



Genetics Information Sheet

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PO Box 317
St. Leonards NSW 1590



(02) 9926 7324



(02) 9906 7529



genetics @med.usyd.edu.au



www.genetics.com.au

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Please note that this information sheet should be accompanied by genetics fact sheets # 1, 3, 5 and 7.

KABUKI SYNDROME

ALSO KNOWN AS

- KABUKI MAKE-UP SYNDROME
- NIIKAWA-KUROKI SYNDROME

GENERAL INFORMATION ABOUT KABUKI SYNDROME

Doctors in Japan first described kabuki syndrome in 1980. The name “Kabuki” was selected because of the facial resemblance to the make up of actors in Kabuki traditional Japanese theatre.

Kabuki syndrome is a rare condition with the following characteristics:

- Intellectual disability;
- short stature;
- unusual facial features;
- abnormalities of the skeleton;
- unusual skin ridge patterns on the fingers, toes, and palms of the hands and soles of the feet.

The majority of the reported cases of this condition have occurred for no apparent reason (*sporadic*). However several cases have been reported to be inherited in families.

MORE INFORMATION ABOUT KABUKI SYNDROME

The most distinguishing features of Kabuki syndrome are the facial features. The following is a list of these features:

- Long palpebral fissures
- Long/ thick eyelashes
- High/ arched eyebrows
- Cleft or high arched palate
- Lowset/ prominent ears
- Broad and depressed nose tip.

Many patients with Kabuki syndrome have a sideways curvature of the spine called *scoliosis*, a short fifth finger that curves inward, and abnormalities of the vertebrae, hands and hip joint. Short stature and abnormal skin ridge patterns on the fingers, toes, palms of the hands and soles of the feet are also common.

Most children with Kabuki syndrome have a mild to moderate intellectual delay. A few, though they may need assistance with speech and fine motor skills, are able to follow the regular curriculum in school.

Many of the older children (in their early teens) have learned or are learning to read at a functional level. Math skills vary; some do fairly well, while others continue to struggle with this.

Over 50% of people with Kabuki syndrome experience hearing loss. The loss in most cases, however, does not result from repeated ear infections (implying damage to the ear).

Most people with Kabuki syndrome have joint laxity. Hip abnormalities, resulting in hip displacements, are a possibility. There is also a possibility of dislocation of the patella (kneecap), most likely arising because of lax ligaments.

At least 30% of the children with Kabuki have heart defects. For most, this is discovered soon after birth and corrected by surgery. A significant amount of the children have kidney or other urinary tract anomalies.

There are many other manifestations being reported such as wide set nipples, premature breast development, early puberty, lowered immunity, microcephaly, undescended testes, umbilical hernias, inguinal hernias, generalised hirsutism (hairiness) and vitiligo vulgaris (patches of depigmentation).

Not all children manifest all these characteristics. The phenotype or physical characteristics of the condition seem to evolve over time making diagnosis in infancy difficult sometimes.

WHAT CAUSES KABUKI SYNDROME?

The majority of cases of Kabuki syndrome are thought to occur for no apparent reason (*sporadic*). There have been a few reports in the medical literature of families in which the condition appears to be inherited, following a pattern called autosomal dominant inheritance. In these families, it is found that the expression or the symptoms amongst affected family members may vary greatly. This is called variable expressivity.

For more information about genes, chromosomes and the autosomal dominant form of genetic inheritance, please refer to Genetics Fact Sheets 1, 3 and 7.

WHO IS AFFECTED BY KABUKI SYNDROME?

Kabuki syndrome is a rare condition that affects males and females in equal numbers. The majority of cases have been reported in people of Japanese ancestry, however, it is still probably greatly under-diagnosed since the number of medical professionals who are familiar with the syndrome is still growing. Diagnosis is further complicated by the fact that the spectrum of "symptoms" is very diverse. As more and more geneticists become aware of Kabuki syndrome, more children are being diagnosed.

IS THERE ANY TREATMENT FOR KABUKI SYNDROME?

People with Kabuki syndrome may benefit from orthopaedic care and physical therapy for prevention of skeletal problems. Cosmetic surgery may also be helpful for some features. Treatment of biliary, respiratory problems or other problems is symptomatic and supportive. Genetic counselling may be of benefit to individuals with Kabuki syndrome and their family.

For more information about what genetic counselling can offer, please refer to Genetics Fact Sheet # 5.

RESOURCES

General peer support and possible contact with other affected individuals or families may be available from:

Association of Genetic Support of Australasia (AGSA)
66 Albion Street
SURRY HILLS NSW 2010
Phone: (02) 9211 1462 Fax: (02) 9211 8077
E-mail: agsa@ozemail.com.au
Home Page: <http://www.agsa-geneticsupport.com.au>

The following Internet Network Group may be able to provide additional information and support:

Kabuki Syndrome Network
Home Page: <http://www.kabukisyndrome.com/>

For information regarding local genetic counselling services:

The Centre for Genetics Education
PO Box 317
ST LEONARDS NSW 1590
Phone: (02) 9926 7324 Fax: (02) 9906 7529
E-mail: genetics@med.usyd.edu.au
Home Page: <http://www.genetics.com.au>

REFERENCES

This information sheet is based on information available from **NORD** (see below) which was last updated in **2000** and **OMIM** which was last updated in **2006**.

NORD National Organization for Rare Disorders (NORD)
Home Page: www.rarediseases.org

OMIM MENDELIAN INHERITANCE IN MAN ON-LINE:
Home Page: <http://www.ncbi.nlm.nih.gov/Omim>