Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by severe developmental delay and intellectual disability, generalized hypotonia, growth failure, hydronephrosis, cardiac anomalies, and dysmorphic craniofacial features (such as microcephaly, hypertrichosis, synophrys, long eyelashes, epicanthus, flat nasal bridge, short, upturned nose, long philtrum, low-set ears, open-mouth appearance, full lower lip, cleft palate, and webbed neck). Thin corpus callosum, tethered spinal cord, intestinal malrotation, anal stenosis, and uterus didelphys have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
2729

Kod OMIM
604916

Kod ICD10
Q87.8

Kod ICD11
LD2Y

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*Źródło

orphanet