DISEASE:
BOR syndrome

NAME:	BOR syndrome
DESCRIPTION:	A rare otomandibular dysplasia syndrome characterized by branchial arch anomalies (branchial clefts, fistulae, cysts), malformations of the ear associated with hearing impairment (malformations of the auricle with pre-auricular pits, conductive or sensorineural hearing impairment), and renal malformations (urinary tree malformation, renal hypoplasia or agenesis, renal dysplasia, renal cysts).
ORPHACODE:	107
SYNONYMS:	Branchiootorenal spectrum disorder Branchiootorenal syndrome Melnick-Fraser syndrome
XREF(S):	Orphanet MeSH OMIM OMIM ICD-10 MedDRA
ANALYTE(S):	SIX1 EYA1 SIX5

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RELATED CONTENT

Related Genetic Tests

- Cataract (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Craniosynostosis (gene panel)
- Renal or urinary tract malformation (CAKUT) (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- EYA transcriptional coactivator and phosphatase 1
- SIX homeobox 1
- SIX homeobox 5

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, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Craniosynostosis (32 genes) - KUL
- test

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