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Bilateral Preauricular Pits

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The Case

A newborn presents with bilateral preauricular pits (Figures 1, 2)

Prenatal and Birth History

- 22-year-old G2P1 African American mother
- Estimated gestational age: 38 weeks
- Spontaneous vaginal delivery in hospital
- Nonconsanguineous parents
- Routine antenatal screen negative
- Apgar scores 9 and 9 at 1 and 5 minutes.
- Birthweight: 3,063 g

Presentation

The infant was active, feeding well, and vital signs were stable. However, the findings on physical examination prompted admission to the neonatal intensive care unit for further evaluation.

Case Progression

Vital Signs

- Temperature: 98.7°F (37.1°C)
- Heart rate: 120 to 150 beats/min



Figure 1.

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Figure 2.

- Respiratory rate: 30 to 50 breaths/min
- 99% oxygen saturation on room air
- Blood pressure on right hand by cuff method

Physical Examination

- Appropriately grown for gestational age
- Head: normocephalic; normal, open, flat fontanelles; symmetric facies; patent nares; intact palate
- Ears: bilateral preauricular pits; normal shaped pinna bilaterally
- Neck: sinus approximately 1 mm in diameter over the medial border of the right sternocleidomastoid muscle. A 2-mm skin tag also was noted over the medial border of the left sternocleidomastoid muscle
- Oral cavity: pink mucosa, intact palate, no lymphadenopathy, normal sucking and rooting reflex



Figure 3.

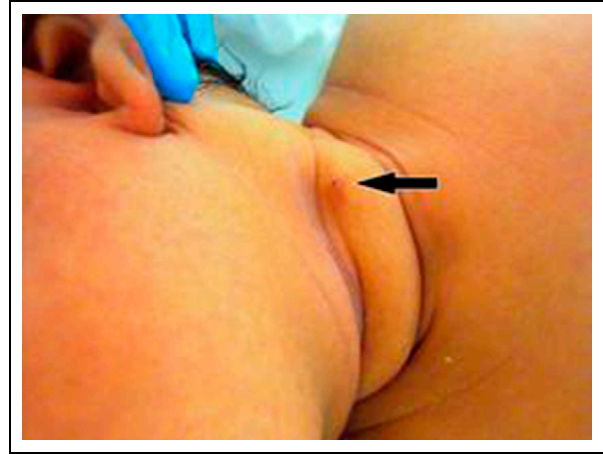


Figure 4.

- Lungs: clear, equal breath sounds; no respiratory distress
- Cardiovascular: normal S1, S2; regular rate and rhythm; no murmurs or gallops
- Abdomen: soft, nondistended, no organomegaly, normal bowel sounds
- Genitourinary: normal term female genitalia; patent anus
- Skeletal: spine appears normal
- Skin: no icterus, birthmarks, or other rashes
- Neurologic: symmetric Moro, normal strength and tone

Family History

The infant's mother was noted to have bilateral preauricular pits and a sinus over the medial border of the left sternocleidomastoid muscle. She also had mild deafness bilaterally. She recollected that her father too had bilateral preauricular pits and a sinus on the left side of his neck.

Differential Diagnosis

- Branchiooculofacial syndrome
- Branchio-oto-renal (BOR) spectrum disorder
- Mandibulofacial dysostosis
- Oculoauriculovertebral dysplasia

Actual Diagnosis BOR Spectrum Disorder

The Experts

Branchial cleft anomalies account for almost 30% of congenital neck masses. They can present as cysts, sinuses, or

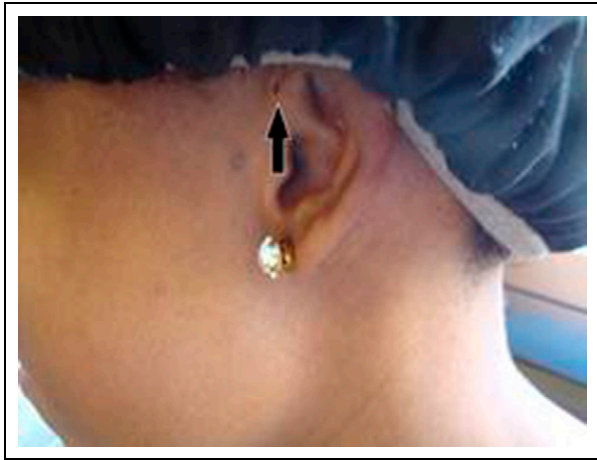


Figure 5.

fistulae. There is no sex predilection, and they may present later in life. Differential diagnoses of branchial cleft anomalies include adenopathy, thyroglossal duct anomalies, lymphatic malformations such as cystic hygroma, dermoid cysts, and cervical clefts. In our patient, the anatomical position of the sinus suggested a branchial cleft anomaly.

Branchial Cleft Anomalies

By the fourth week of gestation, the human embryo has four well-defined arches. Each arch is lined externally by ectoderm and internally by endoderm, with mesoderm in between. Each arch gives rise to specific anatomical parts. Each arch is separated by clefts externally. Branchial anomalies are thought to be the result of incomplete obliteration of the clefts. Branchial cleft anomalies of the first arch account for 1% of the branchial cleft anomalies. These can present as cysts, sinuses, or fistulae located between external auditory canal and the submandibular area. Second branchial cleft anomalies are the most common, accounting for 95% of all branchial cleft anomalies. They present as cysts, sinuses or fistulae in the lower anterolateral neck. Cysts are more common than the rest. An internal opening from a second branchial arch, if present, is located in the tonsillar fossa. Branchial cleft anomalies of the third arch have an external opening at a similar anatomical location as the second cleft anomalies. However, the internal opening is at the piriform fossa. Both third and fourth cleft anomalies normally contain thymic tissue. Anomalies of third and fourth branchial cleft are rare.

Branchio-oto-renal Spectrum Disorders

BOR spectrum disorders include BOR syndrome and branchiootic syndrome (BOS). BOR syndrome, also known as Melnick-Fraser syndrome, is an autosomal



Figure 6.

dominant disorder consisting of second branchial arch anomalies with hearing loss and renal anomalies. The disorder is thought to have 100% penetrance but with variable expressivity. About 10% of the cases have been found to be due to de novo mutations. The true incidence of this disorder is unknown but is possibly between 1 in 40,000 and 1 in 700,000 based on two studies. BOR syndrome may be responsible for almost 2% of profoundly deaf children. Diagnosis of BOR syndrome can be made if three or more major criteria or two major and two minor criteria are present. Only one major criterion is needed in individuals with affected family members.

Major criteria include second branchial arch anomalies, deafness, preauricular pits, auricular deformity, and renal anomalies. Minor criteria include external auditory canal anomalies, middle ear and inner ear anomalies,



Figure 7.

Table. Clinical Features of Branchiootorenal Spectrum Disorders

Otologic features	Preauricular pits; preauricular tags Lop-ear deformity or cup-ear deformity of pinnae Atresia or stenosis of external auditory meatus Malformation, dislocation, or fixation of middle ear ossicles Cochlear hypoplasia Enlargement of cochlear and vestibular aqueducts Hypoplasia of semicircular canals
Second branchial cleft anomaly	Branchial cleft cyst, sinus, or fistulae
Renal anomalies	Renal agenesis/hypoplasia Uretero-pelvic obstruction Calyceal cyst/diverticulum Ectopic ureter; hypospadias Hydronephrosis/vesicoureteral reflux Hypoplastic bladder
Other findings	Lacrimal duct aplasia Cleft palate Retrognathia Euthyroid goiter Facial nerve paralysis Gustatory lacrimation
EYA1, SIX5, and SIX1 are the three genes associated with BOR spectrum disorders. EYA1 mutations account for 40% of the individuals, whereas SIX5 and SIX1 mutations account for 2.5% and 2%, respectively.	

preauricular tags, facial asymmetry, and palate abnormalities. BOS has clinical features similar to BOR syndrome but without structural renal anomalies. Hearing loss in BOR syndrome and BOS can be conductive, sensory, or mixed. Both BOR syndrome and BOS can occur within the same family. The multitude of clinical features described in BOR spectrum disorders are summarized in the Table.

Management

Affected individuals require an individualized care plan with a multidisciplinary team involving a geneticist, otolaryngologist, speech therapist, occupational therapist, radiologist and renal specialist, with the pediatrician coordinating the care.

Second branchial cleft anomalies should be investigated by imaging studies. Computed tomography scan is the preferred modality. The anomalies are associated with a high risk of infection. Complete surgical excision

of the branchial anomaly is the definitive treatment. Auditory functions must be thoroughly assessed and close surveillance must be carried out. Any hearing loss should have prompt intervention, and a supportive educational program must be instituted. Screening for associated renal anomalies includes imaging and renal function tests. Genetic counseling regarding the natural history, inheritance, risks, and screening of other family members should be provided. Pregnant women with a family history of BOR syndrome may be offered prenatal ultrasound at 16 to 17 weeks gestation to evaluate renal malformations in the fetus. Siblings of the affected individuals are recommended to have hearing evaluation and assessment of renal status. Although molecular genetic testing is available, gene analysis may be deferred if the affected individual has the clinical features and family history consistent with BOR spectrum disorder.

American Board of Pediatrics Neonatal-Perinatal Content Specifications

- Know the clinical manifestations of branchial cleft cysts



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