Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by developmental delay, neuropathic visceral dysmotility (resulting in neurogenic megacystis and sometimes chronic intestinal pseudo-obstruction syndrome), intracerebral calcifications, and dysmorphic facial features (including broad forehead, downslanted palpebral fissures, strabismus, protruding and low-set ears, and retrognathia). Microcephaly and renal abnormalities have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
73246

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet