

PKS PERSPECTIVE

FEBRUARY 2018 EDITION

PRESIDENT'S REPORT

With the blink of an eye, 2017 has gone and 2018 is upon us. Reflecting over the past 12 months, the Foundation had an enormous year. Most of our energy was centred towards the Conference. We were very fortunate that 7 families were able to attend. A big thank you to **Tracey HILTON** for all her hard work in organising the conference.



In 2016, we asked families to participate in a research project by a German group (Prof Liehr). This involved sending various facial photos of your children at different ages, from birth to present. The aim was to use characteristic facial features to help predict with a high probability the condition. A journal paper was finally written, and we were acknowledged as co-author in the paper. We will place a copy of this paper on our web page.

We welcomed a new family from Queensland (must be something in the water) as well, as there is also a new family in WA.

The Adelaide conference also gave the Foundation the opportunity to run the AGM. With the unfortunate retirement of **Namita BISWAL** (vice president) and **Spiro CONOMOS** (secretary) saw two new faces stand up to help take the Foundation forward. A big thank you to **Tracey HILTON** (vice president) and **Steve WICKHAM** (secretary and newsletter editor). Namita was our founding executive member and has been active with various fundraising activities over the past few years. On behalf of myself personally, but also from the members, thank you Namita for all your hard work over the past 5 years. Spiro does not go unnoticed, thank you for all your work as secretary.

Nic Acquarola

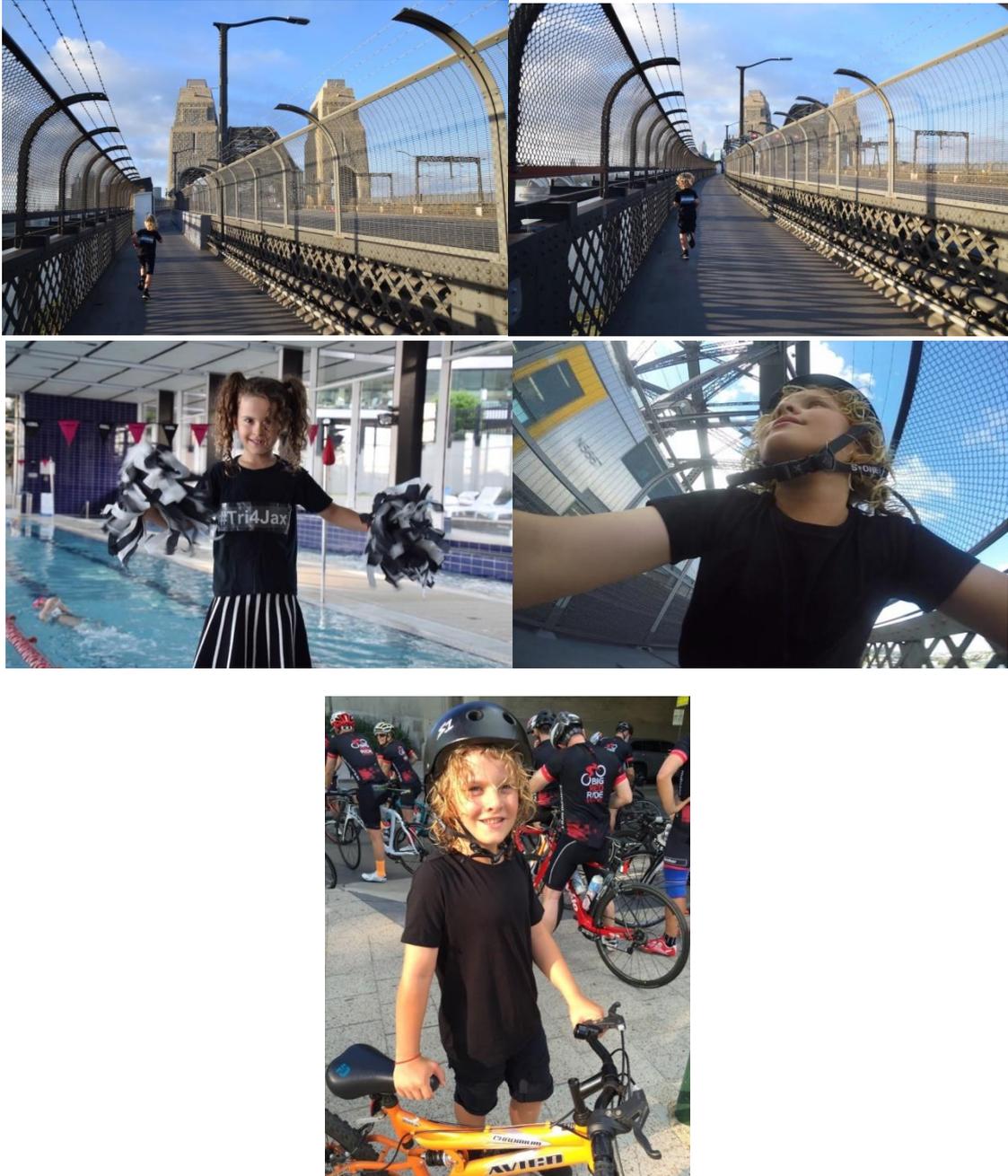
TRI-4-JAX FUNDRAISER

One of our new family members had undertaken in an amazing fundraising event that has raised the bar. The **PARSONS** family (Kristy-Lee, Michael and Jax) had decided to undertake in an extraordinary fundraising event.

Michael along with 4 of his fit friends decided to team up and compete in the prestigious Phuket Ironman 70.3 Triathlon. Using son Jax as the motivator to raise funds, the "Tri-4-Jax" team used the Go Fund Me page for supporters to donate funds. A full detailed story of the event will be placed on the web page soon. At time of press this team had raised over \$53,000!!!!

To make this story even more amazing was the efforts of a family friend of the Parsons. **Rafferty LAIGHT** from Sydney decided to also help contribute to this event by competing in his own “mini triathlon”. Raff competed in a 3.81km, followed by a 400m swim and then a 7.93km ride. Why is this so amazing? Well, Raff is aged 9. Raff raised \$1125 to also add to the “Tri-4-Jax”. Speaking to Raff’s mother, his goal was to raise \$300, so he well and truly surpassed that.

Photographs of Raff’s efforts:



THE PKSFA GRANT PROGRAM

Several families have been successful with obtaining funds for equipment for their child.

Please do not hesitate to contact the Foundation if you would like to seek some funding. The PKS-Kids family conference is being held in Minneapolis, Minnesota on July 12-14, 2018. If any of our family members would be interested in attending and flying the PKSFA flag, please contact Nic if you are interested.

PKSFA MARKETING

Namita BISWAL has kindly offered look and put into practice ways to increase the exposure of the Foundation, through merchandise and advertising. We are looking for volunteers to help Namita in this role. Please contact Nic if you are able to help.

RESEARCH AND PKSFA WEB PAGE

This has been updated to include relevant medical journal articles published up to beginning of 2017. Refer to our web page for details. The Foundation will consider participating in any form of research, so if your medical service provider has any suggestion or thought, please refer them to the Foundation. We have been 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

Following are details of the Paper published with credit to PKSFA as co-author:

Clin Genet. 2017 Jun 29.

Next generation phenotyping in Emanuel and Pallister-Killian syndrome using computer-aided facial dysmorphology analysis of 2D photos.

Liehr T, Acquarola N, Pyle K, St-Pierre S, Rinholm M, Bar O, Wilhelm K, Schreyer J.

Abstract

High throughput approaches are continuously progressing and have become a major part of clinical diagnostics. Still, the critical process of detailed phenotyping and gathering clinical information has not changed much in the last decades. Forms of next generation phenotyping (NGP) are needed to increase further the value of any kind of genetic approaches, including timely consideration of (molecular) cytogenetics during the diagnostic quest. As NGP we used in this study the facial dysmorphology novel analysis (FDNA) technology to automatically identify facial phenotypes associated with Emanuel (ES) and Pallister-Killian Syndrome (PKS) from 2D facial photos. The comparison between ES or PKS and normal individuals expressed a full separation between the cohorts. Our results show that NPG is able to help in the clinic, and could reduce the time patients spend in diagnostic odyssey. It also helps to differentiate ES or PKS from each other and other patients with small supernumerary marker chromosomes, especially in countries with no access to more sophisticated genetic approaches apart from banding cytogenetics. Inclusion of more facial pictures of patient with sSMC, like isochromosome-18p-, cat-eye-syndrome or others may contribute to higher detection rates in future.