DISEASE:
Kabuki syndrome

NAME:	Kabuki syndrome
DESCRIPTION:	A rare multiple congenital anomalies/neurodevelopmental disorder characterized by five major features: intellectual disability (typically mild to moderate), visceral malformations (frequently congenital heart defects), persistence of fetal fingertip pads, post-natal short stature, skeletal anomalies (brachymesophalangy, brachydactyly V, spinal column abnormalities and fifth digit clinodactyly) and specific facial features (arched and broad eyebrows, long palpebral fissures, eversion of the lower eyelid, large prominent, cupped ears, depressed nasal tip and short columella). Various additional features are frequently observed.
ORPHACODE:	2322
SYNONYMS:	Kabuki make-up syndrome Niikawa-Kuroki syndrome
XREF(S):	Orphanet MeSH OMIM OMIM MedDRA ICD-10

ANALYTE(S):	KMT2D KDM6A
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RELATED CONTENT

Related Genetic Tests

- Kabuki syndrome (gene panel)
- Short Stature (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- lysine demethylase 6A
- lysine methyltransferase 2D

Related Gene Panels

- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Kabuki (7 genes) - IPG
- Short Stature (46 genes) - IPG