

## 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 Fara-Chlupackova syndrome

#### Synonimy

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