Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by the association of Pierre Robin Sequence (congenital micrognathia and glossoptosis with airway obstruction and a U-shaped cleft of the soft palate) with joint contractures and developmental delay. Additional variable manifestations include talipes equinovarus, arachnodactyly, radioulnar synostosis, severe hip dysplasia, cardiac anomalies, facial dysmorphism such as crumpled ear helices, and ocular abnormalities, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
5q23 microdeletion syndrome
Zespół mikrodelecji 5q23

Kod ORPHA
436003

Kod OMIM
-

Kod ICD10
Q87.0

Kod ICD11
-

---

*Źródło

orphanet