Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by congenital pterygia (webbing) mainly affecting the neck and large joints, arthrogryposis multiplex, short stature, and craniofacial dysmorphism (including ptosis, downslanting palpebral fissures, high-arched palate, and retrognathia). Additional manifestations are decreased movements, facial weakness, respiratory distress, vertebral anomalies, scoliosis, anomalies of the fingers, and cryptorchidism, among others. The disease is a non-lethal variant of multiple pterygium syndrome.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Autosomal recessive non-lethal multiple pterygium syndrome
Autosomalny recesywny nieletalny zespół mnogich płetwistości
EVMPS
Zespół Escobara
Zespół mnogich płetwistości wariant Escobara
EVMPS
Escobar syndrome
Escobar variant multiple pterygium syndrome

Kod ORPHA
2990

Kod OMIM
265000

Kod ICD10
Q79.8

Kod ICD11
LD26.40

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*Źródło

orphanet