

Supporting Aussie Kids with Kabuki Syndrome



SAKKS NEWS

www.sakks.org

SAKKS TEAM:

petal@sakks.org

adrian@sakks.org

kim@sakks.org

stacey@sakks.org

kelly@sakks.org

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Its hard to believe its almost December—that time of year where we vow that next year will be more organised, and we are already looking around for next years diary so we can write all those appointments in.

There has been a bit of chatter about Kabuki Syndrome Family Get togethers, and we have the first of our State ones next month. See details below :

Victorian Family Get Together

DATE: Sunday 2nd December 2007

TIME: 11.30am

VENUE: Melbourne Botanic Gardens

BYO food and drinks. Come for a relaxing day in the botanic gardens with other KS families. We will have a picnic lunch then enjoy a play on the grass and a walk around the grounds. We look forward to seeing you there. RSVP Stacey

Kelly has organised a State day for March next year and wants to hear from interested families so everybody's needs are met. If you would like to attend the Sydney State Day in March next year please contact Kelly.

If you are from Queensland or looking for an excuse to go to Queensland please contact Kim for details on her State day.

Janelle needs your information and RSVP as soon as possible for those interested in the Weekend Retreat next year.

The SAKKS brochure has undergone some changes and the PDF will be sent to you all shortly.

Don't forget to check out the contacts page and make our newest members feel welcome, we have had several new memberships this month who have already joined our email groups and enjoying meeting new families.

Please find attached an interesting article written by Tiong Yang Tan MBBS FRACP- Clinical Geneticist, who has been kind enough to write this for our SAKKS members.

Be happy,

THE SAKKS TEAM

We have had a few issues with our family day page and have made a couple of changes; I have included a couple of pics of a few of the kids enjoying themselves in Queensland.

If you have any photographs you would like to share please send them to Peta.



A story from a Mum

"We walk through the shopping center, his hand in mine. Jake looks lovingly up at me and says "Gee I love you Mum, and I really love spending time with you". Sundays are special. That's Dad and Son Day. Jake goes out with Dad for special time. Jake loves it. Dad loves it. Sunday mornings start with cuddles in bed and followed by cartoons and toast in the lounge room, we love Sundays. We love being a family. I look around the place, and see the young boys of today, same age at Jake, nearly 12, and the worst punishment in the world is going shopping with Mum, or yuk having to spend time with Dad, or heaven forbid we should ask them to play with their little sister. I have a wonderful, special Kabuki boy. His name is Jake."

What role does a clinical geneticist play in the lives of children and adults with Kabuki syndrome?

Author: Tiong Tan

Children and adults with Kabuki syndrome often see many health professionals. These may include, but are not limited to, their GP, paediatrician, physiotherapist, heart specialist, speech pathologist, dentist, orthotist, immunologist, and eye specialist. Once every couple of years, they might see a geneticist. What does a geneticist do? And what can a clinical geneticist contribute to the lives of families affected by Kabuki syndrome?

A clinical geneticist is a medical specialist who cares for people with conditions that have a genetic component. A large part of clinical genetics practice is the management of children who are born with multiple birth defects, some of whom are diagnosed with a condition such as Kabuki syndrome. Clinical geneticists usually become involved in the lives of such children when they are asked to make a diagnosis to explain the pattern of medical problems experienced by the child. Kabuki syndrome is a rare condition that is distinctive. Its recognition allows advice and management to be tailored specifically for the affected individual. We base this advice on what we know from our collective medical experience of looking after other individuals with Kabuki syndrome.

Making the diagnosis of Kabuki syndrome does not give us the power of a crystal ball. It does not predict what problems will happen, or when they will happen. But it does allow us to draw up a plan to anticipate some of the problems that might happen, and to avoid them, or at least reduce their impact. It is somewhat like drawing up a road map for the future, to help keep the child on the healthiest route. The clinical geneticist is aware of the possible complications of Kabuki syndrome, and is able to guide the whole care team about how to keep the affected child in the best possible health.

Often a diagnosis of a condition like Kabuki syndrome means that the child will have special needs in the future. The clinical geneticist is in the position to advocate for additional help in school to maximise the learning potential of a child with Kabuki syndrome. The clinical geneticist is also in a position to offer support and care to the entire family, not just the affected child. Often parents have questions about whether they might have another affected child, or whether their other children might have an affected child. These are questions that a clinical geneticist can address. We are also aware of any new research findings, and can provide this information or facilitate involvement in an ongoing research project.

By following a child and his or her family over many years, we learn a great deal about some of the difficulties that have to be overcome, and hopefully contribute in a positive and meaningful way in the management of the family's medical and genetic health.

Genetic Health Services Victoria provides clinical and laboratory testing for genetic conditions, as well as counseling and support services, to help all Victorians make informed decisions about their health and wellbeing. Clinics are located all around metropolitan and regional Victoria, allowing our services to be accessible to all Victorian families. If you would like to make an appointment to see a clinical geneticist, or to learn more about our services, please contact (03) 8341 6201, or visit our website www.genetichealthvic.net.au.