

PKS PERSPECTIVE

FEBRUARY 2017 EDITION

PRESIDENT'S REPORT

Hello members and friends, firstly I must apologise for no communication since our last newsletter. Since then, we have had our AGM and the same committee has been re-elected, thank you again for your continued commitment. Without your commitment, the Foundation will not have a future.



A little over a year ago, the Foundation was asked by a research group in Germany to help with their research. The "FDNA Project" (Face2Gene) was designed to improve and aid in the diagnosis of rare genetic conditions, such as PKS, by using characteristic facial features mainly in the early stages of life. Previous research and publications showed this had a very strong correlation with many of the conditions they tested it against and in order to increase their scope they asked the PKS community to provide information. Many PKS families kindly provided photos and information about their child from here in Australia and from around the world. Thank you to those who supported this study. I recently followed up with Dr Liehr and he informed me a paper has been submitted to a medical journal for review and publication. He envisages it should be published this year.

This brings us to the Foundation's commitment towards research in Australia. Research is not cheap, and there are many aspects to medical research which could/would benefit PKS individuals. Even though PKS cannot be cured, most of the research being done is more on what is affecting the individual and how to limit its effect. Visit our web page for an up to date list of journal articles written about PKS. The Foundation is committed to support research and finding the right research project will be difficult. The committee will offer institutions the opportunity to provide us with a research project. The amount of funding has not been decided which is dependent on many factors which need to be assessed on an individual basis. This may involve your child either by providing a biological (blood or tissue) sample or by providing some medical information. More often than not, research requires multiple subjects to make their findings statistically significant, however there are also papers written on single patients which usually demonstrate a unique feature not commonly seen.

The PKS family conference will be held in October this year at the Adelaide Shores Resort in South Australia. Tracey Hilton and her team are working hard to ensure the conference will be a success. The Foundation hopes you will make every effort to attend. The Foundation will be financially supporting the majority of the conference. Details to follow. These conferences are a forum for parents to have a better understanding of many aspects of PKS and disabilities. The organising committee will ensure highly informative speakers are present. If you have a particular topic you think we could cover at this conference please do not hesitate to contact Tracey Hilton or the committee sooner than later as if we find it of benefit we need to allow speakers adequate time to prepare their talks and to organise appropriate travel.

Nic Acquarola

PROFILE – TORAH WADE

With thanks to Manda and Steve Wade, here is Torah's profile.

1. How old is Torah?

11 on the 10th October this year.

2. When did you find out she had PKS?

We found out when she was 4 after 5 contaminated Buccal swabs.

3. What's been the funniest moment in their life so far?

We have had many funny moments and adventures Torah is number 3 of 7 kids there is lots of fun and laughter.

4. What's been the saddest time?

Saddest time was when Torah had emergency bowel surgery at 5 days old we were told she might not survive.

5. What was the hardest time in your life, as parents of Torah?

Hardest times are when the kids ask why Torah can't do things that they can. It's not very often as they just treat Torah the same but sometimes we can't do a lot of family activities as Torah can't join in.

6. Torah, what makes your Mum and Dad and family smile?

When I giggle out of nowhere or laugh at the most inappropriate times.

7. What do you enjoy most about life?

Swimming.

8. How do Mum and Dad and the family know when you are happy?

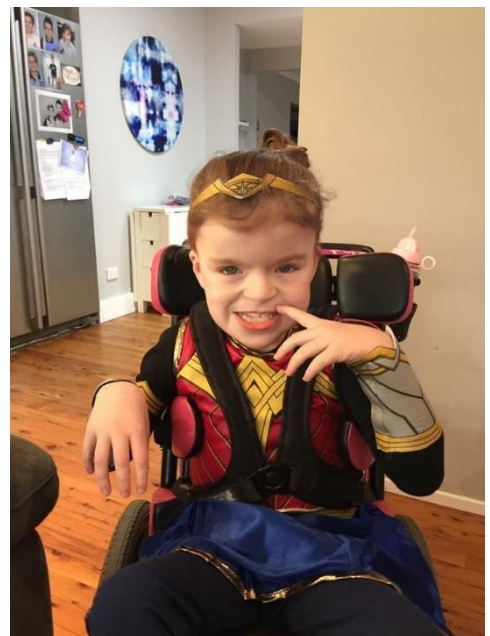
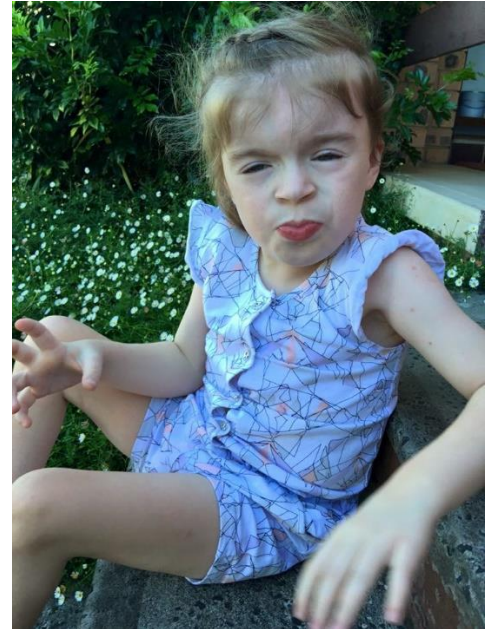
I laugh and giggle and try to hold hands.

9. Now that the PKSFA is established, parents, how do you find it?

We are not ones to ask for help but knowing that there is support if we need it is very reassuring.

10. What are your hopes and dreams?

For Torah to be happy and healthy and enjoy many more fun and memorable moments with our family.



PROFILE – TORAH WADE (CONTINUED)



Photograph of Torah's family.

THE PKSFA GRANT PROGRAM

Several families have been successful with obtaining funds for equipment for their child. In future newsletters, we will bring you reports on some of those. Please do not hesitate to contact the Foundation if you would like to seek some funding.

PKSFA FUNDRAISING

The Foundation did not see undertake any major fund raising event in 2016. The committee will discuss the future of the Entertainment book campaign at our next meeting.

As a result of Namita's Bollywood Mania in 2015, IOZ Events kindly presented the Foundation a \$1500 cheque as they donated their proceeds to the Foundation from a concert held in early 2016. Once again a big thank you to IOZ Events for their support.



WA now has a long term sponsor in Marcel LaMacchia of One Residential. Marcel set up a My Cause account called "Marcel LaMacchia reaching out for PKS" where Marcel is kindly donating \$100 per house sale to the Foundation. His goal is \$5000 and is currently standing at \$1300. This is a novel initiative where an organisation can commit a set amount per sale of a property to the Foundation. If you think you have someone that would like to support the Foundation in a novel way please do not hesitate to pass on our details.



PKSFA WEB PAGE

We will be updating the web page on a more regular basis this year, please visit the site for new and interesting information. Any improvements, corrections or suggestions please let the committee know.