Pallister-Killian Syndrome Foundation of Australia
Education • Awareness • Support

PKS PERSPECTIVE

APRIL 2016 EDITION

PRESIDENT'S REPORT

Christmas, New Year and Easter have all come and gone since our last newsletter. Hopefully this finds you and your family all fit and healthy. The committee have settled down to individually work on various projects which will come into fruition over the next few months.

A big thank you to those who took the time to compile their child's photos and forwarded them to the FDNA group. It's not too late if you still wish to submit these photos and information to the group, details are with our December 2015 edition. The lead researcher informed me that those who forwarded the photos and information was greatly appreciated and will go a long way to aid in the early and non-invasive diagnosis of PKS. If you have any questions or require instructions, please do not hesitate to contact me.

Presidents from the three PKS organisations (PKSFA, PKSKids and PKSKids Italia) have been in discussion over the past 6 months regarding setting up a worldwide organisation (PKS International). Essentially this will be a central registry where individual information is maintained. The concept is quite complex to discuss at the moment as it is still a working progress. World leading specialist Dr Krantz, who has worked closely with the US PKSKids and now the PKSKids Italia, would be on the board. Safety procedures and data access has been of paramount importance in establishing this registry and will be only accessible to a select few people. Meaning all data kept will be maintained in the highest integrity, confidentiality and security.

The plan is also to maintain and promote the PKS Biobank which already has samples from 100 PKS individuals collected over the past 7 years by Dr Krantz's group. If this can be expanded to include other nations, then the availability of material for medical research would be invaluable in better understanding PKS. PKSFA can contribute to the Biobank in several ways, one of which would be though the PKSFA conference, where material can be taken from our children. This material will be kept at the present location the CHoP (Children's Hospital of Philadelphia). This material could conceivably be made available to other international researchers. Biological material release would be vetted and controlled through Dr Krantz and PKS International.

Thank you to all the families that have shared their child's stories with us. In this edition we get to know Sarah Paton.

Nic Acquarola

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MEET SARAH PATON

THIS EDITION of PKS Perspective we profile Sarah Paton, ten-year-old daughter of Tim and Rebecca, and older sister to Hannah.

Sarah was born on her due date in January 2006, and had her PKS diagnosis confirmed when she was eight months old when she was admitted to Royal Children's Hospital to have her cleft palate surgery.

Rebecca has been Vice-President of PKSFA and Tim has been a committee member.

Sarah at 10, with Mum, Rebecca.

1. How old is Sarah? ***10 years' old.***

2. When did you find out she had PKS? Tell us about that.

Sarah was diagnosed at 8 months of age whilst in hospital recovering from surgery to repair her cleft palate. We had known for a couple of months that something genetic was likely to be the issue but it took the whole genetics team and a cheek swab to finally identify it as PKS.

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

Difficult to pick one particular moment but there are times when she gets the giggles about something – anything really and she just lights up the whole room. Sometimes she bursts out with a laugh when someone has made a random comment and her timing is comic perfection.

4. What has been the saddest time?

When she was recovering from her bilateral femoral osteotomies (major hip surgery) and we struggled to find the right level of pain medication for her. Some very long sad nights.

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

Tim says the first few days when we were told she might have Down Syndrome and were waiting for the results for 4 days (which of course were negative).

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

Giggles.

7. What do you enjoy most about life?

Music, balloons, bubbles and cuddles.

8. How do we know when you are happy?

Big smiles, clapping and giggles.

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

We helped create it! We were part of developing the idea but full credit to Nic Acquarola for doing the hard yards.

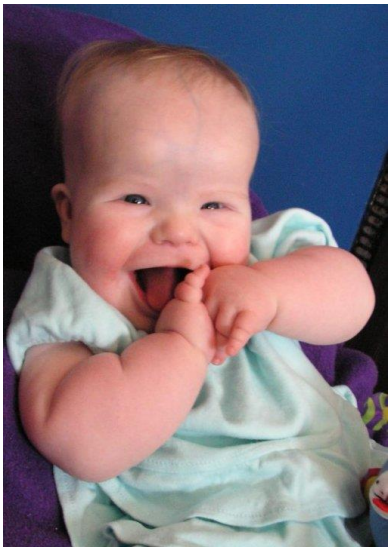
10. Tim and Rebecca, what are your hopes and dreams?

For both of our girls to have long and happy and healthy lives.

From 2013:

We don't know what Sarah will learn to do in the coming years but for now we have become accustomed to the wheelchair, the hoist, the medications, the hearing aids, glasses, orthotics, ramps, van, appointments, specialists, etc., etc., etc. This is our version of normal life and, though it is not the life we would have chosen, we are making it a happy one.

Happy moments: Sarah, Hannah and Tim at hospital in December 2011.



Sarah at 10 months.

Tim and Sarah at Christmas time.

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THE PKSFA GRANT PROGRAM

The grant is open all year round. If you require assistance in the purchase of an item for your PKS child (eg: IPad, software for IPad, sensory toys, bicycle, compression suits, etc), provide additional physiotherapy/OT or even if you would like to attend a PKS related conference. Please do not hesitate to contact the foundation to discuss your wishes.

THE PKSFA CONFERENCE 2016

Bec Paton has been narrowing down venues and speakers for the next conference. A date has not yet been set, but it may be either late 2016 or early 2017. Watch this space. Again, the Foundation will discuss funding arrangements of this event. If you have any suggestions with respect to topics or guest speakers, please do not hesitate to contact Bec.

PKSFA FUNDRAISING

This year we have decided to sell the Entertainment Book again. 20% of each sale goes to the Foundation. We urge you to please support this worthy drive.

Follow the link below if you wish to support the Foundation. Alternatively, please do not hesitate to forward it to family and friends. The more we raise the better the service we provide for your child.

https://www.entertainmentbook.com.au/orderbooks/911786a

WA have decided that 2016 would be the year of the Quiz Night or Bogan Bingo.

Details to follow. Donations and offer for assistance would be greatly appreciated.

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MEDICAL RESEARCH

FDNA project

I was recently contacted by a medical researcher in Germany who is doing work with Facial Dysmorphology Novel Analysis (FDNA). I have had several email correspondences with the lead investigator on this and feel that we can contribute to this study. What the group (www.FDNA.com) have devised is a computer program that looks at the bioinformatics and correlates them with certain genetic conditions.

"Face2Gene provides the genetics healthcare community with information on facial dysmorphic features and patterns of human malformations recognisable in facial photographs. The tool enables genetics professionals to take a photograph of the patient's face, analyse it by looking up syndromes in references, such as Orphanet, and constitute a differential diagnosis. FDNA will continue to improve its technology over time, referencing and synchronising comprehensive phenotype databases. Incorporating Orphanet into Face2Gene will help improve the diagnosis, care and treatment of patients with rare diseases."

Source: http://www.orpha.net/actor/EuropaNews/2014/140918.html

Page 4 provides simple diagram on how it works: FaceBaseFDNA.pdf

How can PKSFA help?

1. Find a series of head/face shot photos of your child, with nothing occluding their face – such as dummy, face painting, spectacles. Rename the file with their age (approximate age), name not required but for ease of the researchers to catalogue you could rename each file as: "JB_2m", "JB_24m", "JB_4y" meaning Justin Beiber 2 months. Preferably yearly (but every two or three years is ok) until present age. You can also include full portrait/frontal of your child (CLOTHED), in lieu of no facial frontal photos, but this is not necessary. Ideally one at birth or near birth, 6m, 12m then yearly.
2. The child does not need to be smiling, but it's important that the eyes are open.
3. The photos need to be submitted as individual photos and not a compilation on one file (so no fancy work with Photoshop).4. They can be of photo format (eg: .jpg, .gif) or PDF. If you only have print photos, then scan one and submit that (especially those with much older children where digital was not around).
4. Once collated, then please email them to Dr Thomas Liehr (Thomas.Liehr@med.uni-jena.de) and in the email please include approximate age of child when diagnosed, if you know the % cells affected when diagnosed, and current age of child now. To help with downloading and having huge files, possibly send two at a time on each email. This should be included in your introductory email.
5. To help with their study can you please have this all sent no later than end of February 2016.

If you have any questions please do not hesitate to contact me directly and I will advise where I can. This is not compulsory in any way and you do not need to inform anyone if you do not intend to provide any information.

WHAT'S HAPPENING IN THE NEAR FUTURE?

- PKS conference to be held in Sydney in 2016. Details will follow.
- PKSKids Face-to-Face Medical conference to be held in the United States in July 2016. Details on the PKSKids website. If you would like to attend please contact the Foundation.

WEB PAGE AND IT

The web page is a work in progress, which will be updated from time to time. Don't forget to check regularly on any updates. Of course, any comments would be appreciated. Facebook, Twitter and Instagram have all been established and are useful in getting short messages out about any upcoming events.