Chapter 3

Hearing Loss in Congenital Microtia

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Additional information is available at the end of the chapter

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Abstract

Congenital microtia occurs in approximately one in 10,000–20,000 live births as a result of the aberrant development of the first and second branchial arches. However, the exact pathogenesis of microtia remains unknown; it is considered a multifactorial disease where both environmental and genetic factors play a role. Microtia and aural atresia are known to be associated with conductive or mixed hearing loss caused by the developmental failure of the auricle, the external auditory canal (EAC), and middle ear structures. Cholesteatoma and mandibular dysplasia are also known to occur in microtia and atresia, as well as rare conditions, such as facial nerve paralysis, chorda tympani dysfunction, and inner ear deformity. The first branchial arch is the origin of the malleus head and the incus body as well as of the mandible, and the second arch derivatives include the stapes bone, the long process of the incus, and the manubrium of the malleus. It has been reported that the grade of microtia and the severity of middle ear abnormalities are correlated, and it is thought that better development indicates more developed middle ear structures. The existence of additional structural anomalies is suggestive of a broader developmental problem in most patients with microtia. This chapter will focus on hearing loss and structural anomalies in congenital microtia.

Keywords: microtia, atresia, facial nerve, taste disorder, hearing

1. Introduction

Congenital microtia occurs in approximately one in 10,000–20,000 live births as a result of aberrant development of the first and second branchial arches [1]. The exact pathogenesis of microtia remains unclear, but it is considered to be a multifactorial disease in which both environmental and genetic factors are thought to be associated with its pathogenesis. Microtia and aural atresia are known to be associated with conductive or mixed hearing loss and are caused by developmental failure of the auricle, the external auditory canal (EAC), and structures

of the middle ear [2, 3]. Other complications, such as cholesteatoma and mandibular dysplasia, are also known to occur in microtia and atresia, as well as rare conditions such as inner ear deformity, facial nerve paralysis, and chorda tympani dysfunction [4, 5]. It is well known that the first branchial arch is the origin of the head of the malleus, body of the incus, and the mandible, and the derivatives of the second arch include the stapes bone, the long process of the incus, and the manubrium of the malleus [2, 3]. Developmental abnormalities of the first and second branchial arches can give rise to congenital microtia and atresia, which have a significant effect on the development of the ear, including the course of the facial nerve.

Most children with congenital microtia are identified by an obvious anomaly at birth; however, they do not always receive the necessary medical care. Although otolaryngologists frequently examine only the hearing level of patients with microtia, those born with both microtia and aural atresia have a complex craniofacial condition that may affect all aspects of their lives, requiring up-to-date and unbiased patient information.

Patients with microtia are unable to undergo corrective plastic surgery until they are in their early teens since their rib bones need to be of a sufficient size for harvesting in order to create an adequately sized graft. Otolaryngologists are responsible for providing care and support to them throughout their lives.

In this chapter, we present the clinical characteristics of congenital microtia and atresia and associated complications observed at our hospital for the benefit of medical staff involved in the care of patients with these conditions.

2. Clinical characteristics and embryology of congenital microtia

The grade of microtia and the severity of middle ear abnormalities are correlated, and it is thought that better development indicates more developed middle ear structures. We investigated 172 patients (191 ears) who underwent reconstructive surgery for their external ear malformations at our university. Consistent with a previous study [1], there was a predominance of right-sided microtia (62%) and boys (64%) (**Table 1**). Although the basis for the sex discrepancy in microtia incidence has not been determined, the difference appears to be more common in Asia. Only 11% of patients had bilateral involvement. **Figure 1** shows the distribution of microtia according to severity using the Marx system proposed by Marx in 1926 [6]. Briefly, grade I microtia corresponds to a normal-shaped but small pinna, grade II to a residual vertical ridge of the tissue, and grade III to the complete absence of the pinna or the presence of only rudimentary soft tissue. The distribution was as follows: 3% was classified as grade I, 27% as grade II, and 78% as grade III (**Table 1**). It has been reported that 74% of cases of microtia are complicated with narrow external auditory atresia [2, 3], and one in five cases with congenital external auditory stenosis are complicated with ear canal cholesteatoma [6]. In our patients, narrow external auditory atresia described as complete atresia was present in approximately 70%, and external auditory stenosis affected 30%.

Careful observation is required to detect external auditory stenosis patients with ear canal cholesteatoma.

Embryologically, the auricle is formed from several protuberances in the first and second branchial arches. These protuberances, known as auricular hillocks, surround the first branchial cleft, which is the space between the first and second branchial arches [8]. Each of the hillocks contributes to a specific component of the auricle, and those in the second branchial arch form most of the ear structure. The external auditory canal and lateral tympanic membrane are derived from the ectoderm of the first branchial cleft and the epithelium of the middle ear cavity, which is derived from the endoderm of the first pharyngeal pouch. The ossicles develop from the mesenchyme of the proximal area of the branchial arches, and the malleus and incus both derive from the first branchial arch (mandibular area and maxillary area, respectively), while the stapes is formed from the second branchial arch.

Marx's classification	**%**
Grade I	3
Grade II	27
Grade III	78
Sex	
Male	64
Female	36
Laterally	
Right	62
Left	27
Bilateral	11

Table 1. Characteristics of patients with congenital microtia.

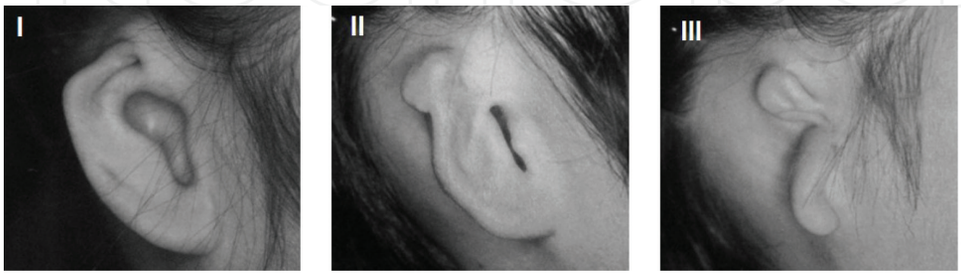

Figure 1. Marx system. Grade I: A normal-shaped but small pinna. Grade II: A residual vertical ridge of the tissue. Grade III: Complete absence of the pinna or the presence of only rudimentary soft tissue.

3. Hearing level

The pure-tone hearing average (average air conduction threshold at 0.5, 1, and 2 kHz) is used as a representative value for the hearing level, and the normal hearing range is generally defined as greater than 20 dB with an air-bone gap within 15 dB. We investigated the hearing levels for our patients with congenital microtia and compared these hearing levels with Marx's classification results (**Table 2**). Marx's classification scores did not show a correlation with the pure-tone hearing level. A previous report also found that the hearing level in microtic ears does not correlate with the degree of microtia [7].

Marx's classification	Air conduction threshold (dB)	Bone conduction threshold (dB)	Air-bone gap (dB)
Grade I	27.5	5.8	21.7
Grade II	60.9	12.8	48.1
Grade III	76.8	11.9	64.9

Table 2. Average hearing level in patients with microtia.

4. Facial nerve and chorda tympani nerve palsy

Facial nerve palsy and chorda tympani are also known to occur in some cases of congenital microtia. In our study [5], facial nerve paralysis (House-Brackmann grade more than III) and change in taste detection threshold due to chorda tympani nerve dysfunction were found in 8 and 10% of patients with microtia, respectively. We found that chorda tympani nerve dysfunction did not correlate significantly with the anatomic structure of the ear anomalies based on Jahrsdoerfer scores. On the other hand, facial nerve paralysis was significantly correlated with the presence of a malleus-incus complex, a pneumatized mastoid, an incus-stapes connection, and an external auditory canal, and facial nerve paralysis patients had a higher Jahrsdoerfer score than the chorda tympani nerve dysfunction patients.

The facial nerve canal arises initially as a sulcus in the cartilaginous otic capsule, and ossification begins from two distinct sites, such as anteriorly near the apex of the cochlea and posteriorly at the pyramidal eminence, at 20 and 25 weeks' gestation, respectively. The bone progressively covers the facial nerve, and the process is usually complete by 3 months after birth. Since the mastoid process and tympanic ring grow after birth, they displace the nerve medially. Therefore, the development of the facial nerve is closely related to the development of the middle ear and the mastoid process. Meanwhile, the chorda tympani branches from the facial nerve at 5 weeks' gestation and subsequently separates the stapes primordium and the incus primordium from the hyoid visceral bar. Unlike the facial nerve, the chorda tympani in the middle ear is not encased by a bony wall. This early branching and development of the chorda tympani may be one of the reasons why our study did not show a significant correlation between chorda tympani nerve dysfunction and facial nerve paralysis; 83% of patients

with chorda tympani nerve dysfunction did not have facial nerve paralysis. In addition, there was no significant difference in Jahrsdoerfer scores for the facial nerve between those with and without chorda tympani nerve dysfunction [5]. It is speculated that facial nerve paralysis, probably including chorda tympani nerve dysfunction, does not always correspond to an anatomic abnormality of the nerve tract.

5. Management of patients with congenital microtia

Because approximately 20–60% of patients with congenital microtia are known to have associated anomalies or an identifiable syndrome [8], patients with microtia should be examined for other dysmorphic features. In our patients, although there were no cases complicated by anomalies in the kidney and spine, there were some children complicated by esophageal atresia, ventricular septal defect, funnel chest, and cleft lip and palate. Especially, symptomatic microtia, which includes Goldenhar syndrome, hemifacial microsomia, trisomy 21, trisomy 18, and Treacher Collins syndrome, may have additional associated congenital anomalies.

Gorlin et al. [9] proposed an encompassing term "oculo-auriculo-vertebral spectrum (OAVS)," which is characterized by facial asymmetry, microtia, ear and facial tags, epibulbar dermoids, microphthalmia, and macrostomia. Hemifacial microsomia, Goldenhar syndrome, and all of its associated anomalies and variations are thought to be included in this spectrum. Extracranial features include renal, cardiac, and vertebral anomalies; at present, there is no consensus on the minimal diagnostic criteria for OAVS [10]. OAVS and microtia share the following characteristics: (1) variable phenotypic expression, (2) asymmetric involvement of facial structures, (3) right-side preponderance, (4) male predilection, and (5) familial occurrence of microtia or related anomalies, such as preauricular tags and pits [10]. Thus, isolated microtia represents a milder phenotype of OAVS.

The clinical expression of congenital microtia and OAVS overlap; hence, clinicians should consider multiple medical assessments when examining patients with microtia. First, all patients with microtia should have a diagnostic ear-specific hearing assessment within the first 6 months of age, to identify hearing loss and to assess the type and severity of hearing impairment. In children with conductive hearing loss, high-resolution CT examination of the temporal bone is useful for evaluating the middle and inner ear structures when the child is of preschool or school age. Renal ultrasound, cardiovascular examination at diagnosis, and cervical spine films at the age of 3 years are also recommended [11]. Treatment for atresia should be considered in the context of hearing, speech and language development, and reconstructive surgery at approximately 10 years of age.

Since lack of landmarks, abnormal anatomies of the facial nerve and middle ear structures, and limited space for sound reconstruction, surgical correction of hearing improvement is sometimes difficult and challenging. Therefore, not only surgery but also hearing acquisition through the use of a device should be considered. To date, osseointegrated implants known as bone-anchored hearing aids (BAHA® by Cochlear) and active middle ear implants known as Vibrant Soundbridge® (VSB by Med-El) have been the most reliable method of hearing

habilitation. These devices have been shown to improve hearing outcomes and quality of life in patients with microtia who might not otherwise benefit from traditional hearing aids. However, in order to use these implants, patients need to underwent surgery, and the portion of the implant exposed to open air has a risk of infection.

More recently, a new hearing device utilizing cartilage conduction has been developed [12]. Since the transducer is not necessarily fixed with pressure, the attachment causes no pain, unlike conventional bone conduction. Moreover, this cartilage conduction device does not require surgery. Cartilage conduction hearing aids have a potential as a useful amplification device for patients with congenital microtia and aural atresia.

6. Conclusion

We conclude that longitudinal care is required for patients with congenital microtia. This care involves the precise and regular evaluation of hearing levels from birth and investigation of malformations of the external auditory canal, middle ear, and inner ear, as well as cholesteatoma and abnormal occlusions occurring at predictable times in relationship with craniofacial growth and development. Microtia can be associated with other congenital abnormalities that are not obvious at birth. Furthermore, the external surface malformations frequently cause adverse psychosocial effects during children's growth process. Patients and their families should be supported in an unbiased manner when making decisions regarding which treatments are the most appropriate for the patient at a particular point of development, and this support must continue throughout the patients' life. In addition, means of hearing improvement should not simply be a difficult operation and instead involve careful consideration of the patient's interests and careful selection among the various options.

Terminology index

Atresia: The absence or closure of the external auditory canal

Auricular hillocks: Six (three-paired) mesenchymal condensations around the first pharyngeal cleft

Branchial arch: Paired structures associated with the pharynx that contribute greatly to the formation of the head and neck

Branchial cleft: The slit-like openings in the gills of fish between the branchial arches

Ectoderm: One of the three primary germ layers in the early embryo

Goldenhar syndrome: A complex congenital anomaly characterized by abnormal development of the eye, ear, and spine

Hemifacial microsomia: A congenital condition in which one or more parts of the face are underdeveloped

Pharyngeal pouch: Saclike diverticula that formed on the endodermal side between the pharyngeal arches

Primordium: An organ or part in the earliest recognizable stage of development

Mesenchyme: A type of undifferentiated connective tissue comprised of loose cells embedded in the extracellular matrix

Microtia: A congenital malformation of variable severity of the ear

Otic capsule: The cartilage that surrounds the developing otic vesicle and develops into the bony labyrinth of the internal ear

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References

[1] Luquetti DV, Leoncini E, Mastroiacovo P. Microtia-anotia: A global review of prevalence rates. Birth Defects Research Part A: Clinical and Molecular Teratology. 2011;**91**:813-822

[2] Harris J, Kallen B, Robert E. The epidemiology of anotia and microtia. Journal of Medical Genetics. 1996;**33**:809-813

[3] Okajima H, Takeichi Y, Umeda K, Baba S. Clinical analysis of 592 patients with microtia. Acta Oto-Laryngologica. Supplementum. 1996;**525**:18-24

[4] Takano K, Takahashi N, Ogasawara N, Himi T. The association of external and middle ear anomaly and mandibular morphology in congenital microtia. Otology & Neurotology. 2016;**37**:889-894

[5] Takano K, Takahashi N, Ogasawara N, Yotsuyanagi T, Himi T. Chorda tympani nerve dysfunction associated with congenital microtia. Acta Otorrinolaringológica. 2017;**137**:686-689

[6] Jahrsdoerfer RA, Yeakly JW, Guilar EA, Cole RR, Gray LC. Grading system for the selection of patients with congenital aural atresia. The American Journal of Otology. 1992;**13**:6-12

[7] Ishimoto S, Ito K, Karino S, Takegoshi H, Kaga K, Yamasoba T. Hearing levels in patients with microtia: Correlation with temporal bone malformation. The Laryngoscope. 2007; **117**:461-465

[8] Luquetti DV, Heike CL, Hing AV, Cunningham ML, Cox TC. Microtia: Epidemiology and genetics. American Journal of Medical Genetics. Part A. 2012;**158**:124-139

[9] Gorlin RJ, Cohen MM, Hennekam RCM. Syndromes of the Head and Neck. 4th ed. New York: Oxford University Press; 2001. p. 1344

[10] Shonka DC Jr, Livingston 3rd WJ, Kesser BW. The Jahrsdoerfer grading scale in surgery to repair congenital aural atresia. Archives of Otolaryngology – Head & Neck Surgery 2008;**134**:873-877

[11] Heike CL, Hing AV, Aspinall CA, Bartlett SP, Birgfeld CB, Drake AF, Pimenta LA, Sie KC, Urata MM, Vivaldi D, Luquetti DV. Clinical care in craniofacial microsomia: A review of current management recommendations and opportunities to advance research. American Journal of Medical Genetics. Part C, Seminars in Medical Genetics. 2013;**163**:271-282

[12] Nishimura T, Hosoi H, Saito O, Miyamae R, Shimokura R, Matsui T, Iwakura T. Benefit of a new hearing device utilizing cartilage conduction. Auris, Nasus, Larynx. 2013;**40**: 440-446