KABUKI SYNDROME

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SYNDROME PROFILE

***Abstract**: Kabuki Syndrome is a congenital disorder present from the birth. It affects 1 in 32,000 Japanese people. It is caused by heterozygous mutation that results in an uncommon systemic progression of the disease. A peculiar facial dysmorphia with dwarfism and varying degrees of mental retardation are the defining characteristics of this syndrome.*

Kabuki syndrome is a rare multisystem disorder characterized by multiple abnormalities including distinctive facial features, growth delays, varying degrees of intellectual disability, skeletal abnormalities and short stature.[1]

HISTORY, ETYMOLOGY & SYNONYMS

Kabuki syndrome was first reported in medical literature in 1981 by Japanese physicians. The name of the syndrome takes inspiration from a form of Japanese theatre called Kabuki, whose actors have a peculiar make-up that resemble the facial features of the syndrome. Kabuki syndrome is also known as Kabuki make-up syndrome, KMS or Nikawa Kuroki Syndrome.[1]

Figure 1 Depiction of an actor of the Kabuki theatre

ETIOLOGY

The onset of Kabuki syndrome is determined by the mutation of one of the two genes – KMT2D and KDM6A. The latter accounts for fewer cases of the syndrome.[1]

CLINICAL FINDINGS

Features of Kabuki syndrome are present at birth (congenital), while other features become apparent as an affected child ages. ec

FACIAL MANIFESTATIONS

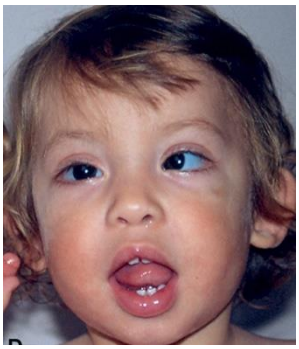

Figure 2 Features of Kabuki Syndrome

Children with Kabuki syndrome have distinct facial appearance, which includes abnormally long openings between the eyelids (palpebral fissures), lower eyelids that are turned outwards (everted), prominent eyelashes, arched eyebrow, a broad nose with flattened or depressed tip and large misshaped ears.[2]

Additional features include a bluish tinge to the whites of the eyes (blue sclerae), drooping of the upper eyelid (ptosis), misaligned eyes (strabismus). Some children may also exhibit seizures, diminished muscle tone (hypotonia) and microcephaly.[3]

ORAL MANIFESTATIONS

A high arched roof of the mouth or a cleft palate, depression involving the inside of the lower lips (lip pits) and abnormally small jaw (micrognathia). Speech delays are not uncommon. These may be as a result of palatal abnormalities and hearing loss.[2]

Growth deficiency is common in individuals with Kabuki syndrome usually becoming apparent during the first year of life (postnatal growth deficiency). In rare cases, children may have partial growth deficiency. In addition, children with Kabuki syndrome can have mild to moderate intellectual disability. Seizures can develop right after birth (neonatal period) or as late as 12 years of age.[2 3]

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