The Journal of Laryngology & Otology
January 2003, Vol. **117,** pp. 32–38

Main Articles

External auditory canal duplication anomalies associated with congenital aural atresia

Nikolas H. Blevins, M.D., Seema V. Byahatti, M.D., Collin S. Karmody, M.D.

Abstract

Maldevelopment of the first branchial cleft can produce a broad spectrum of anomalies in its derivative structure, the external auditory canal (EAC). Failure of the cleft to develop normally can result in either the absence of a normally patent EAC (atresia, or stenosis) or a duplication anomaly (cyst, sinus, or fistula). Despite their common origins, the coexistence of these anatomical abnormalities is quite unusual. We present four patients with both aural atresia and duplication anomalies of the EAC. Three patients had non-syndromic unilateral aural atresia and presented with periauricular lesions originating from the first branchial cleft. The other patient had a variant of Treacher Collins syndrome and presented with draining infra-auricular fistulae.

The classification and management of first branchial cleft anomalies is reviewed in light of these cases. An understanding of the embryogenesis of the external ear is necessary to successfully recognize and treat this spectrum of deformities. A classification system is presented that encompasses the full spectrum of first cleft anomalies.

Key words: Ear; Abnormalities; Ear Canal; Branchial Region; Embryology

Introduction

Numerous head and neck structures have their origins in the branchial apparatus. Abnormal development of the branchial system can result in a spectrum of congenital anomalies including the absence, maldevelopment, or duplication of an anatomical structure. Most commonly, abnormalities of branchial origin result in persistent cysts, sinuses and fistulae. Of the branchial clefts, only the first cleft normally persists through embryogenesis, developing into the EAC and squamous layer of the tympanic membrane. Anomalies related to the development of the first cleft are rare and can result in a wide range of external ear malformations. It is possible that more than one type of first cleft anomaly can co-exist. In such cases, an incompletely formed EAC can be present along with a congenital cyst, sinus, or fistula. Combinations of such anomalies may potentially present a diagnostic and therapeutic challenge. We have encountered four patients who presented with anomalous epithelial-lined spaces in conjunction with congenital aural atresia. This rare occurrence can provide insight into the embryogenesis of the EAC from the first branchial cleft.

Case reports

Case 1

A 29-year-old white male presented with a three-week history of right otalgia and post-auricular swelling. He had a history of a congenital atretic right ear canal and pre-auricular skin tag. He had the expected conductive hearing loss associated with the atresia, and no other otological history. When the otalgia began, he was initially treated elsewhere with both oral and intravenous antibiotics that failed to relieve his symptoms. He subsequently developed an area of granulation tissue in the superior aspect of his atresia pouch, and a punctum that drained purulent material when pressure was applied to an area of post-auricular fluctuance. He also developed mild trismus. An audiogram documented at 60 dB conductive hearing loss.

A computed tomography (CT) scan was obtained that demonstrated a bony atresia plate and a well-aerated middle ear. There was no evidence of middle-ear fluid or cholesteatoma (Figure 1). A gadolinium-enhanced magnetic resonance image (MRI) was obtained that demonstrated a right postauricular cyst with a tract running into the

From the Department of Otolaryngology/Head and Neck Surgery, New England Medical Center, Tufts University School of Medicine, Boston, MA 02111,USA.
Accepted for publication: 7 August 2002.

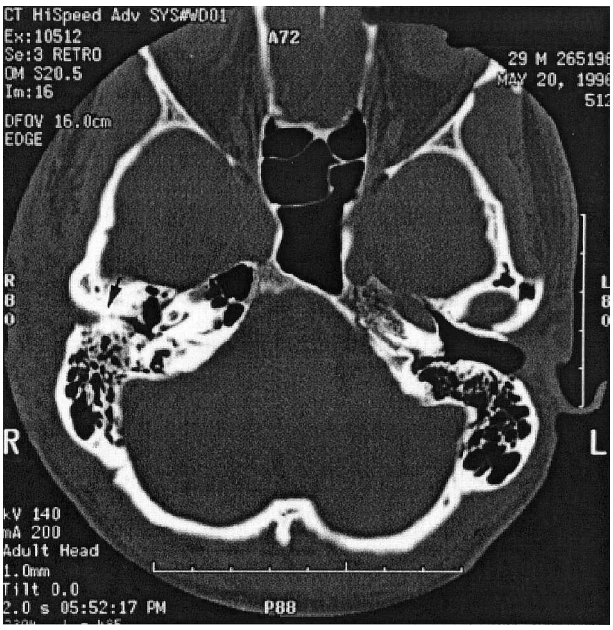

FIG. 1

An axial temporal bone CT scan of patient 1, a 29-year-old man presenting with aural atresia and a postauricular lesion draining into the atresia pouch though a sinus tract. There is a narrowing of the EAC on the right (arrow), which initially appeared to represent a bony stenosis. At the time of surgery, however, this was found to be a complete bony atresia plate. There is a normal inner ear and a well aerated middle-ear and mastoid cavity. There is soft tissue swelling adjacent to the squamous portion of the temporal bone. No obvious cyst or tract is appreciated. There is no evidence of a destructive process.

antero-superior aspect of the external auditory canal remnant. There was a moderate degree of inflammation adjacent to the tract and temporomandibular joint (Figure 2).

The patient underwent surgical resection of an infected cyst through a post-auricular approach. The cyst was deep to the temporalis muscle, abutting the

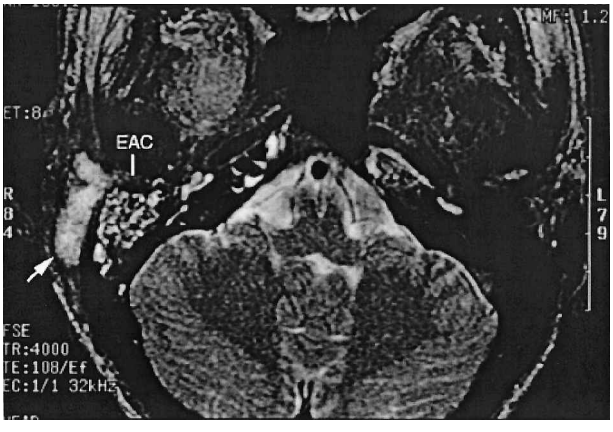

FIG. 2

A T1-weighted axial MRI scan with gadolinium of *Case 1* is shown at the same level as the CT scan shown in Figure 1. It demonstrates a cyst (arrow) in the superior postauricular region with a tract running anteriorly to communicate with the external auditory canal (EAC). There is a moderate amount of oedema around the tract. There is bright signal present in the mastoid air cells, consistent with inflammatory changes.

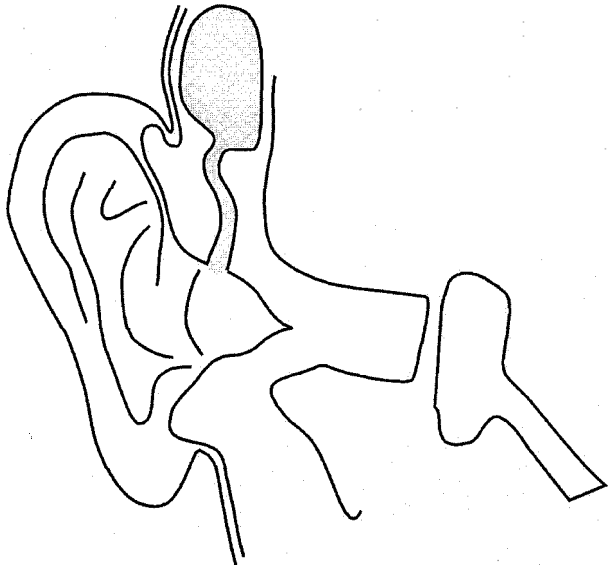

FIG. 3

Schematic depiction of the first cleft anomaly found in *Case 1*. There is a bony aural atresia and a sinus tract communicating with the lateral superior aspect of the external auditory canal remnant. The sinus tract does not involve the tissues medial to the atresia plate.

squamous portion of the temporal bone. A well-defined tract terminated in the lateral atresia pouch. A bony atresia plate was found to be complete. The EAC and middle ear were not reconstructed during this procedure. No cholesteatoma was found. Histopathological examination of the specimen revealed acute and chronic inflammation with copious granulation tissue. Areas of squamous epithelium were disrupted by chronic inflammation. After five years of follow-up, the patient has remained free of any symptoms of recurrence or infection, and has not required any additional otological procedure. A schematic depiction of this lesion is seen in Figure 3.

Case 2

A 26-year-old white man presented with a 10-year history of a slowly-enlarging left post-auricular mass. He had a history of an ipsilateral congenital aural atresia and microtia. He had no history of infection, pain, or drainage. The mass was approximately 3 cm in its largest dimension, fluctuant, and non-tender. The canal was atretic, and the auricle was small and dysmorphic, although all anatomical subunits were present. An audiogram showed a maximal conductive loss on the left.

The patient requested resection of the cyst, and wished no additional reconstruction be attempted. At surgery, the cyst was found to be well encapsulated with no evidence of a tract. Histopathological inspection revealed a squamous epithelial lining and some chronic inflammation. The patient has had no recurrence or further symptoms at three years follow-up. A depiction of his ear anatomy is shown in Figure 4.

https://doi.org/10.1258/002221503321046612 Published online by Cambridge University Press

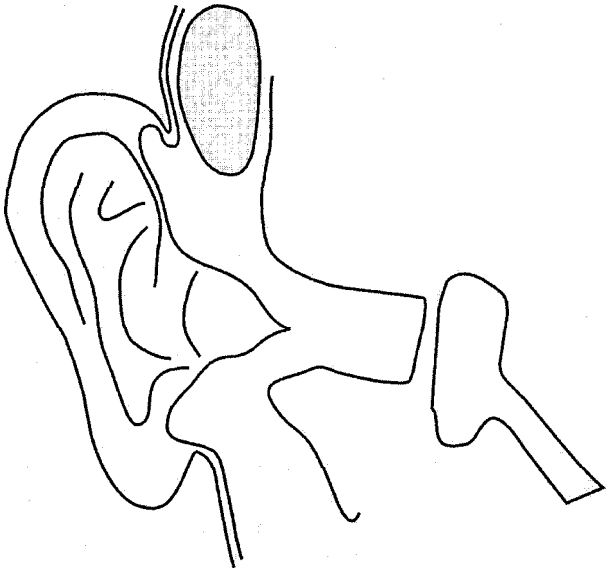

FIG. 4

Schematic depiction of the first cleft anomaly found in *Case 2*. There is a congenital aural atresia and a post-auricular cyst. There was no evidence of a tract communicating with the skin or any ear structure.

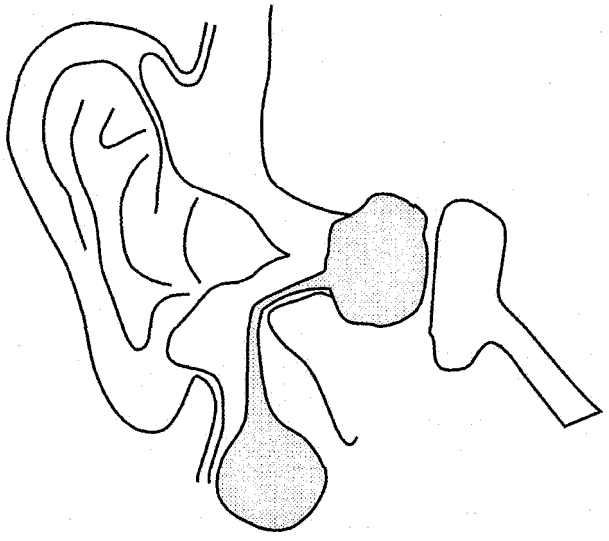

FIG. 6

Schematic depiction of the first cleft anomaly found in *Case 3*. There is a bony atresia plate and a cholesteatoma in the medial aspect of the external auditory canal adjacent to the atresia plate. This cholesteatoma is joined to a cyst in the neck by a squamous-lined tract and appeared to originate from the same duplication anomaly.

Case 3

A 20-year-old Asian woman with a right congenital aural atresia presented with a mass at the ipsilateral angle of the mandible just anterior to the sternocleidomastoid muscle. Two weeks after its first appearance, the mass became tender and indurated, and was incised and drained externally. Several months later, the drainage recurred at the incision site. The patient had a malformed tragus and pre-auricular skin tag. Since this patient presented prior to the advent of CT, plain tomograms were performed, and confirmed the presence of a bony atresia plate with apparently normal middle and inner ears. A radiolucent area was identified lateral to the atresia plate (Figure 5). Her audiogram revealed a 45–50 dB right-sided conductive hearing loss.

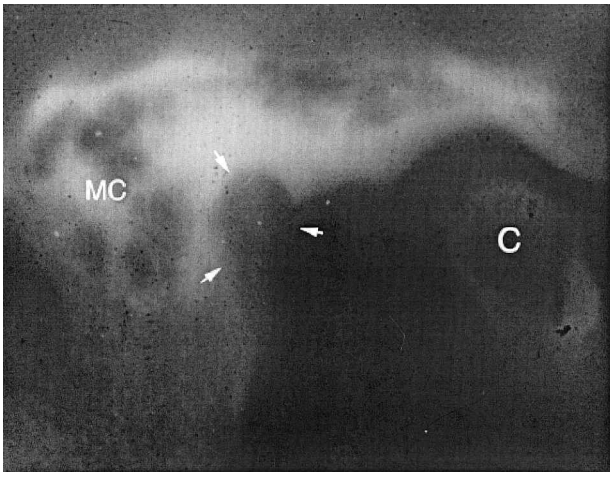

FIG. 5

A sagittally-oriented tomogram of the temporal bone in *Case 2*, a 20-year-old woman with a bony aural atresia and duplication anomaly is shown. There is a lucency (arrows) lateral to the middle ear which proved to be a cholesteatoma. The cholesteatoma was continuous with a squamous-lined cyst at the angle of her mandible. The mastoid air cells (MC) and mandibular condyle (C) are also shown.

The infected neck mass was surgically explored, and was found to consist of a tract extending from the cervical skin to a keratin-filled cyst just lateral to the bony atresia plate, and just deep to an intact skin-lined atresia pouch. The cyst and tract were excised, and pathology confirmed these to be lined by squamous epithelium, and filled with keratin debris. No cartilage was identified in the specimen.

Two years later, the patient developed right otalgia and otorrhoea from the atretic right external auditory canal. On exploration, this appeared to be secondary to recurrent or recidivistic squamous epithelium lateral to the bony atresia plate. A canal wall down tympanomastoidectomy was performed, and the bony atresia plate was removed. The malleus was malformed, while the incus, stapes and the facial nerve were normal. One year later, the patient had a clean mastoid bowl and middle-ear cleft. A depiction of this patient's original branchial cleft anomaly is shown in Figure 6.

Case 4

A patient with a variant of Treacher Collins syndrome had had bilateral aural atresia, microtia and cutaneous cervical fistulae since birth. The patient had had chronic serous drainage from the right fistula throughout infancy and childhood. Intermittently, the drainage would become purulent when the patient had an upper respiratory infection.

https://doi.org/10.1258/002221503321046612 Published online by Cambridge University Press

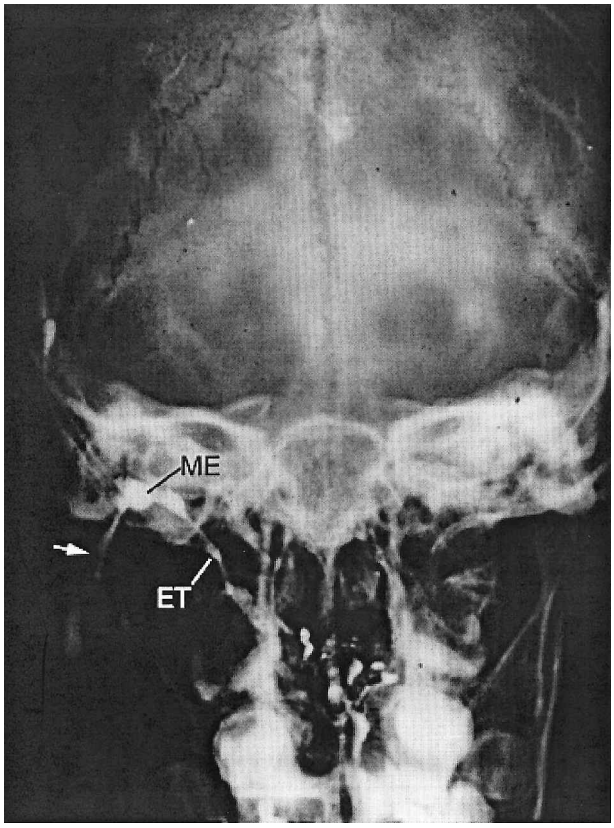

FIG. 7

AP skull film with contrast of *Case 3*, a child with congenital aural atresia and bilateral draining neck fistulae. Contrast can be seen filling the fistula (arrow) confirming its continuity with the middle-ear cleft (ME). Contrast can also be seen draining into the nasopharynx through the eustachian tube (ET).

The patient had multiple other congenital anomalies including hypoplasia of the midface and mandible, antimongoloid slant of the eyes, a fish-shaped mouth, and high arched palate. He also had severe myopia of the right eye, short fingers and toes, webbing of the second and third fingers, bilateral lid ptosis and mental retardation. He had a severe bilateral mixed hearing loss and used bone conduction hearing aids.

The patient had undergone multiple reconstructive procedures for his microtia, but no procedures had been undertaken to correct the aural atresia. Plain tomograms demonstrated bilateral bony aural atresia. The middle-ear spaces were aerated bilaterally and the ossicles appeared to be malformed. The inner ears appeared normal. At 13 years of age, the patient had a fistula-gram of the right draining fistula which confirmed the presence of a tract extending from the skin to the middle ear cleft, allowing drainage of contrast down the eustachian tube into the nasopharynx (Figure 7). Because of the patient's additional numerous malformations, no further intervention was undertaken. The patient continued to have intermittent drainage from the right fistula tract. A depiction of this lesion is shown in Figure 8.

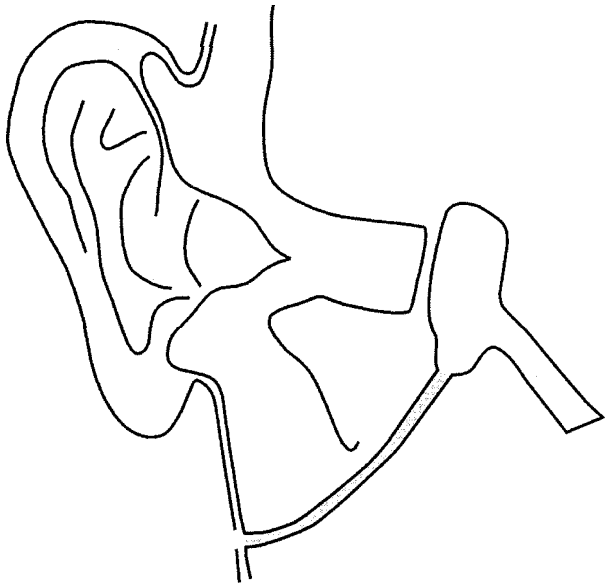

FIG. 8

Schematic depiction of the first cleft anomaly found in *Case 4*. There is a bony atresia plate in the medial ear canal with a near complete membranous atresia of the remainder of the EAC. A fistula tract runs between the middle ear and neck skin.

Discussion

The branchial apparatus begins to develop in the fourth gestational week and gives rise to many of the structures of the head and neck. The system consists of clefts, arches and pouches. The external clefts are lined by ectoderm and, with the exception of the first cleft, are obliterated during embryogenesis. The first cleft persists and develops into the external auditory canal. The arches, containing mesoderm, give rise to the cartilaginous, muscular, nervous and vascular elements of the head and neck. The internal pouches are lined by endoderm and give rise to the lining of the upper aerodigestive tract.[1] Multiple anomalies of the branchial apparatus can occur and have been extensively discussed in the literature. The most common are the second branchial cleft anomalies. First branchial cleft duplication anomalies are far less common, accounting for less than one per cent of all branchial anomalies.[2]

Embryologically, the first cleft is first seen at the fourth week of human gestation as a small indentation on the ectodermal surface between the first and second branchial arches. As the first and second arches grow, the cleft deepens with dorsal and ventral components.[3] The first and second arches project buds or hillocks superior and inferior to the cleft, which fuse at six weeks to create the pinna.[4] The first to third hillocks are from the first arch and give rise to the tragus, crus of the helix and the helix. The fourth to sixth hillocks originate from the second arch, and give rise to the antihelix and antitragus.[4] The first and sixth hillock fuse anteriorly to obliterate the ventral aspect of the first cleft.[5] The dorsal aspect of the cleft continues to grow towards the tubotympanic recess (elongating first pouch)

https://doi.org/10.1258/002221503321046612 Published online by Cambridge University Press

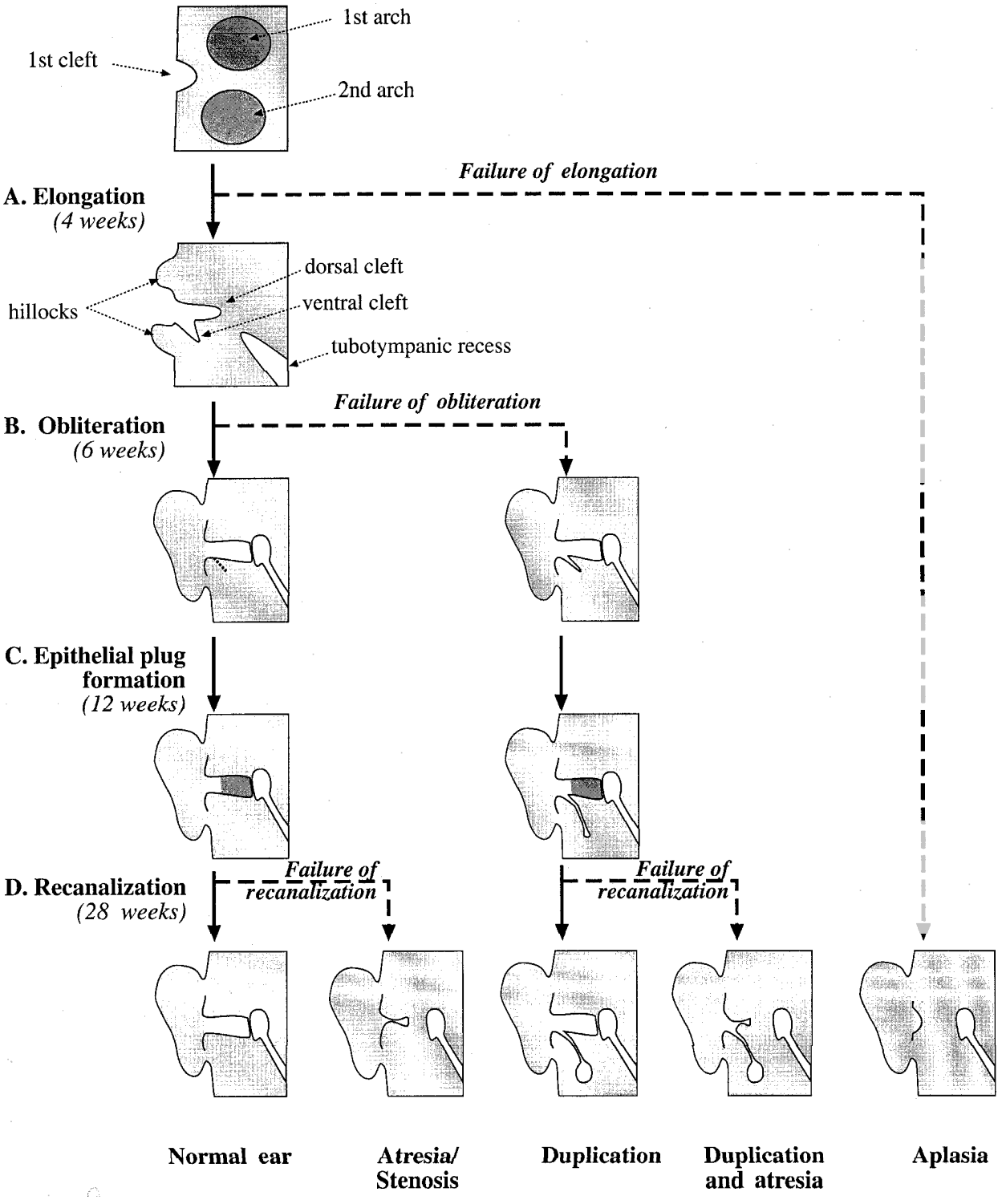

FIG. 9

In this flow chart, the normal development of the first cleft is illustrated. A schema for potential abnormal development is also shown.

(a) At four weeks, there is a dimpling of the ectodermal surface that elongates via two mechanisms: medial growth of the first cleft and lateral growth of the first and second arches. Failure of elongation of the cleft at four weeks secondary to lack of medial growth results in aplasia of the EAC.

(b) Normally at six weeks, there are two limbs to the cleft: ventral and dorsal. The dorsal component is the precursor to the final EAC, the ventral cleft is obliterated. Failure of obliteration of the ventral cleft at six weeks results in a duplication anomaly. This may be a cyst, sinus or fistula.

(c) At 12 weeks, the EAC is obliterated by an epithelial plug.

(d) At 28 weeks, the EAC is recanalized to from a normally patent canal. Failure of recanalization results in atresia or stenosis of the EAC.

Duplication anomalies and atresia can coexist if a failure of obliteration precedes a failure of recanalization.

https://doi.org/10.1258/002221503321046612 Published online by Cambridge University Press

until about the third month of gestation. At that time, the dorsal cleft is obliterated by a meatal plug comprised of epithelial cells. This obliterated tract recanalizes at about seven months gestation to create the normal patent EAC.[1] A schematic illustration of this process and possible malformations are shown in Figure 9.

Several classification systems have been established to clarify the clinical presentation and treatment approach for first cleft anomalies.[5–7] In his landmark 1972 study, Work used both histological and clinical factors to classify first branchial cleft cysts, sinuses and fistulae into two types.[7] Work's Type I branchial cleft anomalies are defined as being 'pure first branchial cleft anomalies', composed solely of ectodermal elements. Clinically, these Type I anomalies present as periauricular masses or abscesses. They may communicate with the EAC, usually at the bony-cartilaginous junction. Type I lesions have a close association with the inferior division of the facial nerve. In contrast, Work's Type II anomalies are thought to involve elements of the first or second arch (mesoderm) in addition to the first cleft (ectoderm). For this reason, such anomalies contain cartilage, a derivative of arch mesoderm, in addition to an epithelial lining. Type II anomalies usually present as masses at the angle of the mandible, and may communicate with the EAC. The often have an intimate anatomical association with the main trunk of the facial nerve.[7] Many authors have commented on the difficulty in categorizing first cleft anomalies into Work's classification since the clinical picture does not always correlate with the histologic findings.[5,6,8,9]

Traditionally, the term 'first cleft anomaly' has been limited to imply the existence of an abnormal epithelialized space, whether it is a cyst, sinus or fistula. Such a narrow definition does not account for the entire complexity of the embryology of the first cleft. There would appear to be at least three major steps in the normal development of the first cleft and its derivative structures (1) elongation of the cleft, (2) obliteration of the ventral portion of the cleft, and (3) recanalization of the cleft. Abnormalities affecting any of these stages may produce the spectrum of abnormalities in the cleft's derivative structures observed clinically. A classification can be proposed which is based on the following defect subtypes: (1) failure of elongation resulting in aplasia, (2) failure of obliteration resulting in a duplication anomaly and (3) failure of recanalization, resulting in atresia or stenosis (Table I, Figure 9).[6]

The most fundamental defect of the first cleft will result from a failure of cleft elongation at four weeks gestation. Anatomically this presents as an aplastic EAC with complete absence of a tympanic ring, and retrodisplacement of the mandibular condyle. The middle ear and inner ear can be normally developed in these patients since they are derived from the first arch, first pouch and otic placode, and not from the first cleft. A true aplastic first cleft anomaly cannot co-exist with failure of obliteration or recanalization anomalies since these are dependent on the pre-existing elongation of the first cleft which by definition, is lacking in aplasia.

Failure of first cleft obliteration results from persistence of the ventral aspect of the first cleft.[5] This obliteration normally occurs at five to six weeks of gestation, and failure of this obliteration may result in duplication anomalies of the EAC. Such a persistent space may become an epithelial-lined structure (cyst, sinus or fistula) traditionally thought of as first branchial cleft lesions.[5] Rarely, they can present as fistulae between the middle ear and the skin as in our fourth case. This type of communication likely occurs as the result of a breakdown in the tissues normally separating the first cleft from the tubotympanic recess (first pouch). This 'closing membrane' normally goes on to become the tympanic membrane.[5] A break in the closing membrane results in communication between the cleft and pouch, thereby potentially allowing a fistula tract to communicate between the middle ear and skin as seen in *Case 4*. An atresia may still develop in such cases if the first cleft fails to completely recanalize.

Failure of recanalization results when the meatal plug in the medial dorsal cleft persists. Normally, recanalization of the EAC occurs at seven months gestation.[1] Failure of recanalization may result in atresia or stenosis of the EAC. In cases of partial recanalization, squamous epithelium may be trapped medial to a tight stenosis or membranous atresia resulting in a canal cholesteatoma.[10]

In light of this potential classification system for first branchial cleft anomalies, our patients presented here suggest that various combinations of the latter two anomalies can occur. The obliteration of the ventral cleft and the recanalization of the dorsal cleft occur at different times during gestation. Failure of obliteration, combined with a later disordered recanalization could produce the coexistence of the anomalies presented here. The presence of an early obliteration anomaly may increase the likelihood of developing a second anomaly in the further devel-

TABLE I

PROPOSED CLASSIFICATION FOR FIRST CLEFT ANOMALIES. THIS TABLE SUMMARIZES THE DEVELOPMENTAL ABERRATIONS, THE GESTATIONAL TIME AT WHICH THE DEFECT OCCURS, AND THE RESULTING CLINICAL FINDINGS. COMBINATIONS OF FAILURE OF OBLITERATION AND RECANALIZATION CAN OCCUR AS ILLUSTRATED BY THE CASES PRESENTED

Developmental defect	Gestational age	Result
Failure of elongation	4 weeks	Aplasia
Failure of obliteration	6 weeks	First cleft duplication (cyst, sinus, fistula)
Failure of recanalization	28 weeks	Atresia or stenosis

https://doi.org/10.1258/002221503321046612 Published online by Cambridge University Press

opment of the EAC. First and second arch abnormalities can also coexist with first cleft anomalies, producing mesodermal elements in duplications or as maldevelopment of middle-ear structures, facial nerve or stapedial artery.

It is important to differentiate a true duplication anomaly from the canal cholesteatoma occasionally seen in malformed ears. Partial recanalization of the EAC may result in a tight stenosis, thereby preventing the normal migration of keratinizing epithelium of the medial canal. Similarly, squamous epithelium could be trapped behind a membranous atresia. In either case, a canal cholesteatoma may develop as a result. Such a canal cholesteatoma could be mistaken for a coexisting duplication anomaly, especially if it becomes infected, and drains to the external skin. Further confusing the differentiation is the fact that a duplication anomaly may itself result in the development of a cholesteatoma. Persistent keratinizing epithelium from the ventral cleft may proliferate within a closed space, resulting in a cholesteatoma. Cholesteatoma may be more likely to occur from a duplication of the EAC if it coexists with an atresia that prevents the egress of keratin debris. This was the presumed aetiology of the cholesteatoma in our third case, although it is impossible to say this with certainty. It can be particularly difficult to appreciate the origin of a cholesteatoma in a malformed ear since the diagnosis may be made late, after bone erosion has further distorted the anatomy.

As illustrated by our cases, the clinical diagnosis of duplication anomalies coexisting with aural atresia can be difficult. Three out of four patients presented in young adulthood with periauricular lesions. Two had become infected, and were refractory to usual medical therapy and surgical drainage. In such patients, a thorough exam of the EAC pouch for pits or tracts suggestive of a duplication anomaly is advised. Of course, other more common disorders that also occur in normally developed ears should be considered, including otitis media, neoplasms, adenitis, coalescent mastoiditis, and sebaceous cysts.

Imaging studies are helpful in establishing the diagnosis and treatment plan of a coexisting duplication anomaly in an atretic ear. A CT scan will help to demonstrate the nature of the atresia or stenosis, and evaluate involvement of the middle ear and mastoid. Two of our patients were treated before the advent of CT scanning and in those patients, useful information was extracted from plane films and tomograms. The use of contrast material injected into a fistula (fistula-gram) may also be helpful in delineating the course of a tract.

Once a duplication anomaly in an atretic ear is diagnosed, treatment can be tailored to the specific lesion. Duplications lacking any communication with structures medial to the atresia plate may be treated with complete surgical excision without the need to simultaneously address the aural atresia (as for *Cases 1* and *2*). Anomalies that communicate with the middle ear or EAC medial to an atresia require simultaneous repair of the atresia (as for *Case 3*). In cases with minimal symptoms, and no evidence of infection or enlarging mass, the duplication may not have to be resected at all (as for *Case 4*). Complete excision of duplication anomalies occurring in conjunction with atresia or stenosis can be curative. It is important to remember that the facial nerve may be closely associated with these anomalies, and the surgeon must be prepared to identify the nerve safely at the time of the procedure.[11]

Conclusion

Duplication anomalies of the first branchial cleft can occur in conjunction with congenital aural atresia. The clinical presentation of such coexisting first cleft abnormalities may be confusing. Radiographic imaging is helpful in identifying such abnormalities and planning treatment. Complete excision of cysts, sinuses and fistulae is often necessary, and in some cases may require the simultaneous repair of the atresia or stenosis. In light of the spectrum of anomalies that may result from aberrant first branchial cleft development, we present a potential classification scheme based on the embryologic stages of first cleft development.

References

1 Sadler T. *Langman's Medical Embryology*. Baltimore: Williams and Wilkins; 1985:281–319
2 Finn DG, Buchalter IH, Sarti E, Romo T, Chodosh P. First branchial cleft cysts: clinical update. *Laryngoscope* 1987;**97**:136–40
3 Michaels L, Soucek S. Auditory epithelial migration on the human tympanic membrane: II. The existence of two discrete migratory pathways and their embryological correlates. *Amer J Anat* 1990;**189**:189–200
4 Karmody C, Annino DJ. Embryology and anomalies of the external ear. *Facial Plast Surg* 1995;**11**:251–6
5 Olsen KD, Maragos NE, Weiland LH. First branchial cleft anomalies. *Laryngoscope* 1980;**90**:423–36
6 Karmody CS. A classification of the anomalies of the first branchial groove. *Otolaryngol Head Neck Surg* 1979;**87**:334–8
7 Work W. Newer concepts of first branchial cleft defects. *Laryngoscope* 1972;**982**:1581–93
8 Arndal H, Bonding P. First branchial cleft anomalies. *Clin Otolaryngol* 1996;**21**:203–7
9 Ikarashi F, Nakano Y, Nonomura N, Kawana M, Okura T. Clinical features of first branchial cleft anomalies. *Amer J Otolaryngol* 1996;**17**:233–9
10 Nishimura Y, Minatogawa T, Kumoi T. Intractable retroauricular abscess associated with microtia and aural atresia – some views in relation to the congenital cholesteatoma and microtia. *Ann Plast Surg* 1989;**23**:74–80
11 McRae R, Lee KJ, Goertzen E. First branchial cleft anomalies and the facial nerve. *Otolaryngol Head Neck Surg* 1983;**91**:197–202

Address for correspondence:
Nikolas H. Blevins, M.D.,
750 Washington Street,
Boston, MA 02111, USA.

Fax: 617 636-1479
E-mail: nblevins@lifespan.org

N. Blevins, M.D. takes responsibility for the integrity of the content of the paper.
Competing interests: None declared