

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	Synonimy
Zespół wad wrodzonych	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	Kod OMIM	Kod ICD10
2990	265000	Q79.8

Kod ICD11
I D26.40

* Źródło

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