

Otoradiological evaluation of patients with aural atresia: a retrospective study

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Abstract

Patients affected by aural atresia and microtia are frequently evaluated by multiple specialists (ENT, maxilla-facial and plastic surgeons, specialist in audiology and phoniatrics) and most of them require a radiological evaluation for the correct rehabilitative and surgical planning. The present study, including 190 patients currently in follow up at the Otorhinolaryngology Clinic of the Azienda Ospedale Università Padova, aim to identify the timing of execution of the CT in our cohort of patients. Moreover, the prevalence of vascular, auricular, mandibular and central nervous system anomalies detectable by means of CT will be discussed, focusing on the possible clinical impact of the detection of these malformations. In our cohort, most patients underwent imaging in the first year of life. The most frequently reported anomalies are those of the posterior labyrinth and the mandibular condyle. Even though a limited amount of patients presented carotid artery and cervical vertebrae anomalies, they should be remembered for their clinical relevance.

Keywords: Atresia auris, microtia, CT, ear, otology.

Introduction

Congenital anomalies of the external ear can involve the pinna (resulting in microtia) or the external auditory canal (resulting in aural atresia). These auricular defects in some cases can be only the first of multiple abnormalities affecting nearby districts, as in oculo-auriculo-vertebral spectrum (OAVS) (OMIM 164210). (Kokavec 2006) OAVS is a rare spectrum of heterogeneous congenital malformations (male:female ratio 3:2, incidence 1:3500 – 5600 live births) (Poswillo 1974, Grabb 1952, Rollnick 1987), in which the craniofacial structures derived from the first and second pharyngeal arches are incompletely developed on one (85% of cases) or both sides (Grabbe 1952, Mastroiacovo 1995). Even though the pathogenesis is not identified yet, abnormal embryonic vascular supply (Ottaviano 2006), haematomas and drugs used in the early phases of gestation have been supposed to have a role in causing these anomalies (Hartsfield 2007).

In other cases, the external ear malformations are isolated condition (prevalence 0.83 – 17.4/10,000 lives) (Forrester 2005, Mastroiacovo 1995, Suuturla 2007). Most frequently, the disease is unilateral (in 79 – 93% of cases) affecting the right side (60%), but both sides may be affected at different degree (Forrester 2005, Harris 1995, Mastroiacovo 1995, Suuturla 2007).

For all these reasons, it is easy to understand that patients affected by external ear anomalies require a multidisciplinary approach, since these patients may present not only ear anomalies but also facial, ocular and cervical vertebrae involvement. When all these districts are concomitantly affected, this clinical feature is classified as Goldenhar syndrome (Goldenhar 1952). Consequently, ENT, maxillo-facial and plastic surgeons can be involved, as well as ophthalmologists, geneticists and pediatricians. In this context the role of the specialist in audiology and phoniatrics is crucial since the development of au-

ditory, linguistic and oral functions should be strictly monitored. These functions should be checked and rehabilitated with the extremely important support of technicians in audiology and speech therapists. In the most severe cases, rehabilitation is not sufficient, and even surgical intervention should be considered. The most frequently performed surgical procedures are maxillofacial surgery for occlusion problems, plastic or reconstructive surgery of the auditory canal or the pinna or surgical positioning of bone conduction hearing aids.

Any rehabilitative process should be planned considering as much information as possible for the global benefit of the patient. The timing of each step should be evaluated by all the specialists involved. For example, the correction of occlusion problems might require a dental management in some cases, maxillo-facial surgery in others or even both in some other cases: consequently, a correct planning is pivotal for the success of these procedures.

Imaging is considered crucial for the evaluation of these patients, but the timing for the execution is frequently debated among experts. Computed tomography (CT) is a technology based on X-rays that are not recommended in the first phases of life, for their impact on the crystalline and their cancerogenic potential.

Nonetheless, imaging can be critical for the clinical management as well as for surgical planning. Previous studies showed that patients affected by anomalies of the external ear may present concomitant inner ear,

central nervous system (Brotto 2017, Davide 2017, Manara 2015), and salivary glands malformations (Brotto 2018). All these anomalies might have a tremendous impact on patients' life, such as the possible increased risk of meningitis or the impairment of oral functions.

In order to analyze what kind of information revealed imaging about the patients currently in follow up at the Otorhinolaryngology Clinic of the Azienda Ospedale Università Padova, a retrospective study based on clinical and radiological reports was conducted.

Material e Methods

In the present study the clinical and radiological reports of the patients currently at follow up at the Otorhinolaryngology Clinic of the Azienda Ospedale Università Padova were considered.

Only patients who underwent CT before the age of six were selected.

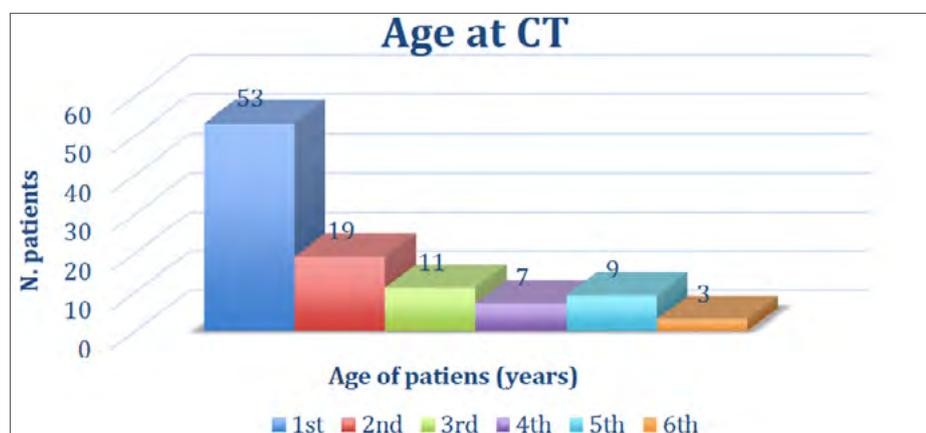
Among the 190 patients presenting aural atresia, 102 underwent CT in the first five years of life.

Results

In our cohort, 53/102 (52%) patients underwent CT in the first year of life, 19/102 (19%) in the second, 11/102 (11%) in the third, 7/102 (7%) in the fourth, 9/102 (9%) in the fifth, 3/102 (3%) between 5 and 6 years old (see Figure 1).

The patients were 59 males and 43 females. The mean age at the moment of the CT was $1,108 \pm 1,475$ years old.

Figure 1: Schematic representation of the number of patients who underwent CT in the first six years of life.



Six out of 102 (6%) patients presented anomalies of the course of the internal carotid artery, 4/102 (4%) presented anomalies of the cervical vertebrae, 13/102 (12%) presented anomalies of the semicircular canals,

29/102 (28%) anomalies of the course of the VII cranial nerve and 29/102 (28%) presented anomalies of the mandibular condyle (see Figure 2).

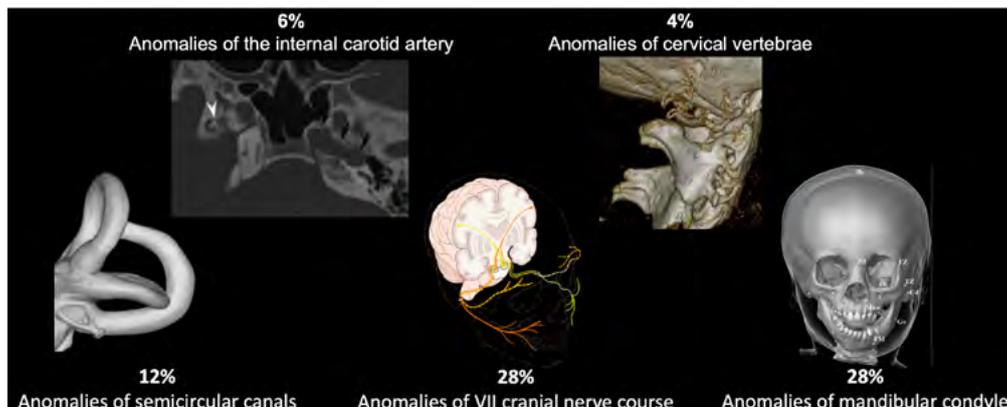


Figure 2: Graphic representation of the involvement of semicircular canals, VII cranial nerve course, mandibular condyle, internal carotid artery and cervical vertebrae.

Discussion

Aural atresia and microtia are well known faces of the OAVS. These pathologies are known to involve the structures derived from the first and second pharyngeal arches as characteristic clinical features such as aural atresia/microtia, hemifacial microsomia, epibulbar dermoids and cervical vertebrae anomalies.

The recent literature highlighted that even structures that are not strictly derived from the pharyngeal arches can be frequently altered, for example the inner ear, the brain (Davide 2017, Brotto 2017), the carotid artery (Ottaviani 2007), the cranial nerves (Manara 2015), and the salivary glands (Brotto 2018).

These small studies, with limited cohorts, highlighted these extremely useful data and that some of these anomalies can only be detected by means of the MRI and CT imaging. Evaluations on large cohorts are still missing, though.

Our study involved the clinical and radiological reports of 190 patients. Among them, the 54% underwent a CT in the first six years of life. This percentage can be an underestimation, since a part of the global cohort may be patients who never performed radiological

imaging (for example adults for whom the CT imaging was not available in their childhood).

Most of the patients who underwent CT imaging (52%) performed the exam in the first year of life, while the percentage is reduced in the following years.

This may be due to several reasons. Parents are frequently motivated to understand the clinical status of the child soon after birth, in order to better plan the future of their son/daughter. The radiology seems to give them a clearer understanding of what their child's future will be. Moreover, CT scans nowadays require only few seconds for a full head imaging (around 30 seconds). Since the whole exam can be performed in few minutes, in toddlers the CT imaging can be executed while the child is spontaneously sleeping and this is easier in the first months of life.

Among these patients, 28% presented anomalies of the mandibular condyle; this information can be extremely useful while evaluating the maxilla-facial and dental status of the child, as well as the risk to develop dental occlusion problems. The identification of mandibular condyle anomalies may impact the dental management of the child not only in the first years of life but even in the adoles-

cence. In addition, 3D reconstruction can help in the surgical planning for a harmonious aesthetic reconstruction of the face (Manara 2016).

The same percentage of patients also present anomalies of the course of the VII cranial nerve. The development of the VII cranial nerve is extremely influenced by the presence of anomalies of the external and middle ear as well as the mastoid. In case of surgery of the auricular region, the identification of the course of the VII cranial nerve is a pivotal information both in emergency procedures and in planned reconstructive surgery. Knowing that in around one third of patients with auricular atresia the course of the VII cranial nerve is abnormal is extremely useful in this context.

The 12% of patients also presented anomalies of the semicircular canals. Also, these structures, in particular the lateral semicircular canal, are surgical landmarks in ear surgery. Moreover, even though the impact of posterior labyrinth congenital anomalies on the vestibular function is not fully characterized, we cannot exclude that these anomalies might have an impact on the balance of the subject.

A small, but not negligible, number of patients (6%) presented anomalies of the carotid artery, that was found absent, hypoplastic or with altered course. This information should be considered in the planning of otologic and facial surgery. In the past, these anomalies were also considered as a possible explanation of the pathogenesis of the OAVS (Ottaviano 2007), but the small prevalence of these malformation seems to exclude this hypothesis.

Finally, 4% of patients presented anomalies of the cervical vertebrae. These malformations may range from fusion of the vertebrae to morphological abnormalities leading to

abnormal posture. Moreover, the possible presence of these anomalies should be remembered in case of life support procedures, since neck hyperextension is frequently performed and the presence of these anomalies can impair the maneuver or even force fractures of the spine.

The abovementioned data can be extremely useful in the clinical practice but some weak points of the present study should be mentioned.

This study is based on radiological reports and not on direct evaluation of the imaging, so several bias can be present: multiple observers, methodologies and expertise are involved. Consequently, even if the number of patients involved is extremely high for this rare pathology, it is even possible that the reported percentages can be underestimated.

Moreover, radiological examination of patients with craniofacial anomalies requires an advanced expertise in the field and it is possible that the description can be extremely heterogeneous since the patients performed the imaging in different Italian hospitals.

Conclusions

A comprehensive characterization of clinical and radiological features of patients with external ear anomalies is far to be achieved.

In our opinion, an early radiological evaluation by means of CT in the first years of life is feasible and can provide useful information for the clinical and surgical management of these patients.

The availability of new imaging techniques, such as the cone beam CT, that can provide equal imaging quality with low radiation dose, should be recommended especially in the first phases of life.

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