

Opis choroby *

Definicja

Thakker-Donnai syndrome is a rare, genetic, lethal, multiple congenital anomalies/dysmorphic syndrome characterized by facial dysmorphism (including long, downward slanting palpebral fissures, hypertelorism, posteriorly rotated ears, broad nasal bridge, short nose with a bulbous tip and anteverted nares, downturned corners of the mouth) as well as vertebral (occult spina bifida, hemivertebrae), brain (ventricular dilatation, agenesis of corpus callosum), cardiac (tetralogy of Fallot, ventricular septal defect) and gastrointestinal (short esophagus with intrathoracic stomach, small intestine, spleen and pancreas, anal atresia) malformations. There have been no further descriptions in the literature since 1991.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Dysmorphism-multiple structural anomalies syndrome Zespół Thakker'a i Donnai

Kod ORPHA	Kod OMIM	Kod ICD10
1780	227255	Q87.8

Kod ICD11

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*Źródło

orphanet