

Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by epiphyseal and vertebral dysplasia and abnormalities of the external ears (severe microtia or anotia) and the nose (hypoplastic nose with bifid tip, triangular nares, or anteverted nares). Additional variable findings include short stature, localized aplasia cutis, hypodontia, synophrys, agenesis of the corpus callosum, and cardiac, gastrointestinal, and/or urogenital malformations, among others. Psychomotor development may be delayed.

Dane

Klasyfikacja

Zespół wad wrodzonych Epiphysial-vertebral-ear dysplasia-nose-plus associated findings syndrome

Synonimy

Zespół dysplazji nasadowo-kręgosłupowo-usznej, płaskiego nosa plus/oraz towarzyszących wad

Kod ORPHA

496751

Kod OMIM

616854

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet