Opis choroby *

Definicja

Otofaciocervical syndrome is a rare, genetic developmental defect during embryogenesis syndrome characterized by distinct facial features (long triangular face, broad forehead, narrow nose and mandible, high arched palate), prominent, dysmorphic ears (low-set and cup-shaped with large conchae and hypoplastic tragus, antitragus and lobe), long neck, preauricular and/or branchial fistulas and/or cysts, hypoplastic cervical muscles with sloping shoulders and clavicles, winged, low, and laterally-set scapulae, hearing impairment and mild intellectual deficit. Vertebral defects and short stature may also be associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Fara-Chlupackova syndrome
Zespół Fara i Chlupackova
Zespół OFC
OFC syndrome

Kod ORPHA
2792

Kod OMIM
615560

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet