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Defects of the first branchial cleft

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Abstract
Four cases of first branchial arch defects are presented. The literature relating to such defects is reviewed and a possible embyrogenic mechanism is proposed with particular reference to the disposition of the facial nerve. The classification of these defects into Types I and II by Work (1972) is supported.

Key words: Branchial apparatus

Introduction

Developmental anomalies of the branchial apparatus have been recognized for over 160 years (Von Ascherson, 1832). Whilst such anomalies are not uncommon, comparatively few abnormalities resulting from anomalous development of the first branchial cleft have been reported. We report four cases of reduplication defects of the external auditory meatus.

The external auditory meatus develops from the first of a series of branchial clefts which groove the sides of the head of the embryo between the fifth and eighth weeks of gestation and correspond to endodermally lined outpouchings of the pharynx. The clefts are separated by paired arches. It is the mesoderm of the first and second arches which gives rise to the fibrous middle component of the tympanic membrane, with the first arch giving rise to the malleus and incus and the second arch giving rise to the stapes, stapedius muscle and pretrematic chorda tympani which invaginates itself into the mesodermal layer of the tympanic membrane en route to the mucosa of the tongue. Although there is a well-defined funnel-shaped tube at the site of the membranous external meatus, the ectoderm of the first cleft and the endoderm of the first pouch remain separated by thick mesoderm until the eighth week of gestation when, at the time that the ventral cleft is obliterated by fusion or overgrowth of the first and second arches, a cord of ectodermal cells grows inwards from the dorsal end of the first cleft towards the endoderm of the first pouch. For a short time the ectodermal plug may abutt the endoderm of the first pouch, but ingress of mesoderm results in their separation by the future mesodermal fibrous component of the tympanic membrane. Four ossification centres arise around the deep end of the ectodermal plug. These eventually give rise to the tympanic ring of the adult bony meatus. The ectodermal plug remains as a solid structure until the seventh month of gestation at which stage the central cells degenerate in a process of canalization which begins medially and progresses laterally until a patent canal is formed.

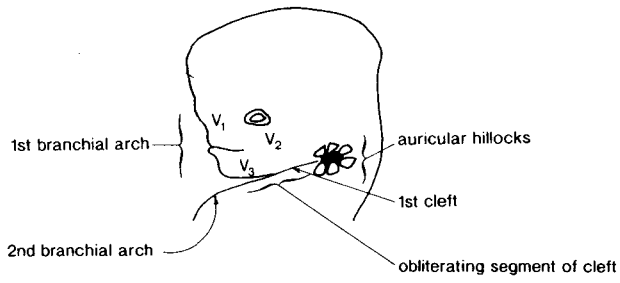

Fig. 1
Diagrammatic representation of 40 days gestation.

The external ear develops from six thickenings or hillocks which develop from the first and second arches around the dorsal end of the first branchial cleft. Initially distinct they become obscured and merge. The three hillocks which derive from the first arch probably give rise to the tragus and part of the helix, whilst the three from the second arch probably give rise to the lobule, part of the helix, antitragus, antihelix and concha. Cartilage arising within the hillocks forms the auricular skeleton and also grows inwards parallel to the ectodermal plug to form the cartilage of the cartilaginous meatus.

It is thought that the more commonly seen pre-auricular cysts and sinuses arise as a result of the disordered fusion of the hillocks and although they may occur in conjunction with meatal anomalies they represent separate developmental entities (Streeter, 1922; Work, 1972); (see Figures 1 and 2).

Case reports (see Figure 3)

Case 1

A 26-month-old male child presented with a history of recurrent abscess formation in the right retromandibular/parotid area. At the time of presentation he had already undergone four separate incision and drainage procedures for such abscesses. On examination there was no evidence of neck or meatal sinuses, but a cystic swelling was noted below the cartilaginous meatus. He failed to return for follow-up until aged seven years when he represented with a further abscess at the same site. On this occasion there was a definite sinus below the ear.

After two further episodes of infection surgical exploration was undertaken and amidst an area of dense scarring a cavity full of cheesy material was identified and excised along with two fibrous extensions which were traced to a firm attachment to the cartilaginous meatus adjacent to the tympanic ring. Healing was complicated by keloid formation requiring further surgery and triamcinolone injection.

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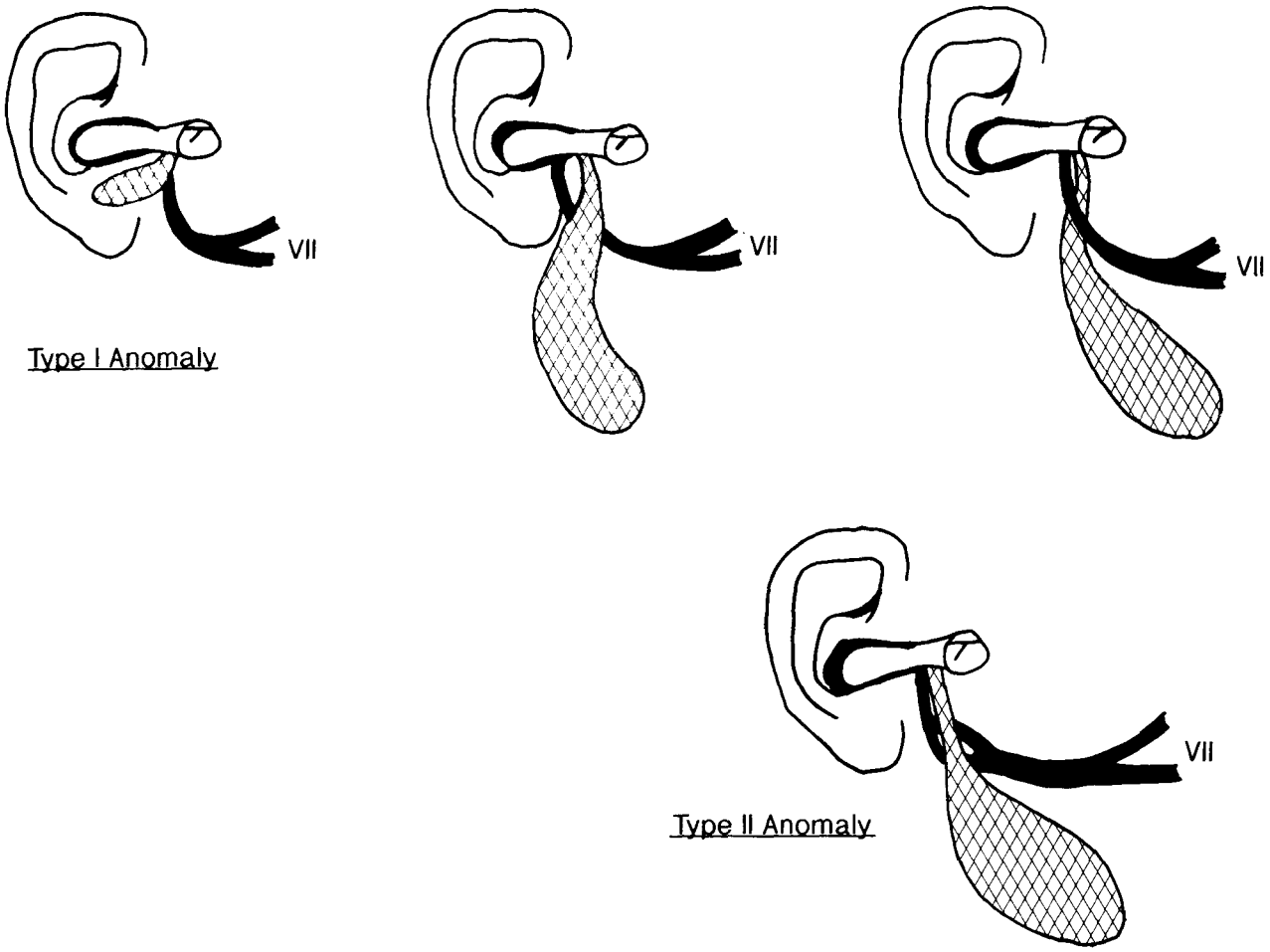

FIG. 2
Anomalies of the first branchial cleft (after Work, 1972).

Histopathological examination of the specimen revealed an ellipse of skin with a nipple-like opening from which leads a 3.5 cm ductular structure lined with keratinized squamous epithelium and filled with keratin. The features are those of an embryonic remnant.

Case 2 (see Figure 4)

A one-year-old female child presented with persistent unilateral otalgia. On admission for grommet insertion she was noted to have a cystic swelling of the floor of the external meatus which resulted in some displacement of the annulus and tympanic membrane antero-inferiorly. The cyst was incised and a cheesy material obtained. Post-operatively she became pyrexial and developed left neck swelling associated with persistent discharge from the site of incision of the meatal cyst. The episode of acute infection settled but a residual neck swelling persisted and was eventually biopsied by a general surgeon, obtaining inflamed salivary gland tissue. After several more episodes of acute infection associated with discharge from the ear, it was decided to explore the area formally. A fibrous mass was found lying postero-inferior to the main trunk of the facial nerve with two extensions running superiorly under the nerve, one running for a short distance anteriorly and containing an apparently cartilaginous component, whilst the other, which was more fibrous and had an obvious lumen, extended up to the floor of the external meatus and opened into the meatus at the junction of the bony and cartilaginous meatus.

Histopathological examination showed a cyst (2 × 11 cm) filled with lamellated squames and lined with stratified squamous keratinizing epithelium with hair follicles, sebaceous and apocrine glands and mature cartilage in the wall. Leading

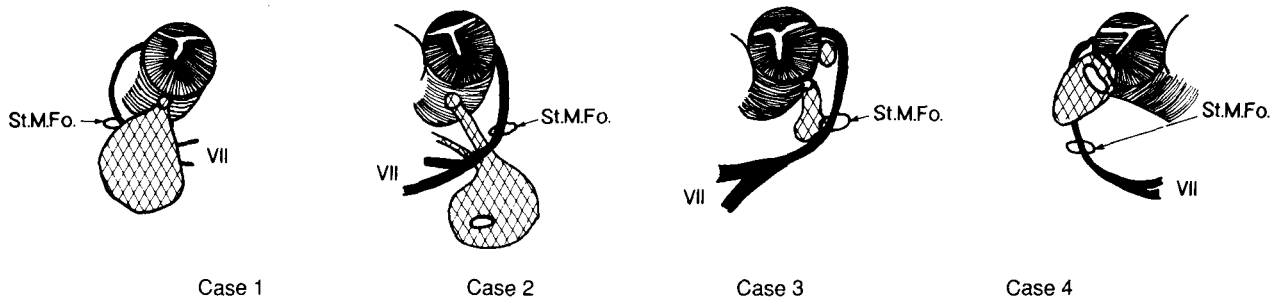

FIG. 3
Diagrammatic representation of the pathological findings in Cases 1–4. St.M.Fo. = Stylomastoid foramen nerve.

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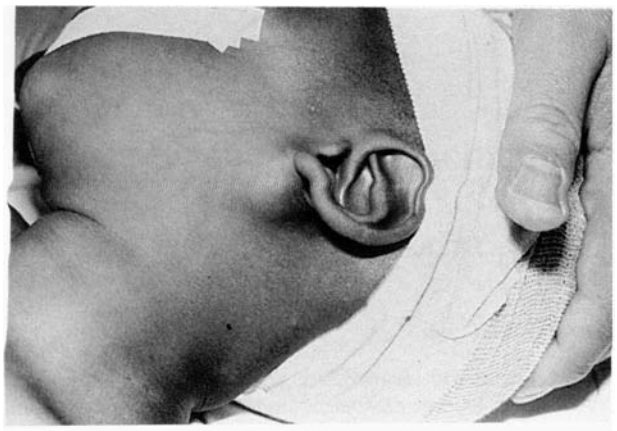

FIG. 4
Pre-operative photograph of Case 2.

from the cyst was a 1.5 cm sinus tract which showed signs of a chronic inflammatory cell infiltrate. An adjacent salivary gland showed signs of compression atrophy. The features are compatible for an inclusion dermoid with a fistulous tract.

Case 3

A 48-year-old woman with no prevous history of otological problems presented with an 11-day history of left otalgia and a complete lower motor neurone facial palsy. Examination revealed a normal tympanic membrane and two defects in the bony meatus 2–3 mm lateral to the annulus, one posteriorly and one inferiorly, each containing cheesy keratinaceous material. A provisional diagnosis of a meatal re-entry by cholesteatoma was made and the ear was explored surgically. The middle ear was normal and there was no evidence of cholesteatoma. The posterior pocket was found to end blindly in a granulation-filled dilatation about 1.5 mm in diameter adjacent to the vertical portion of the facial nerve at the level of the pyramid. The inferior defect was found to extend below the tympanic ring to the stylomastoid foramen where it ended in continuity with an area of inflammatory tissue adjacent to the exit of the facial nerve. The nerve was decompressed from the level of the pyramid to the stylomastoid foramen. Five months post-operatively there has been partial recovery of facial nerve function. Histopathological examination showed chronic inflammatory tissue only.

Case 4

A 53-year-old man presented with a six-month history of right otorrhoea. Nine years prior to this presentation he had had a right Bell's palsy which proved self-limiting. There were no other otological problems. On examination a swelling with a granular discharging punctum was noted arising from the floor of the meatus postero-inferiorly just lateral to the annulus. The cystic swelling was uncapped and marsupialized into the meatal lumen under general anaesthesia. It extended deep to the annular groove, which was intact, to form a bulge into the facial recess. The cyst was chronically inflamed with underlying carious bone which was curetted until apparently healthy bone was reached before the defect was lined with repositioned meatal skin.

Discussion

Aberrant development of the membranous external meatus may take the form of recanalization, atretic defects, or reduplication defects. The former may be associated with middle ear anomalies, whilst the latter generally occur in conjunction with normal middle ear development (Bellucci, 1981).

Work (1972) suggested a classification of the reduplication defects on the basis of their histological constitution: Type I defects were comprised exclusively of tissue of ectodermal origin, and Type II defects comprised of both ectodermal and mesodermal tissue. If their classification is applied to our cases then Case 2 may be classed as a Type II defect whilst the others would represent Type I defects.

The precise manner in which reduplication defects arise remains unclear. We would propose that Type I defects may result from abberrant recanalization of a single epithelial cord or recanalization of two parallel cords. Type II defects may result from the communication of the normal meatus with ectoderm trapped during the process of obliteration of the ventral portion of the first cleft by fusion or overgrowth of the first and second arches, creating an epithelially lined blind-ending sinus in communication with the meatus, a collaural fistula opening onto the neck along the line of fusion or an isolated inclusion dermoid which may subsequently fistulate to the neck skin or external meatus when infected (Kerr, 1963). In the first situation the anomaly might be expected to consist of a purely epithelial reduplication, whilst the second situation might conceivably give rise to an anomalous structure with both ectodermal and endodermal components. In both cases entrapment of desquamating squamous epithelium will result in the production of a cholesteatomatous process resulting in erosion of the bony external meatus, tympanic annulus and hypotympanum. This is one possible explanation of the phenomenon of 'congenital ear canal cholesteatoma'. The late presentation of Cases 3 and 4 was presumably the result of the superimposition of an infective process onto a pre-existing anomaly which resulted in the conversion from a previously self-cleaning pocket into a non-self-cleaning pocket with the result that the entrapped desquamating epithelium assumed the characteristics of an erosive cholesteatoma.

The major consideration in the surgical exploration of the defects is their disposition with respect to the facial nerve. If our hypothesis is correct then one would expect that the anomalous tract or cavity would lie superficial to the main trunk of the facial nerve in a Type I defect. With a Type II defect, arising as a result of disordered fusion of the ventral portions of the first and second branchial arches, the variability of the depth and position of the trapped ectoderm and the uncertainty as to the origin of the mesodermal, usually cartilaginous, component of the defect means that the disposition of the tract or cavity with respect to the facial nerve will be variable. In Case 2 the tracts ran deep to the trunk of the facial nerve, but a review of cases in the literature reveals that the nerve may lie deep or superficial to the tract (Aimi and Takino, 1962), or indeed may be split by it so that two separate portions embrace the track (Crymble and Braithwaite, 1964). It is not surprising that several authors report facial nerve injury after incision and drainage of abscesses associated with Type II defects. The differentiation between Type I and Type II defects will often only be possible in retrospect and their exploration should therefore be undertaken with great care in order to avoid damage to the facial nerve.

The superficial disposition of the Type I defects with respect to the facial nerve does not eliminate the risk of damage to the nerve during surgery, as such defects may lie in close apposition to the nerve in the region of the stylomastoid foramen. In Cases 2 and 4 both patients had a history of facial nerve palsy prior to surgery and in Case 3 the proximity of inflamed tissue derived from the anomalous tract to the facial nerve would suggest a causative relationship.

In a Type II defect it is likely that the substance of the parotid gland will embrace the defect in a similar manner to the facial nerve as it develops by a process of divisive outpouching for the oral cavity. Again intimate association with the parotid gland appears to be a common finding in surgical exploration of Type II defects. In the Type I defects this does not appear to be the case.

Conclusions

We report four cases of first branchial cleft abnormalities.

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Consideration of the likely patterns of embryological development of these and other cases in the literature supports the classification of such defects into Types I and II as proposed by Work (1972).

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