

A Courageous Battle Yash and Pallister-Killian Syndrome



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Pallister-Killian syndrome (also tetrasomy 12p mosaicism or Pallister-Killian syndrome) is an extremely rare genetic disorder occurring in humans.

Pallister-Killian occurs due to the presence of the anomalous extra isochromosome 12p, the short arm of the twelfth chromosome. Affected individuals have unusual facial features, mental retardation, seizures, patchy color differences in the skin, and various other physical abnormalities. Many fetuses with Pallister-Killian syndrome die during pregnancy or soon after birth. There are 12 to 15 known cases in Australia with 5 known in Sydney of which 3 cases were just recently reported among sufferers who are now adults.

In a recent interview with Ms. Namita Biswal, Secretary of the PKS Foundation Australia, we delved into this subject to help highlight a syndrome although considered rare, if not very rare, which needs to be considered a part in the list of disability afflicting individuals. The Indian Telegraph spoke with Ms. Biswal, to know more about PKS, the Foundation, and her own story with her youngest child Yash. It is beyond doubt that she knows the daily challenges of this syndrome, the sacrifices being made and the future of caring for Yash should this disorder not make into the National Disability Scheme.

Yash was born as any other normal child, meaning there was no hint or otherwise to suggest the challenges to be faced as a family. Having a 7 years old son, Namita knew exactly what to look out for and the care needed for a newly born child. It was after six months, the parents noticed something not normal about Yash. His development seemed a bit lacking and as she put it, "we raised the red flag and took him to the nearest community nursing centre to put our mind at ease..." Little did they realize the full impact about to be faced as they were told at the nursing centre the usual 'kids will take time to develop' and it is 'too early to tell' responses. With the red flags waving, they took him to Sydney Children's Hospital where the methods of testing and analysis albeit being more thorough, the diagnosis was more confounding. By this time Yash was 11 months old and was suffering from 'Global



Developmental Delay', which is a generic term used for disorders either too rare to diagnose or unknown to the medical profession.

However, a geneticist took up their case. Dr. Rani Sachdev, delved deeply into Yash's condition. She noticed his facial features such as large forehead, 'coarse features' such as droopy eyelids, widely spaced eyes to name a few. Any unknown diagnosis like Global Developmental Delay and using terminology such as 'coarse features' only adds to the stress and fears on to the parents, but after 18 months of waiting, Dr. Sachdev had an answer. Pallister-Killian Syndrome (PKS). Ms. Namita Biswal recalls the day, "Dr. Sachdev calls us up and she asks if we want to know the good news or the bad news and having asked for the good news first we knew what the condition was, Pallister-Killian syndrome but the bad news was the lack of knowledge and research

into this disorder." This also explains the lack of correct diagnosis at the Children's Hospital. It is a disorder known mainly by the geneticist due to the genetic disorder of PKS, coupled with the fact that it is a very rare condition.

The major challenge now facing the family, not just Yash, was dealing with this disorder. On the one hand, there was a perfectly normal and healthy child, on the other quite the opposite needing special care. With no family support, nor this disorder being part of any governmental scheme to claim assistance, Namita faced a bleak choice. She had to quit her job to take full time care of her youngest child, not knowing the proper therapy required she did her best to cope and started to look out for families with the same problem. This was going to be the genesis of the Foundation. By the end of 2011, the Founding families were getting in contact with one another. They held



a series of conversations and correspondences to form, shape and develop the PKS Foundation Australia. So far fund raising activities have taken place in Western Australia and New South Wales (under Namita Biswal's charge), one is to be held soon in Victoria.

In terms of monetary challenges faced by families with PKS kids, the cost of speech therapist alone is somewhere between \$300-\$500 per hour, the cost of a wheelchair equipped to assist a child with low mobility skill is at a whopping \$1500 each. Needless to say, not everyone can access such care and facilities, especially if the speech therapist themselves do not know the method and approach required in dealing with kids suffering from PKS. As there is more research and development in dealing with sufferers of Aspergers or Down Syndrome, there is also the same need required for PKS without which

any efforts to assist a kid will not be fully realized. Therefore, to assist families for the necessary funds, PKS Foundation undertaken fund raising activities last year.

There is a three-fold approach to PKS Foundation Australia, firstly, to garner support and improve the quality of life for sufferers of this disorder. Therefore, the first point of support is for the kids and family as they bear most of the stress and pressures. Another aspect is for grants and therapy, finding and supporting the best means and methods to increase the chances of support. Secondly, increase research activity in this area. For instance, 95% of kids do not talk; they have visual and auditory impairment or both as in Yash's case (he cannot focus directly on an object, rather looks from the corner of his eyes). Research is needed to study the neurological condition and therapy practices



required to best deal with this syndrome. Lastly, by means of the above two, educate and raise awareness of this syndrome. As stated before, the medical profession was unaware of this condition by giving a generic term to the condition suffered by Yash and more many other similar situations.

This year, Namita intends to give 5 grants to very deserving families as recognized by PKS Foundation Australia, increased grant from 2 last year, of \$1500 each. On 28th July, The Pallister-Killian Syndrome Foundation of Australia is conducting a fund raising event at the Lane Cove National Park (entry from Lady Game Drive gate, at the Riverside Walk). Entry is free, and donations towards the cause are appreciated.