Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome usually characterized by microcephaly, ocular anomalies such as microphthalmia, and apple-peel intestinal atresia. Facial dysmorphism is reported in some cases and may include narrow or sloped forehead, hypertelorism, microphthalmia, dysplastic, edematous deep-set eyes, short palpebral fissures, large or low set ears, broad nasal root, anteverted or broad nasal tip, long philtrum, micrognathia, thin upper vermillion, large mouth and skin tag on the cheek. Motor delay and intellectual disability have been reported. Heart, brain, craniofacial abnormalities, renal hypoplasia and other anomalies (e.g. lower limb edema, thrombocytopenia) are variably present. Rarely, cases without intestinal atresia, microcephaly or developmental delay can be found. Severe lethal cases have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Apple-peel intestinal atresia-ocular anomalies-microcephaly syndrome
Jejunal atresia-microcephaly-ocular anomalies syndrome
Apple-peel intestinal atresia-ocular anomalies-microcephaly syndrome
Jejunal atresia-microcephaly-ocular anomalies syndrome

Kod ORPHA
506307

Kod OMIM
243605

Kod ICD10
Q13.8

Kod ICD11
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*Źródło

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