## Opis choroby *

Definicja

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.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Kabuki make-up syndrome
Zespół makijażu Kabuki
Zespół Niikawa i Kuroki
Niikawa-Kuroki syndrome

**Kod ORPHA**
2322

**Kod OMIM**
300867

**Kod ICD10**
Q87.0

**Kod ICD11**
LD2F.1Y

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*Źródło

orphanet