



KABUKI SYNDROME FAQ

What is Kabuki syndrome?

Kabuki syndrome is a rare genetic disorder occurring in approximately 1 in 32,000 births worldwide. Individuals affected by Kabuki experience a variety of symptoms which may include: mild to moderate intellectual impairment, growth delays, low muscle tone, feeding difficulties, heart defects, cleft palate, skeletal abnormalities, vision and hearing impairments, communication delays and autistic-like behaviors.

Who can diagnose Kabuki syndrome?

A Geneticist. Kabuki syndrome can be clinically diagnosed via physical features, or genetically diagnosed through blood work.

Is Kabuki syndrome contagious?

No! Kabuki syndrome is a genetic disorder, it cannot be spread to others.

Is Kabuki syndrome a progressive disorder?

Kabuki syndrome is not considered a progressive disorder, however new complications may present over time.

Can a child outgrow Kabuki syndrome?

No. Kabuki syndrome is a genetic disorder. Early intervention and treatment may lessen complications associated with Kabuki, but it is a lifelong disorder.

Is Kabuki syndrome a fatal disorder?

Kabuki syndrome does not shorten an individual's life span, however underlying conditions may.

What will my child's future look like?

While the effects of Kabuki syndrome vary from individual to individual, many children and adults share similar underlying diagnoses. The impact of each diagnosis varies from person to person, as does their future.

Can an individual with Kabuki syndrome live independently as an adult?

The physical and cognitive effects of Kabuki syndrome affect individuals differently. Some higher functioning adults have gone on to marry and have children.

If I have more children, will they have Kabuki syndrome too?

If the mutation is de novo (new - the parents are not carriers) there is a 1% chance. If either parent is a carrier, or has Kabuki syndrome, there is a 50% chance.

Do all children with Kabuki syndrome experience developmental delays?

Most, but not all! Many parents report delays in feeding (eating by mouth vs feeding tube), sitting up, crawling, walking, and talking.

For more information please visit our website:

www.AllThingsKabuki.org

Can children with Kabuki syndrome play sports?

Possibly! Because Kabuki syndrome affects each individual differently, it depends on the child. Some children with complex medical needs are not physically able to play sports, while others have successfully kept up with their peers in a variety of sports.

Do all children with Kabuki syndrome require therapeutic intervention?

While Kabuki syndrome affects each individual differently, early intervention has proven to play a vital role in developmental progress for many. Therapy services may include: Occupational, Speech, Physical, Vision and Feeding.

Can children with Kabuki syndrome attend regular classes in school?

Absolutely! Cognitive delays vary with each child so it is important that an IEP (Individualized Education Plan) or 504 Plan be established ahead of time. These plans ensure that a child has the appropriate accommodations in place, to allow them the best possible educational experience. Children with Kabuki syndrome can attend mainstream classes, special education classes or a combination of both. Additionally, some individuals may require one-on-one aides, while others may not.