

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

*Źródło

orphanet