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## Abstract

Temporal bone histopathological findings of two patients with trisomy 18 syndrome are described. Many of the abnormalities previously described were seen in the present cases; namely, atresia of the external auditory canal, aberrant course of the tensor tympani muscle, malformed stapes, aberrant course of the facial nerve with an obtuse angulation at the first genu and displacement of geniculate ganglion cells into the internal auditory canal, shortened cochlea with decreased spiral ganglion cell population, and vestibular anomalies, such as bony and membranous blockage of the superior semicircular canal. Moreover, an extremely underdeveloped malleus and incus continuous with a persistent Meckel's cartilage were observed.

**KEYWORDS:** temporal bone, pathology, trisomy 18

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## Temporal Bone Histopathology in Trisomy 18 Syndrome : A Report of Two Cases

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Temporal bone histopathological findings of two patients with trisomy 18 syndrome are described. Many of the abnormalities previously described were seen in the present cases; namely, atresia of the external auditory canal, aberrant course of the tensor tympani muscle, malformed stapes, aberrant course of the facial nerve with an obtuse angulation at the first genu and displacement of geniculate ganglion cells into the internal auditory canal, shortened cochlea with decreased spiral ganglion cell population, and vestibular anomalies, such as bony and membranous blockage of the superior semicircular canal. Moreover, an extremely underdeveloped malleus and incus continuous with a persistent Meckel's cartilage were observed.

*Key words*: temporal bone, pathology, trisomy 18

Since the initial report by Kos *et al.* (1) in 1966, temporal bone findings in 11 cases of trisomy 18 syndrome have been reported in the literature (2-9). However, the features and localizations of the anomalies have varied with each individual case, and no general pattern of anomalies of the stato-acoustic organ in the disease has yet been established. In the present study, microscopic findings of the temporal bone in 2 new cases of trisomy 18 syndrome are described, and a comparison with the findings reported hitherto is made.

### Cases

*Case 1.* A 4-day-old male, the first child of a 34-year-old mother, delivered by caesarean section at the 40th week of pregnancy. The child weighed only 2360 g at birth, and showed left microtia and atresia auris. Great difficulty of breathing and

severe cyanosis were observed immediately after delivery. These problems led to death on the 4th day. Diagnosis of trisomy 18 syndrome was confirmed by chromosomal analysis. The postmortem examination revealed left external hydrocephalus, left atresia auris, microtia, micrognathia, atrial septal defect, ventricular septal defect, and duplication of the right pelvis and ureter. The temporal bones were removed 25 h after death. They were fixed with 10% formalin, decalcified, embedded in celloidin, and sectioned horizontally at 20 microns. Every 10th section was stained with hematoxylin and eosin, and mounted for light microscopic examination.

*Case 2.* A 50-day-old female, the second child between a 31-year-old father and a 28-year-old mother, born spontaneously after 41 weeks of gestation. Body weight at birth was only 2220 g, and micrognathia, microphthalmia and low set auricle were observed. Heart murmurs were audible immediately after birth, and cyanotic attacks occurred repeatedly. The patient died of bronchopneumonia 50 days after birth. Diagnosis of trisomy 18 syndrome was confirmed by chromosomal

analysis. Postmortem examination revealed a low set auricle, micrognathia, microphthalmia, hypertelorism of the eye balls, ventricular septal defect, patent ductus arteriosus, fatty liver, dislocation of the pancreas and the jejunum, and flexion contracture of the limbs. The temporal bones were removed 12 h after death and examined as in case 1.

### Temporal Bone Histopathology

#### Case 1

*Left ear.* Atresia auris formed by soft connective tissue was observed. The attic space was hypoplastic and contained a malformed incus which articulated with a cartilaginous malleus. The cartilaginous malleus

was still in a developing stage and was connected with a large cartilage which appeared to be a persistent Meckel's cartilage. The manubrium of the malleus and the long process of the incus were defective. The stapes was monocrucal (columella type) and was fixed osseously to the edge of the oval window at the posterior and inferior margin of the footplate (Fig. 1). The stapedial muscle and tendon were missing. Although the tensor tympani muscle was present, it ended at the cochleariform process without a tendon. The tympanic cavity was small with remnant mesenchymal tissue under the mucous membrane. The round window niche was not pneumatized. The facial nerve was generally thin, with geniculate ganglion cells displaced into the



Fig. 1 Case 1. Left ear. Photomicrograph shows monopodal stapes (S) with bony fixation at posterior rim of footplate (arrow) and persistent Meckel's cartilage (M). F, facial nerve.  $\times 14$ .

internal auditory meatus and obtuse angulation at the first genu. The Fallopian canal was missing from the horizontal portion downwards. The facial nerve was exposed widely in the middle ear cavity, and it disappeared immediately after the branching of the chorda tympani at the level of the lower edge of the oval window. In the inner ear, the ampullated crus of the superior semicircular canal was osseously closed (Fig. 2), and the crista ampullaris was also missing. The posterior semicircular canal (both osseous and membranous labyrinths) was hypoplastic. The cochlea was formed, with 2 and a half turns, and no remarkable changes were found in the organ of Corti, tectorial membrane and Reissner's membrane. A decrease in the

spiral ganglion cell population was observed at the basal turn.

*Right ear.* The middle ear cavity was destroyed at the time of removal of the temporal bone, with the malleus and the incus being displaced and the stapes dislocated into the vestibule. The stapes was deformed with the anterior crus forking into two crura, one of which was displaced to the posterior part of the footplate. The tendon of the stapedial muscle was missing. The facial nerve was generally thin, with geniculate ganglion cells displaced into the internal auditory meatus and obtuse angulation at the first genu. The nerve descended without a horizontal segment. In the inner ear, the membranous superior semicircular canal was almost com-

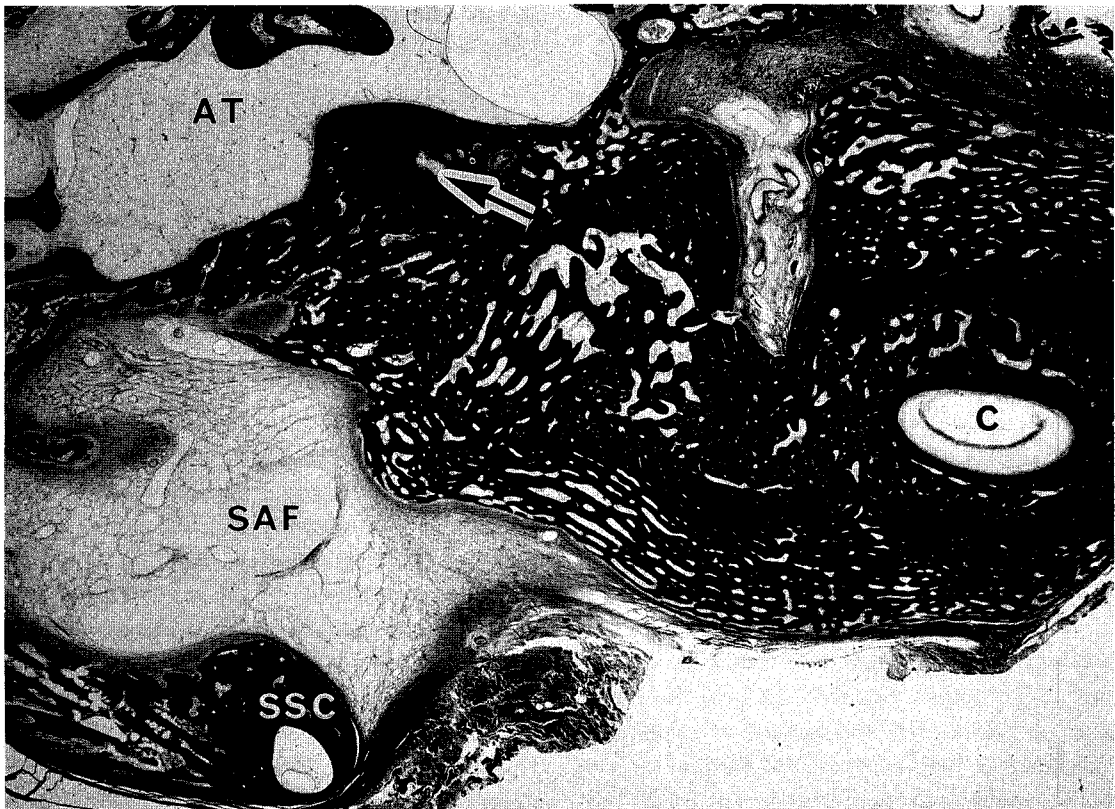


Fig. 2 Case 1. Left ear. Absence of lateral limb of superior semicircular canal (arrow). C, cochlea. SSC, medial limb of superior semicircular canal. AT, attic. SAF, subarcuate fossa.  $\times 6$ .



**Fig. 3** Case 1. Right ear. Stricture of membranous superior semicircular canal (arrow).  $\times 90$ .

pletely occluded (Fig. 3). The membranous posterior semicircular canal was also narrowed. Cyst formation was observed in the dark cell area around the crista ampullaris of each semicircular canal, and the spiral ganglion cell population was decreased at the lower basal turn. A large subarcuate fossa was noted in the otic capsule.

#### Case 2

*Left ear.* The tympanic membrane, the malleus and the incus were found to be normal. The stapes was thick at both the crura and footplate, and the posterior crus was displaced to the center of the footplate. The membranous superior semicircular canal was markedly constricted (Figs. 4 a, b). Dilatation of the endolymphatic duct was observed. The

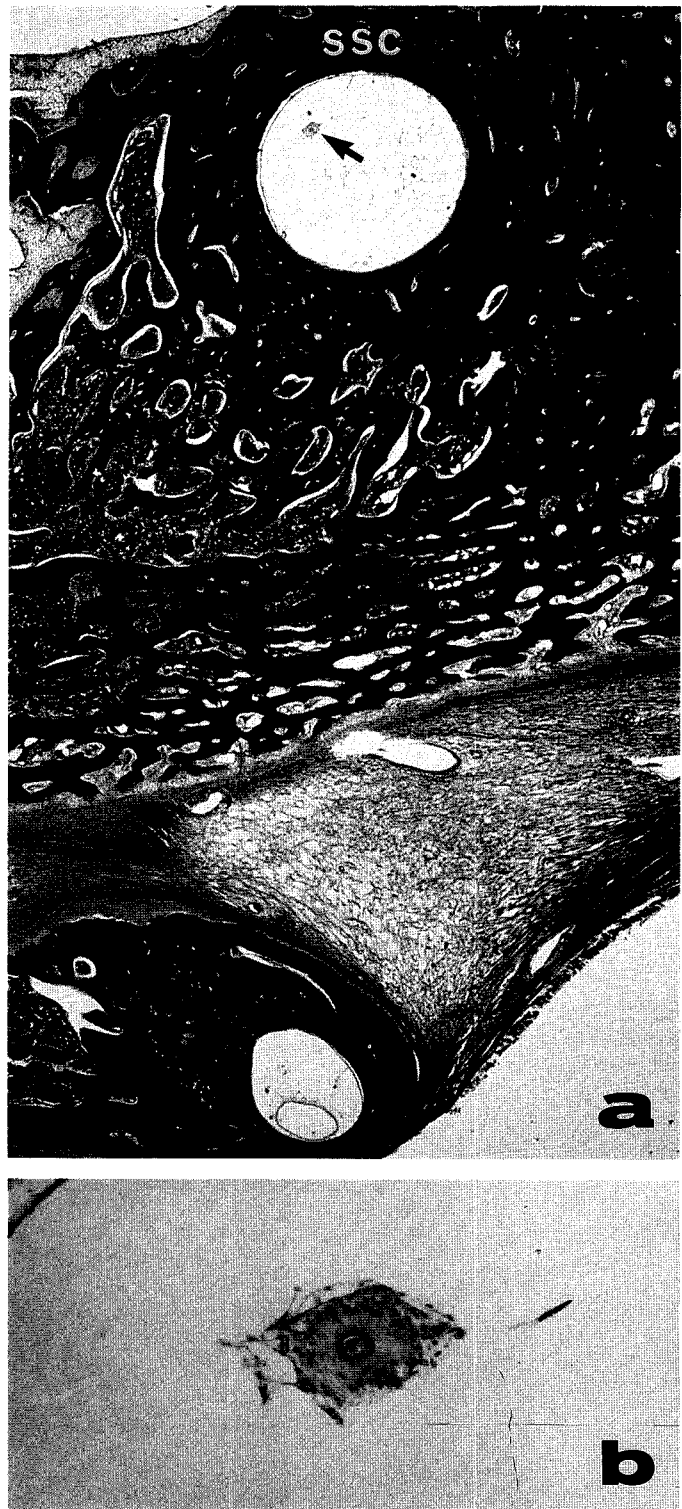
cochlea had only 2 spirals. The cell population of the spiral ganglion in the basal turn was markedly decreased.

*Right ear.* The tympanic membrane, the malleus and the incus were normal. The stapes was thick at both the crura and the footplate. The anterior crus was fat and forked into two crura, and the posterior crus was displaced to the center of the footplate (Fig. 5). The tensor tympani muscle took an aberrant course. The facial nerve showed obtuse angulation at the first genu, and the ganglion cells were displaced to the internal auditory meatus. The semicircular canals and the vestibule were normal. The endolymphatic duct was dilated. The cochlea had only 2 turns and vascular remnants were found at the scala vestibuli and Reissner's membrane. The cell population of the basal spiral ganglion was markedly decreased (Fig. 6).

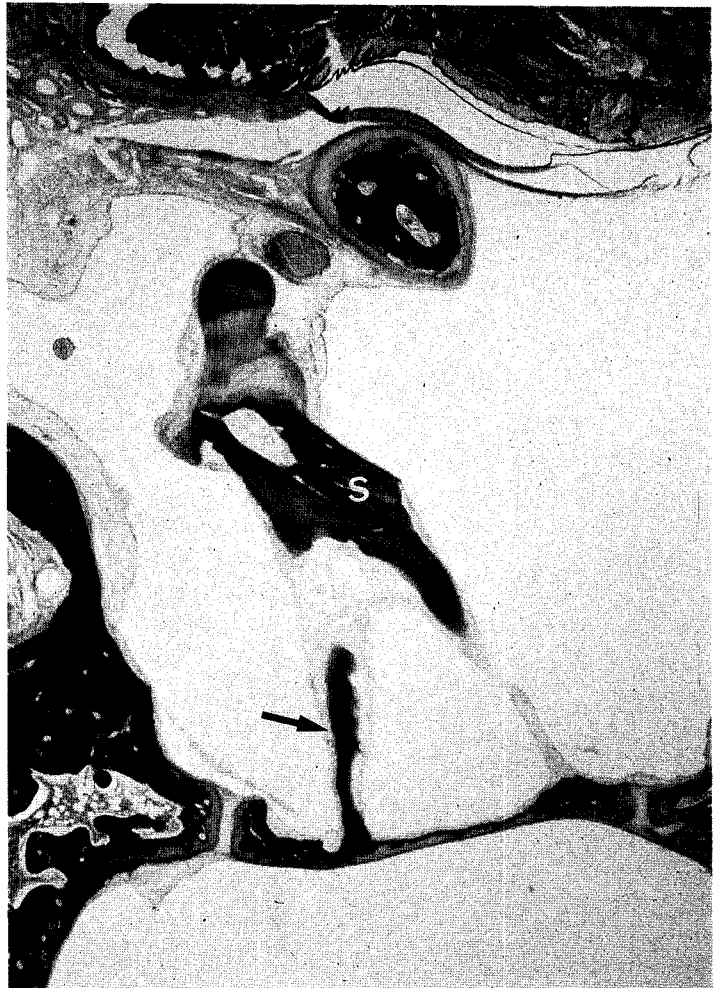
#### Discussion

Variability of temporal bone anomalies has been reported in patients with trisomy 18 syndrome. In our present cases of trisomy 18 syndrome, the severely involved parts were derivatives of the second branchial arch, particularly, the stapes, the manubrium of the malleus, the long process of the incus, the stapedial tendon and a portion of the Fallopian canal. These parts were usually malformed or underdeveloped. The facial nerve took an abnormal course, and its ganglion cells were displaced to the internal auditory meatus. Narrowing or atresia of the external auditory canal, an aberrant course of the tensor tympani muscle, and a widened subarcuate fossa were also observed. In the inner ear, a shortened cochlea associated with a hypoplastic modiolus, absence of the crista and a portion of the superior semicircular canal, and an enlarged vestibular aqueduct

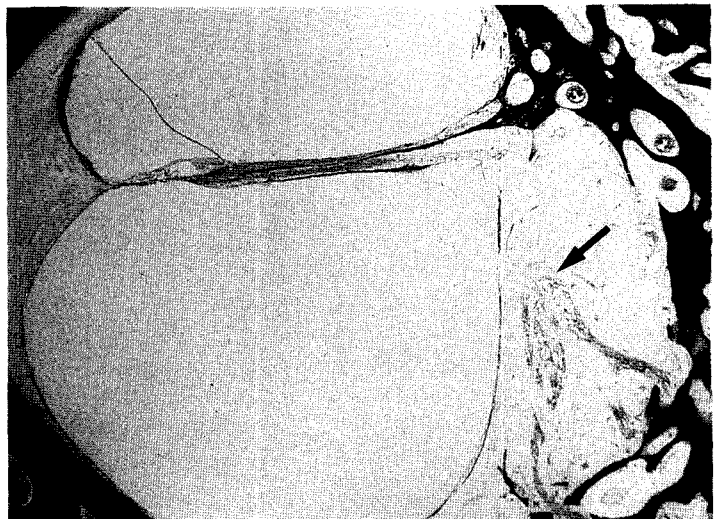
**Fig. 4** Case 2. Left ear. a: Stricture of membranous superior semicircular canal (arrow).  $\times 15$ . b: High magnification of strictured membranous canal.  $\times 100$ .



**Fig. 5** Case 2. Right ear. Photomicrograph shows malformed stapes (S) with displaced posterior crus (arrow).  $\times 14$ .



**Fig. 6** Case 2. Right ear. Decreased spiral ganglion cell population at basal turn (arrow).  $\times 36$ .





associated with hypoplasia of the endolymphatic duct and sac were observed. The cochlear nerve was also involved, with a decrease or absence of the spiral ganglion cells. These findings have also been observed in 10 cases of this syndrome reported by others and by us (1,2,4-9). The unique findings in the present cases were the extremely underdeveloped malleus and incus continuous with a persistent Meckel's cartilage.

As for the mechanism of the development of such anomalies as found in the inner ear, Miglets *et al.* have stated that these anomalies can be explained by developmental arrest (6). However, all of the anomalies observed cannot be fully explained by developmental arrest alone, and it must also be considered that aberrations of the autosomal chromosomes of group E may cause profound influences at various developmental stages of the statoacoustic organ, since a very wide range of anomalies of the inner, middle and external ears are found in the temporal bone of patients with this type of chromosomal aberration. Studies on many more cases are necessary to clarify these problems.

## References

1. Kos AO, Schuknecht HF and Singer JD: Temporal bone studies in 13-15 and 18 trisomy syndrome. *Arch Otolaryngol* (1966) **83**, 439-445.
2. Kelemen G: Rubella and deafness. *Arch Otolaryngol* (1966) **83**, 520-532.
3. Kelemen G, Hooft C and Kluyskens P: The inner ear in autosomal trisomy. *Pract Oto-Rhino-Laryngol* (1968) **30**, 251-258.
4. Sando I, Bergstrom L, Wood II RP and Hemenway WG: Temporal bone findings in trisomy 18 syndrome. *Arch Otolaryngol* (1970) **91**, 552-559.
5. Lindsay JR: Profound childhood deafness: Inner ear pathology. *Ann Otol Rhinol Laryngol* (1973) **82**, Suppl: 5.
6. Miglets AW, Schuller D, Ruppert E and Lim DJ: Trisomy 18: A temporal bone report. *Arch Otolaryngol* (1975) **101**, 433-437.
7. Saito R, Sonobe N, Matsumura M, Ogura Y, Yoda T, Ubuka S and Nakamura S: Temporal bone findings in trisomy 18 syndrome. *Pract Otol (Kyoto)* (1977) **70**, 927-937.
8. Saito H and Furuta M: Temporal bone findings in patients with E trisomy. *Ear Res Jpn* (1983) **14**, 337-339.
9. Minatogawa T, Kimura J and Ito H: Trisomy 18: A temporal bone report. *Ear Res Jpn* (1984) **15**, 299-300.

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