

Kabuki syndrome

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. The condition is also known as Niikawa-Kuroki syndrome.

Symptoms

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

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.

The cause of Kabuki syndrome

Kabuki syndrome usually is caused by a fault in the gene MLL2. Everyone has two copies of the MLL2 gene and Kabuki syndrome occurs when one of the two genes is faulty. Kabuki syndrome is inherited as an autosomal dominant disorder. However, in most children with Kabuki syndrome, it has occurred as a new genetic change, rather than having been inherited from a parent.

A fault in the MLL2 gene is not identified in all children with suspected Kabuki syndrome. In these children, it may be that the diagnosis is incorrect, or alternatively there may be a second gene for Kabuki syndrome that has not yet been discovered.

Diagnosis

Kabuki syndrome is difficult to diagnose for three main reasons:

- 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 MLL2 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 defect
- 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

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 the people 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.

Where to get help

- Your doctor
- Paediatrician
- Genetic counselling services – available at most large public maternity hospitals
- Genetic Health Services Victoria, Royal Children's Hospital Tel. (03) 8341 6200
- Supporting Aussie Kids with Kabuki Syndrome (SAKKS)
- Association of Genetic Support of Australasia Tel. (02) 9211 1462
- The Centre for Genetics Education Tel. (02) 9462 9599

Things to remember

- 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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