

Kabuki syndrome is a rare genetic disorder first described in 1981 by Japanese researchers, Dr. Norio Niikawa and Dr. Yoshikazu Kuroki.^{1,2} Dr. Niikawa noted similarities between the facial features of those with Kabuki syndrome and the make-up used in traditional Japanese Kabuki Theater and called the condition Kabuki make-up syndrome (KMS). Kabuki syndrome has been referred to as KMS, Niikawa-Kuroki syndrome, and Kabuki syndrome.

In 2010, researchers at the University of Washington discovered the first gene responsible for Kabuki syndrome: *KMT2D*, formerly known as MLL2.³ In 2012, *KDM6A* was confirmed as the second gene to cause Kabuki syndrome.⁴ Variants (or spelling changes) causing Kabuki syndrome on these genes usually occur randomly, but Kabuki syndrome can be inherited (passed from parents to children).

Kabuki syndrome occurs in approximately 1:32,000 births and is believed to occur equally across all ethnicities and both sexes. Each person with Kabuki syndrome is unique. Symptoms and their severity can vary widely between individuals. Some of the most common symptoms include^{5, 6, 7}:

Brain

- Mild-to-moderate intellectual disability
- Speech difficulties
- Poor coordination and visual-spatial awareness
- Anxiety and obsessive behaviors
- Sleep disturbances
- Seizures

Muscles/Skeleton

- Persistence of fetal finger pads
- Low muscle tone
- Joint hypermobility
- Growth deficiency
- Skeletal abnormalities such as scoliosis

Gastrointestinal/Renal/Immune

- Feeding difficulties including reflux
- Kidney abnormalities
- Endocrine disorders
- Immune deficiencies

Head/Face

- Hearing loss
- Blue sclerae, eye and vision disorders
- Cleft lip and/or palate
- Missing and/or widely spaced teeth
- Long opening between the eyelids
- Short nose with a depressed tip
- Large or cupped ears

Heart

- Congenital heart defects

Infants usually have normal growth measurements at birth but often later exhibit "failure to thrive" due to a combination of symptoms such as poor growth and trouble feeding.

Join us! Scan the QR code to visit
www.KabukiSyndromeFoundation.org,
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