Please fill in the following form if you would like to register with SAKKS or to make a tax deductible donation.

There is no cost to become a member; our memberships include: Family, Associate and Professional.

( ) I would like to become a member
( ) I would like to make a donation

First Name_____

Surname_____

Street_____

Suburb_____

State_____

Telephone _____

Email _____

Comments:
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Send to: SAKKS— PO Box 318, Rundle Mall, South Australia, Australia, 5000

www.sakks.org

SAKKS—Supporting Aussie Kids with Kabuki Syndrome was established in 2004 by the parents of a child with Kabuki Syndrome who were faced with the isolation that can be associated with this rare syndrome. Since its inception, SAKKS has grown into a Non Profit Incorporated Association with representatives in most states.

Our mission is to support those affected by KABUKI SYNDROME by offering:

- Information, external links to genetics departments and research as well as links to medical information.
- Contacts page—a link to families
- Story pages written by parents as well as photograph pages displaying many of our special members and their journey so far.
- Forum and stories page exclusively for siblings.
- Regular newsletters with the current activities of SAKKS.
- Professional membership; a forum for professionals to aide in information sharing.
- Annual family days in each state and Nationwide Retreats bi-annually.

To contact one of our team members go to our " contact us " page at www.sakks.org - we look forward to hearing from you.

SAKKS

Supporting Aussie Kids with Kabuki Syndrome Inc.

www.sakks.org

What is Kabuki Syndrome?

KABUKI SYNDROME — Also known as Niikawa-Kuroki Syndrome.

Kabuki Syndrome is a rare genetic disorder that occurs in approximately 1:32,000 births.

There are more than 300 individuals with Kabuki Syndrome published worldwide, but it is likely that there are many more that are not published. It was first described in 1981 by Niikawa and Kuroki who observed several children with similar characteristics.

There are many features which can occur in Kabuki Syndrome but not all are seen in every child.
Some of the features include:

- Arched, interrupted eyebrows
- Long palpebral fissures
- Large and low-set ears
- Depressed nasal tip
- Short stature
- Skeletal abnormalities such as short fingers, loose joints
- Intellectual disability
- Cleft lip and palate
- Cardiac abnormalities
- Urogenital and kidney problems
- Anorectal and intestinal problems
- Immune abnormalities
- Ear infections and hearing loss
- Hypotonia

In most cases of Kabuki Syndrome, there is no family history of the syndrome. Kabuki Syndrome is found in males and females equally.

There is no cure for Kabuki Syndrome but there is a lot that can be done to ensure good health in a person with Kabuki Syndrome, and to make sure that each person with Kabuki Syndrome achieves their full potential.

Health care professionals that are likely to be involved include a pediatrician, geneticist, and other specialists depending on the problems in the child. Most children will require the input of speech therapists, physiotherapists and other allied health professionals.

Reference: American journal of Medical Genetics 127A:118-127 (2004)
Reviewed by: Dr Sue White—Clinical Geneticist, 2008.
Reviewed by: Dr Tiong Tan—Clinical Geneticist, 2010.

For further information please visit

www.sakks.org