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