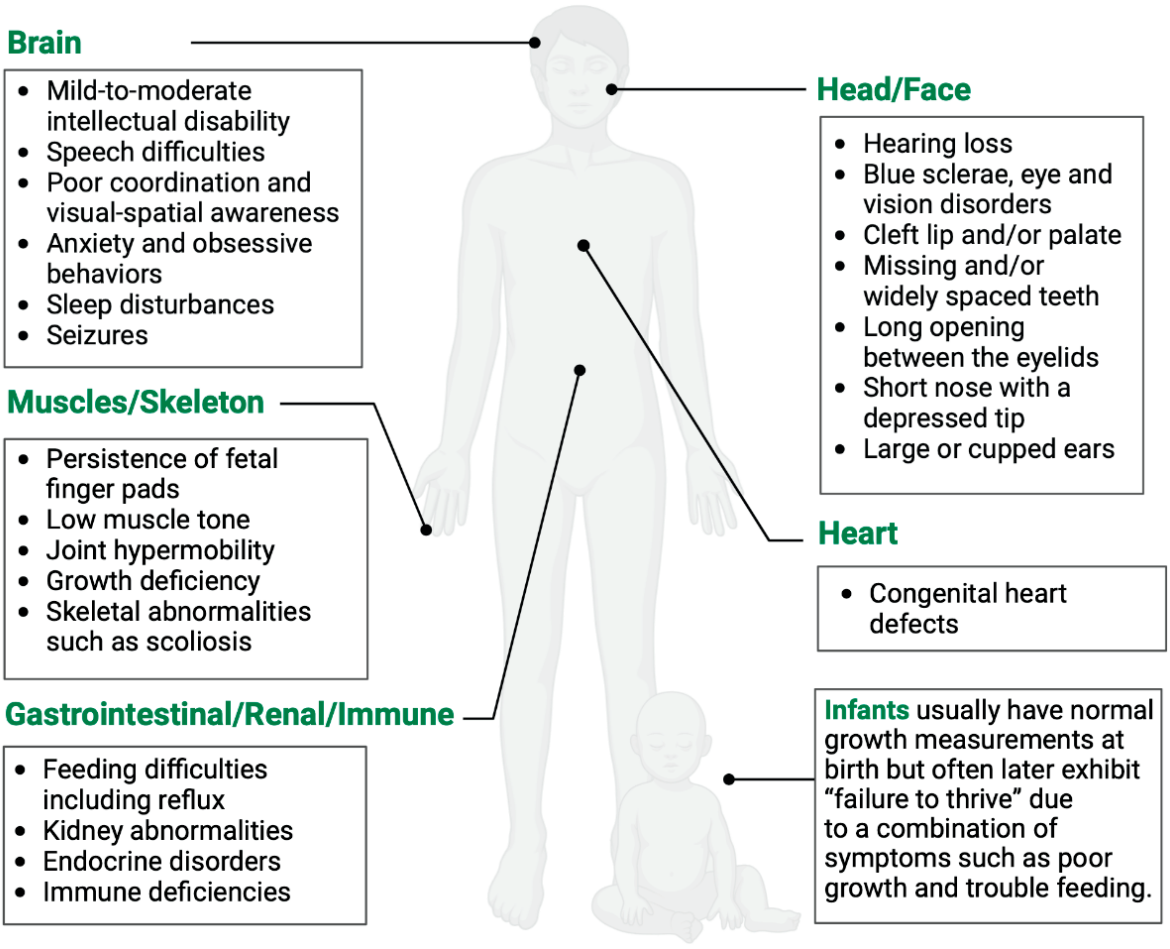
Common Symptoms of Kabuki Syndrome

Kabuki syndrome is a rare genetic disorder that affects an estimated 1 in 32,000 births across genders, races, and ethnicities. Kabuki syndrome is caused by mutations in two known genes (KMT2D and KDM6A), and possibly others yet to be identified. The gene mutation usually occurs randomly, but Kabuki syndrome can be passed on by a parent to their child.[3, 4, 5]

Each person with Kabuki syndrome is unique. Symptoms and their severity can vary widely between individuals. Some symptoms can be present at birth, while others can appear later in life. The most common symptoms include:[5, 6, 7]



Kabuki syndrome was first described in 1981 by Japanese researchers, Dr. Norio Niikawa and Dr. Yoshikazu Kuroki. 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.[1, 2]

More resources including recommended evaluations, cited publications, and current research opportunities are available at www.KabukiSyndromeFoundation.org