OTOLOGIC IMPAIRMENTS IN ACHONDROPLASIA: A NOSOLOGIC ASSESSMENT

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Achondroplasia is an autosomal dominant dysplasia of the skeleton; the main features are shortness of the limbs, cranio-facial and odonto-maxillary anomalies and prominent forehead.

Hearing disorders in achondroplasia require a methodical analysis of their clinical and pathogenetical characteristics. The hearing disorders may differ largely from a mild to a severe congenital sensory-neural hearing loss or from a fluctuant to a permanent conductive hypoacusis. This large variability suggests a classification problem.

The literature is very scanty about this subject. We dare say that ear involvement in achondroplasia may lead to two different syndromes:

1. Congenital dysplasia syndrome
2. Tubal-tympanic syndrome

The former includes permanent hearing losses, both sensory-neural and conductive or mixed, due to the dysplasia of the labyrinthine bone or of the ossicular chain; the latter is referred to hypoacusis following inflammatory diseases of the tubo-tympanic lining.

1. Congenital dysplasia syndrome

It is useful to remember that temporal bone has a membranous and cartilaginous development; the latter concerns the labyrinthine capsule and the ossicular chain. The development of the labyrinthine capsule and the ossicular chain is different from that of any other bony structure of the body.

The primordium of the labyrinth, which is of neural ectodermic origin, faces the mesenchymal tissue which goes towards a definite transformation following a particular pattern after the appearance of fourteen ossification centers. In this area the embryonic cartilage changes into bone through its previous complete differentiation into cartilage. But the stage of this differentiation is not uniform everywhere, leaving a layer of embryonic avascular cartilage between the outer
periosteal layer and the inner endosteal layer. This distribution will be life-long definite.

Differentiation of the ossicles has many analogies with that of the labyrinthine capsule. They have some areas of endochondral cartilaginous tissue with characteristics of immaturity. Anyway they reach complete development during the fetal stage.

The noxa which is responsible for achondroplasia seldom interests these structures, through an alteration of the primary chondrogenesis.

Many authors have found that some areas of ossification are mixed with others of osteosclerosis; the endosteal layer loses its sharp outline, the ossicular chain changes its shape and structure. There are no congenital alterations in other areas of the temporal bone, particularly in the auditory tube.

The likeness between the bony development of the long bones and that of the temporal bone does not correspond to equivalent clinical manifestations: the growth changes of the temporal bone are very rare while the changes in development of the long bones are constant.

The rarity of such manifestations of the labyrinth and of the middle ear in achondroplasia may be due to two factors:

. there is a great difference in the time of development between the temporal bone and the long bones;
. the structures of the temporal bone are definite at birth and are constant for the life span (while the long bones change their development after birth).

The diagnosis of all the malformations of the ear in achondroplasia is based upon three parameters:

. audiological assessment. The sensory-neural hearing loss may vary from mild to severe deafness, due to the malformation of the labyrinth. A conductive or mixed hearing loss may be found when there are malformations of the ossicular chain; in this case the hearing loss is permanent in time, while that due to tubal-tympanic disturbancies is fluctuant.
. clinical findings. The otoscopic examination is negative in cases with sensory-neural deafness. When there is conductive hearing loss some dysmorphic features of the malleus may be observed: the manubrium may be stumpy, thinner than usual, shorter or horizontally retracted, showing a deformity of the anterior process.
. radiographic evaluation. The radiologic diagnosis is based upon linear and pluridirectional tomography; the last release of the CT scan is able to explore the smallest structures of the temporal bone, defining all the dysmorphic changes of the ossicular chain and the labyrinth.

2. Tubal-tympanic syndrome

Serous otitis is the major feature, while otorrhea and its chronic sequelae are less frequent. The clinical findings are similar to those of