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Osteogenesis Imperfecta – A Human Temporal Bone Case Study

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Osteogenesis imperfecta (OI) is defined as one of the inherited connective tissue disorders that can result in fragile bones, blue sclera, and deafness. This genetic disease affects collagen type 1 (COL1), which leads to different manifestations depending on the mutation. There are four subtypes of OI, based on different defects in the collagen synthesis, types I, II, III, and IV.

OI type I, previously known as OI tarda, is the most common and least severe subtype, is autosomal dominant, and results in blue sclera, increased risk of bone fracture and progressive hearing loss in 50% of patients. OI type II, previously known as OI congenital, is the most severe form, it usually leads to perinatal death and is autosomal recessive. OI Type III is the most severe type among children who survive the neonatal period. The degree of bone fragility and the fracture rate vary widely. OI Type IV diagnosis often occurs later in life and usually represents the patients that do not fit into subtypes I–III (1,2).

Deafness is one of the main features of osteogenesis imperfecta and conductive hearing loss is the most common form, but mixed or pure sensorineural hearing losses are not uncommon (3). In the University of Minnesota temporal bone collection, we found one case that fits in the OI diagnosis.

CASE REPORT

We are presenting a case of a baby boy who died one day after he was born diagnosed with OI, as he had typical signs, such as multiple fractures of extremity long bones, multiple fractures of ribs, marked hypoplasia of skull and abnormal tensile strength of connective tissue, being classified as a type II OI. Death was attributed to fetal bleeding coming from umbilical arteries.

The temporal bones were removed, decalcified in TCA (trichloroacetic acid), embedded in celloidin, sectioned at 20 μ m, and every tenth slide stained with hematoxylin and eosin. The middle ear and mastoid were not included in the temporal bone removal.

Histopathologic Findings in Temporal Bones

Right and left temporal bones have similar findings. The middle ear structures are missing, except for the stapes footplate, which was mainly cartilage, with very little ossification.

The otic capsule shows very irregular endosteal bone, very thin to absent in some places. The middle layer of the otic capsule has remaining osseous architecture consistent with OI, with less septation or no septation in some areas, and plenty of chondrocytes. The septations are a thin layer of bone deposited on the periphery of calcified cartilage. There are large hemopoietic marrow areas (fig. 1A).

In the cochlea, perilymphatic and endolymphatic spaces are normal. Spiral ligament is normal and the stria vascularis shows hypotonic degeneration in all turns and basophilic concretions (fig. 1A and B). There are a normal number of spiral ganglion cells present. Reissner's membrane is normal and well positioned. The bony modiolus and osseous spiral lamina show no abnormalities. There is a microfracture at the lateral semicircular canal in the left ear (fig. 2A and B). There are no signs of new bone formation.

These temporal bones showed evidences of abnormal ossification at the otic capsule, compatible with OI, manifested also by the large number of chondrocytes as cartilage remnants, the signs of microfracture confirmed the fragility of the bone. Our findings are in agreement with those described by Schuknecht (4), and reinforce Santos (5) hypothesis of the delayed endochondral ossification in OI type II. The presence of basophilic concretions within the stria vascularis was not previously described in OI cases, but in syndromes that presents with heart defects. Despite of those defects are not common to occur associated with OI, the autopsy records were not available.

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Figure 1.

A and B: Horizontal histopathologic section of the right ear (HB 571), stained with hematoxylin and eosin, 20X magnification, showing the cochlea (1). The otic capsule has a very thin endosteal bone (→), the middle layer has few septations and is filled with hemopoietic bone marrow (■). There are large remnants of cartilage (◆). The perilymphatic and endolymphatic spaces are normal. The stria vascularis is hypotrophic and shows basophilic concretions (⇐).

Figure 2.

A: Horizontal histopathologic section of the left ear (HB 571), stained with hematoxylin and eosin, 10X magnification. 1: cochlea; 2: stapes footplate 3: lateral semicircular canal. B: Horizontal histopathologic section of the left ear, stained with hematoxylin and eosin, 100X magnification, showing the microfracture (↙) in the lateral semicircular canal (3).