DISEASE:
Otofaciocervical syndrome

NAME:	Otofaciocervical syndrome
DESCRIPTION:	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.
ORPHACODE:	2792
SYNONYMS:	Fara-Chlupackova syndrome OFC syndrome
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	EYA1 PAX1
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/1008

RELATED CONTENT

Related Genetic Tests

- Cataract (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- EYA transcriptional coactivator and phosphatase 1
- paired box 1

Related Gene Panels

- Cataract - UGent
- test