
DISEASE:
Branchiootic syndrome

NAME:	Branchiootic syndrome
DESCRIPTION:	Branchiootic syndrome is a rare, genetic multiple congenital anomalies syndrome characterized by second branchial arch anomalies (branchial cysts and fistulae), malformations of the outer, middle and inner ear associated with sensorineural, mixed or conductive hearing loss, and the absence of renal abnormalities. Typical ear findings consist of malformed auricles (e.g. lop or cupped ears), preauricular pits and/or tags, and middle and/or inner ear dysplasias (including cochlear, vestibular and semicircular channel hypoplasia, malformation of the ossicles and of middle ear space).
ORPHACODE:	52429
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM
ANALYTE(S):	EYA1 SIX1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/2984>

RELATED CONTENT

Related Genetic Tests

- [Cataract \(gene panel\)](#)
- [Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophthisis / Bardet-Biedl syndromes and kidney cancers \(gene panel\)](#)
- [Renal or urinary tract malformation \(CAKUT\) \(gene panel\)](#)

Related Laboratories

- [Centre de Génétique-Institut de Pathologie et de Génétique \(IPG\)](#)
- [Centrum Medische Genetica - UZ Gent](#)

Related Analytes

- [EYA transcriptional coactivator and phosphatase 1](#)
- [SIX homeobox 1](#)

Related Gene Panels

- [Cakut \(congenital anomalies of the kidney and urinary tract-1\) \(69 genes\) - IPG](#)
- [Cataract - UGent](#)
- [Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophthisis, Bardet-Biedl syndromes and kidney cancers \(146 genes\) - IPG](#)
- [test](#)