra syndrome".

There are now about 400 people in the world with "KS" and it is so rare not much is known about it. Typically, those with the syndrome have moderate physical and intellectual disabilities and experience developmental delays.

Almost three years on from getting a diagnosis, we still feel incredibly lucky that we received that answer. Our lives have changed a lot since getting that word to put in a box. Olivia has learned how to walk and say a few words – milestones we were never confident she would reach. And while knowing what may lie ahead in her future is not always easy – it has helped us plan for her future care. Things like getting a disability permit for the car, funding for equipment and respite care, and even helping her get a place in a fantastic specialist school have all been easier because we have a name.

Photo courtesy of the McMahon Family

I think you always assume, until you learn differently, that doctors hold the answers to most ailments. We probably would be searching for answers still if Olivia had not been put forward to participate in the Melbourne Genetic Alliance Research project, which was a pilot to show the effectiveness of Exome and genetic sequencing to diagnosing rare syndromes. We were warned in advance the chance of her Exome sequence finding something was only 20 per cent, which had really haunted us. We wanted more children in the future, but were not prepared to shoulder the unknown risk of having another child with Olivia's issues.

We got a call about ten months after the test was sent away to come in for a chat with our geneticist, Dr Zornitza Stark. She told us she had found the tiny "spelling mistake" in Olivia's genes which resulted in Kleefstra. There were a lot of tears and hugs.

Reading the brochure on KS, it was like someone was perfectly describing our daughter. It was so overwhelming and inspiring to see other families talking about how their children were at walking and talking – milestones we were never sure until that moment if Olivia would be able to meet. We are now linked in with many parents whose children have KS on Facebook and we know that there are many, many very tough challenges to come for Olivia and ourselves.

Reflecting back, living with the "undiagnosed genetic syndrome" label was like standing in the ocean. Sometimes the water would be still and clear, but you could never ignore the waves that might be lurking over the horizon. Sometimes, out of the blue, I would be hit with waves of exhaustion, regret and grief. It was easy on those days to be totally overwhelmed and to run through every detail of the pregnancy and birth wondering if at any point, we had done something wrong

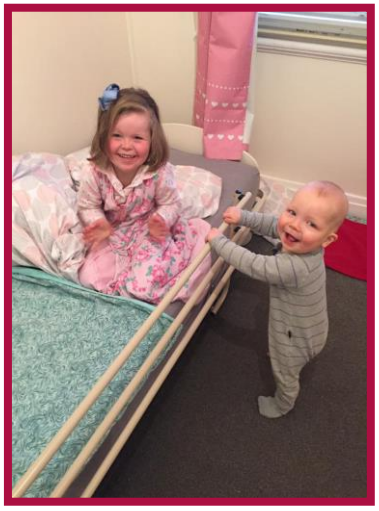


Photo courtesy of the McMahon Family

The Victorian State Government committed $25 million to fund the Melbourne Genetics Alliance program in the 2015 State budget and in 2017 they committed a further 8.3 million to genome sequencing. In time, this will have the capacity to diagnose many more people with undiagnosed syndromes.

In October 2016, we were thrilled to welcomed baby William into our lives, something which would never have happened if had not been Olivia's diagnosis. Tim and I are both proud to be advocates for SWAN and MCRI in the hope that more families will be able to get the answers they are longing for.

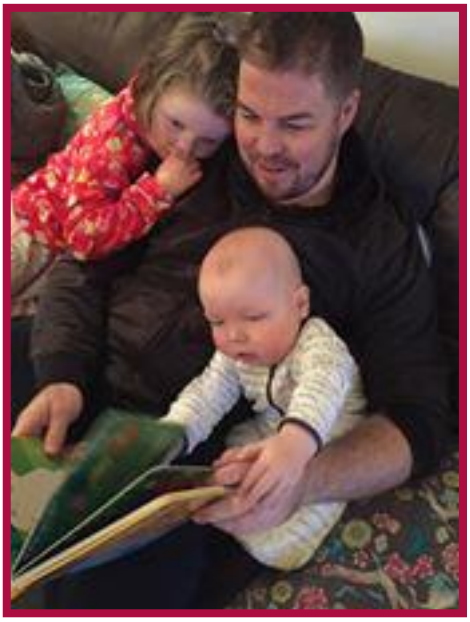


Photo courtesy of the McMahon Family

Written by Kate McMahon

SWAN WOULD LIKE TO THANK IT'S SPONSORS

FINANCIAL CONTRIBUTION

Your donations allow us to support more families, helping them establish enduring, mutually supportive relationships and giving them the information they need to make informed choices for their children. SWAN provides opportunities for families to connect over workshops and seminars, family fun days (such as Undiagnosed Children's Awareness Day) and parent dinners, where parents can have a night off to mingle and chat with other people who understand.

HOW TO DONATE

SWAN needs your assistance to help us grow and support more members. You can donate by visiting our website: https://swanaus.org.au/donate/ or https://www.givenow.com.au/swanaus. All donations over $2 are tax deductible.

SWAN CONTACT DETAILS

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