branchio-oto-renal syndrome

Eponyms:
- BOP
- BOR syndrome
- BOS
- branchio-otic syndrome
- EYA1
- Melnick-Fraser syndrome

Inheritance: autosomal dominant

Semiological Synthesis: Facio-oto-urological disorder. Preauricular pits on the helix or ear tags, branchial cysts or fistulas internal/external opening, renal hypoplasia/agenesis. Frequency: one out of 40,000 live births.

Group Sub group Signs:

AUDITORY DISORDERS
- deafness
  - cholesteatoma, keratoma
  - deafness conductive type
  - deafness sensorineural, including unspecified type
  - deafness, in syndromic association

DERMATOLOGICAL DISORDERS
- cutis lesions, not including nodules-blistering
  - cutis, fistulas

LABORATORY DATA
- chromosomal assignment
  - chromosome 8q localization
- gene, structural-functional anomalies
  - BOR (EYA1) eyes absent, drosophila, human homolog, gene chr.8q13.3
  - gene analysis-DNA analysis

NEUROLOGICAL DISORDERS
- cranial neuropathy
  - facial palsy

OCULAR DISORDERS
- lacrimal gland-sac, defects
  - duct defect, puncta defects, lacrimal canalis defect, tear duct defects
  - epiphora

OROCRANIOFACIAL ANOMALIES
- external ear malformations
  - ear lobe, cup shaped
  - ear lobe, hypoplastic, small, poorly shaped, dysplastic pinnae
  - ear lobe, malformed, unspecified type
  - ear, external canal atresia, stenosis
  - ear, prominent; ear lobe, protruding
  - preauricular pits,
  - facies, modified appearance
    - facies, asymmetric, unilateral atrophy, hemifacial atrophy
    - neck, modified appearance
    - neck, fistula

OTHERS
- inheritance
  - inheritance, autosomal dominant
- supergroups
  - cutaneous-facio-neuro-oculo disorders
  - cutaneous-facio-oto-urological disorders
  - facio-neuro-oco-uro disorders

PRENATAL-NEONATAL MODIFIED DATA
- foetal changes
- foetal changes, recognized by laboratory data
- foetal changes, recognized by ultrasound techniques

**prenatal diagnosis**
- prenatal diagnosis, echographic

**UROLOGICAL DISORDERS**

**kidney, malformations**
- kidney, malformation, including horseshoe kidney
- kidney, small, underdeveloped, including renal agenesis

**renal dysfunctions**
- renal failure, including: nephritis, pyelonephritis, glomerulonephritis

**urinary tract-bladder, malformations**
- urinary reflux, vesicoureteral reflux
- urinary tract duplication, bladder duplication
- urinary tract malformations, unspecified type

**Super group:**
cutaneous-facio-neuro-ocular disorders
cutaneous-facio-oto-urological disorders
facio-neuro-oculo-oto disorders
oto-urological disorders

**Super aggereg. Aggregations:**
DEAFNESS
defauness, in syndromic association

**FOETAL CHANGES**
foetal changes, recognized by laboratory data
foetal changes, recognized by ultrasound techniques

**Differential diagnosis:**
28464 BOR-Duane-hydrocephalus contiguous gene syndrome
3760 branchial arch defects Aksu type
3765 branchial arch defects X-linked
3770 branchial cleft anomalies
3780 branchio-ocular-facial syndrome
3790 branchio-oto syndrome
3802 branchio-oto-ureteral syndrome
6830 cryptomicrotia-brachydactyly syndrome
13960 kidneys small syndrome
14510 Lee-Nicholson-Hitchcock syndrome
15725 Marres-Cremers syndrome
27650 preauricular pits-renal disease syndrome
333 renal agenesis, unilateral
25680 Townes-Brocks syndrome

**Bibliography**
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Smith's Recognizable Patterns of Human Malformation. 6th Edition pag.272-273
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