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

PKS PERSPECTIVE

DECEMBER 2015 EDITION

PRESIDENT'S REPORT

HI MEMBERS AND FRIENDS, much has happened since the July edition in the Foundation. The AGM was held in August which showed the Foundation has thus far held up to expectation. We have had a slight reshuffle of committee members as well as a new member. Firstly, I would personally like to thank **Bec PATON** for her role as Vice President since the inception of the Foundation. Bec contributed so much to the Foundation in the early stages and has gladly offered to step aside to allow Namita the opportunity to take on the duties of Vice President.

Bec was instrumental in driving the concept of having a support network (similar to the PKSKids in the USA), without her drive I do not think we would have been able to have set the model for a successful Foundation. Bec is still actively involved with the Foundation as our current Conference Coordinator. No doubt we will see Bec back in the driving seat in the very near future. We welcome Spiro CONOMOS to the executive committee as our Secretary. After a month of being barraged with legalities, Spiro has settled in quite comfortably. Terzita ACQUAROLA presented the financials over the year which showed we are financial. However, what is of concern is the reluctance of our members to take on our Grants program. This may be for equipment, supplementary PT/OT which is additional to what your child currently has or even for the attendance of a PKS related conference both nationally and internationally. No matter how large or small please do not hesitate to contact the executive committee to discuss how we can help. All requests will be kept confidential if you require.

We have not heard back from the UK group regarding the questionnaire, which they were waiting for ethics approval.

We have been approached for another research group for support, which I will outline below. After several emails to the principal organiser of this research I am confident that PKSFA with the help of its members can help out with their request.

Namita BISWAL successfully organised her first major fund raising event in October, which two PKS families supported. Well done Namita, all that hard work paid off. Thankyou also to the families who supported this event, one came from Queensland!

The Foundation still has many of the information brochures, so if you would like to have some please do not hesitate to contact your friendly neighbourhood committee member. We are also planning to revamp the brochures with may take on two forms; one for the general public/family and one for the medical professionals.

With Christmas and the New Year just around the corner, PKSFA would like to extend our wishes to each and every one of you and thank you for your continuing support. We hope that 2016 is a good year for you.

MEET THE HILTONS (SARAH, AND PARENTS, CHRIS & TRACEY)

I SUPPOSE having disabilities ourselves made it easier to accept Sarah. Chris is partially blind and I am totally deaf without my cochlear implant. No one plans to have a special needs child though. My dreams for Sarah's future died the day we had our appointment with the geneticist. Being told she would be dependent on us for the rest of her life when she was six years old turned our world upside down. Looking back, it's a blessing in a way that Chris didn't want to have a diagnosis earlier as we didn't know the statistics about PKS; that only 30% walk.

Baby Sarah.

Sarah Ruth was born in the small country hospital of Dalby, Qld in December 1995. Our one and only after many miscarriages but that's another story. Nothing was amiss those first few days or weeks that followed. Questions began to arise in my mind at four months, when she wasn't doing things like another baby, Hannah. Merrilyn, had given birth to her fifth baby just ten weeks before I had Sarah.

Every week at church I learned so much watching Hannah and talking to Merrilyn. By nine months old I knew that something was wrong and help was needed NOW. God led us to the right help at the right time. I was sharing about Sarah to our local librarian who immediately suggested I order Glenn Doman books. In these books was the information to help us develop a daily routine to get Sarah back on track to achieve the milestones. Between fifteen months and five years old we turned our home into a mini gym.

Chris and Sarah playing trumpets at home.

The hardest part (emotionally) was the daily, repetitive program of essential exercises which we had to perform on Sarah with enthusiasm and glowing happiness.

Very vital and yet very hard; with so little results to show for it all for many months.

She sat on her own at fifteen months.

Crawled at twenty-two months (four months' work).

Walked at thirty months (eight months' work).

Her Paediatrician arranged for us to have Qld government department Family Services come to see us on a monthly basis. This gave us access to physio, O.T and Speech. Unfortunately, they didn't like Doman and challenged us about our insistence we get Sarah crawling before walking. Their argument being "She needs to catch up to her age group." We were not to be deterred.

One funny incident happened, probably when she was six. I was with a group of ladies and their children were all playing together. Sarah was just walking around watching them. One boy came over to his mum upset with Sarah. "She called me a good girl!!" I laughed and explained that because Sarah was an only child and Chris and I said "good girl" to her, she was quite repetitive saying it, it was almost the ONLY thing she said.

She shows her happiness by singing or babbling away in a lovely sweet voice. Flaps her hands and dances in her own way. Having our speechie teach Sarah how to type a few years ago opened the door for communication. She now uses her iPad (with me giving her wrist support) and we have discovered who our daughter is.

She likes bananas, monkeys, sharks, the beach and her DVD's "Donut Man." She enjoys going twice a week to day-respite where she has her own lovely support worker to care for her needs. She's learning how to go out on public transport and is slowly making friends.

PKSFA makes parenting Sarah so much easier! Knowing other families whose children face similar challenges, it's reassuring of the support and information that's there as we all travel this journey to make our PKS children's lives the best they can be.

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BOLLYWOOD MANIA

Bollywood dancers performing at Bollywood Mania which raised almost $14,000 for PKSFA.

THE PKS BOLLYWOOD MANIA on October 10th was well attended by over 220 guests in NSW. The event raised almost $14000 and generated a lot of awareness about PKS via local media and word of mouth.

The special guest speakers for the event were Dr Rani Sachdev and Joanne Gardiner. Dr Sachdev (pictured below right) is a renowned geneticist from the Sydney Children's Hospital. She spent nearly 18 months trying to get the answers for the Biswal family and finally got the diagnosis of PKS through a skin biopsy. Joanne is the Principal of Sir Eric Woodward Memorial School for kids with special needs that Yash Biswal attends.

Joanne Gardiner's speech was both inspiring and moving.

The event showcased performances from the northern and southern parts of India and there was an engaging, contemporary Bollywood performance that got the guests on their feet.

With Henna Tattooing, dance floor and Photo Booth the event was enjoyed by all age groups.

A delicious, lip smacking 3 course Indian meal made the event delectable!

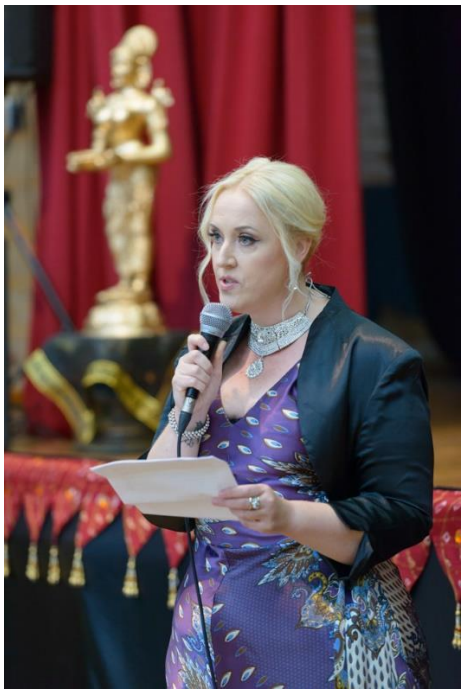
This event wouldn't have been possible without the support of a dedicated and passionate team of volunteers and our corporate supporters and donors.

A BIG thank you to everyone that believed in us and made this possible, including the guests.

Namita and Yash Biswal.

The Martin, Hilton and Biswal families represented.

More fun on the night with the Bollywood dancers and guests performing.



Special guest speaker, Joanne Gardiner.

Bollywood musicians performing.

Jared and Sue Martin.

Glenn and Sue Martin and family.

THE PKSFA GRANT PROGRAM

A reminder of the PKSFA grants program. No matter how large or small you are seeking, the Foundation is there to help support you.

If you are interested in attending any of the international PKS conferences, please do not hesitate to contact the committee to discuss grant support.

THE PKSFA CONFERENCE 2016

Bec Paton is currently researching 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

The Foundation is considering a nationwide fundraising event for 2016, which will be open to the public. This may take the form of a raffle or an auction (similar to Ebay). Namita and Bec are in the process of looking at the best format and prizes which would be attractive to the public. Watch this space. This fundraising campaign will be used to finance the conference and as such we will be reliant on your support for its success. Should you know of anyone or business who is able to support us with a donation, please do not hesitate to discuss it with the Foundation.

If you would like to organise a fund raising event in your area please discuss it with one of the committee first as we have the appropriate certifications for you to produce. Remember: help us to help you.

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.

PKSFA MAJOR SPONSOR

Many thanks to Nick Acquarola from Pentana Solutions in Melbourne who is pictured presenting Nic Acquarola (President, PKSFA) with a cheque for $2000. The welcome donation will be used to provide much needed equipment for children like Alessia. This outdoor stroller will allow Alessia to experience and enjoy the outdoors just like anyone else. Families often are faced with huge financial burdens when it comes to special needs equipment. Along with specialised equipment, the Foundation also uses funds to provide compression suits which help children to improve their muscle tone.

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