
Kabuki syndrome

Summary

- Kabuki syndrome is a rare genetic disorder with a range of characteristics, including intellectual disability, distinctive facial features and skeletal abnormalities.
 - There is no cure – treatment aims to reduce the risk of complications and improve quality of life.
 - Since Kabuki syndrome is so rare, there is limited information on the long-term outlook.
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Kabuki syndrome is a rare genetic disorder with a range of characteristics, including intellectual disability, distinctive facial features and skeletal abnormalities. Estimates suggest that Kabuki syndrome occurs in about one in every 32,000 births. However, Kabuki syndrome is thought to be underdiagnosed, so it could be more common.

The condition affects males and females equally, and there is no cure. Kabuki syndrome is also known as Niikawa-Kuroki syndrome.

Symptoms of Kabuki syndrome

Kabuki syndrome has a wide range of characteristics, but not all are present in every child with the condition.

The most common characteristics include:

- distinctive set of facial features, including widely spaced eyes, low-set or prominent ears, exaggerated eyebrow arch, flattened nose tip and a very high palate or cleft palate
- short stature
- skeletal abnormalities, such as scoliosis, short fingers or lax joints
- intellectual disability, ranging from mild to severe.

Other health problems with Kabuki syndrome

Kabuki syndrome may be associated with other health problems, including:

- heart defects such as coarctation (narrowing) of the aorta, ventricular or atrial septal defects (holes between the right and left heart chambers)
- hearing loss
- kidney abnormalities
- dental problems, such as missing or misshapen teeth
- eye problems, such as drooping eyelid (ptosis) or strabismus
- immune system dysfunction, such as the autoimmune disease idiopathic thrombocytopenic purpura (ITP) – a bleeding disorder
- small skull (microcephaly)
- undescended testicles
- ongoing middle ear infections
- epilepsy
- behaviour problems
- early puberty
- substantial weight gain at puberty.

Causes of Kabuki syndrome

Kabuki syndrome is usually caused by a change in the gene *KMT2D* (formerly *MLL2*). Everyone has two copies of the *KMT2D* gene and Kabuki syndrome occurs when only one of the two genes is changed. This is an 'autosomal dominant' inheritance pattern. However, in most children with Kabuki syndrome, it has occurred as a new genetic change, rather than having been inherited from a parent.

A change in the *KMT2D* gene is not identified in all children with suspected Kabuki syndrome. Changes in a second gene, *DKM6A*, are a less common cause of Kabuki syndrome.

Diagnosis of Kabuki syndrome

Kabuki syndrome is difficult to diagnose for three main reasons, being:

- Every child with the condition presents with a slightly different set of characteristics.
- Instead of being present at birth, characteristics may develop over time.
- Many doctors may not be familiar with Kabuki syndrome because the condition is so rare.

A geneticist usually makes the diagnosis based on the child having the distinctive facial features and other signs suggestive of Kabuki syndrome. Genetic testing of the *KMT2D* gene can be used to confirm diagnosis.

Treatment of Kabuki syndrome

Kabuki syndrome is permanent and there is no cure. Treatment aims to reduce the risk of complications and improve quality of life. Options may include:

- Surgical treatment – for example, an operation to correct a heart problem
- medication to manage epileptic seizures
- orthopaedic treatment for scoliosis and joint laxity
- restorative and cosmetic dental treatment
- regular eye examinations and treatment
- physical therapy to strengthen muscles
- occupational therapy to improve fine motor skills
- speech therapy
- sign language training
- social skills training
- behaviour modification therapy
- special education assistance
- cosmetic surgery – for example, to pin back prominent ears.

Long-term outlook for Kabuki syndrome

Since Kabuki syndrome is so rare, there is limited information on the long-term outlook. Issues to consider include:

- One study of three adults with Kabuki syndrome found that while they could manage day-to-day life and hold part-time jobs, they still needed supported care accommodation.
- A person with Kabuki syndrome appears to have a normal life span, but is likely to have ongoing medical problems associated with the condition, which will require medical management.
- People with Kabuki syndrome may suffer from obesity in adolescence or adulthood, which increases the risk of a range of health problems, including cardiovascular disease and diabetes. Weight management is important.

Genetic counselling and Kabuki syndrome

If your child or another family member has been diagnosed with Kabuki syndrome, or if the condition runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Kabuki syndrome and what causes it, how it is inherited, and what a diagnosis means for your child's health and development, and for your family. Genetic counsellors are

trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If Kabuki syndrome runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by Kabuki syndrome.

Where to get help

- Your **GP (doctor)**
- **Paediatrician**
- **Victorian Clinical Genetics Services (VCGS)**, Royal Children's Hospital Tel. **1300 118 247**
- **Genetic Support Network of Victoria** Tel **(03) 8341 6315**
- **Genetic Alliance Australia** Tel. **(02) 9295 8359**
- Genetic counselling services – available at most large public maternity hospitals
- The **Centre for Genetics Education** Tel. **(02) 9462 9599**
- **Better Start for Children with a Disability** Tel. **1800 242 636**

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