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CONGENITAL RIGHT MICROTIA AND ASSOCIATED LEFT PREAURICULAR SKIN TAGS, A CASE REPORT

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Case report

Key words: congenital microtia, preauricular tags, congenital abnormalities, newborn

Summary. Congenital malformations of the external and middle ear may have a genetic or an acquired background. Among the congenital malformations about 30% are associated with syndromes involving additional malformations. Also, microtia is associated with congenital defects of the external auditory canal (EAC) of varying degrees. *Case report*. In a full-term, spontaneously born infant, microtia on the right side and preauricular skin tags on the left side were noted immediately after birth. In addition to the clinical examination, additional consultations and imaging examinations were performed. A lobule type microtia of the right pinna and external auditory canal (EAC) atresia on the right side were noted, the left pinna was fully formed with multiple accessory skin tags. The audiologic examination showed normal hearing on the left side. No additional anomalies were found during diagnostic evaluation. The newborn will be followed up by an ear, nose and throat (ENT) specialist. External ear reconstruction is planned around the age of 10 years. *Discussion and conclusion.* When it comes to bilateral anomalies of the external ear, it is important to search for associated anomalies, taking into account the relatively high probability of ear malformations being associated with genetic syndromes. Imaging methods are indispensable for the planning and successful outcome of operative reconstruction and rehabilitation procedures. If normal hearing is confirmed in the contralateral ear of a newborn with isolated microtia, normal growth and development (including speech development) is expected. Family should be made aware of currently available treatment options.

Introduction

The external and middle ears are derived from the first and second branchial arches and grooves. Congenital malformations of these structures may have a genetic or an acquired background. Atresia auris congenita, involving malformation of the outer and middle ear, with the characteristic finding of an atretic external auditory canal (EAC), showed an incidence between 1:6.000 to 1:8.000, but severe malformation can be expected in 1:10.000 to 1:20.000 newborns.[1]

Gross malformation or aplasia is reported in 1:17.500 newborns. According to Schloss, the prevalence of microtia is higher; 3:10.000.[2] Microtia occurs with a higher prevalence in some ethnic populations like Navajo Indians (1/900–1/1.200 live births) and is less frequent in white populations.[3]

Among the congenital malformations about 30% are associated with syndromes involving additional malformations and/or functional loss of organs and organ's systems. Also, microtia is associated with congenital defects of the external auditory canal of varying degrees, from different grades of stenosis to complete absence of the cartilaginous and/or osseous parts of the canal, blind pouches, or tracts. These associations are explained by the common embryological origin of these structures. Other facial abnormalities may also be in association with microtia: preauricular tags, appendages, fissures, cysts or sinuses, abnormalities of the facial nerve, underdevelopment of the middle and inner ear, small ear, mandible hypoplasia, cleft lip or palate, macrostomia, facial clefts, anophtalmia, microphtalmia, epibulbar dermoids, facial asymmetry. The non-syndromal ear malformations present with anomalies of the ear without any other malformations, such as limb reduction defects, urinary tracts anomalies, cardiac defects, polydactyly, vertebral defects, holoprosencephaly.[4]

The diagnostic steps include clinical examination, audiological evaluation, genetic analysis and, especially, imaging methods. The precise description of the ear malformations by means of imaging methods is indispensable for the planning and successful outcome of operative reconstruction and rehabilitation procedures.[5]

Case report

A girl was born vaginally in 2019 at our maternity ward. Birth was uncomplicated, with Apgar scores of 10 at both 1 and 5 minutes of age. Infant birth weight was 3610 grams, body length 52 cm and head circumference 35 cm, gestational age estimate according to Farr was 40,6 weeks. The mother was 31 years old, with no history of chronic illness and gestation diabetes, gravida 2, para 1, 41/42 weeks gestational age by ultrasound estimate, no abnormalities were detected on fetal ultrasound examinations, maternal thrombocytopenia was monitored from 22 weeks of gestation with lowest value 121 000/uL and post-partum thrombocyte count was still low.

During the first physical examination of the infant after birth a lobule type microtia of the right pinna was

Figure 1. Microtia of the right ear

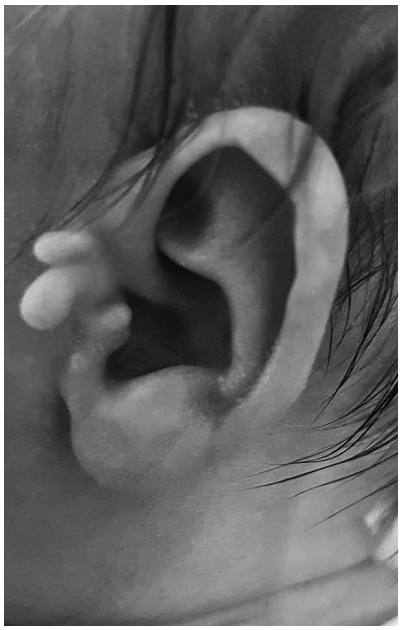

Figure 2. Associated left preauricular skin tags

noted (classified with Nagata classification)[6], with only small skin appendages visible in place of the external ear *(figure 1)*. Subsequently, during detailed physical examination no external meatus of the EAC was visible, which led to a clinical suspicion of EAC atresia on the right side. The left pinna was fully formed with multiple accessory skin tags visible around the tragus *(figure 2)*. No additional visible anomalies were found, face and neck did not appear asymmetrical. Routine complete blood count results were within normal ranges for age.

Specific diagnostic work-up of an infant with above described bilateral external ear anomalies included: audiologic testing by otoacoustic emissions (OAE), ear, nose and throat (ENT) consultation, head multi-slice computed tomography (MSCT) with extra attention given to the right temporal pyramid, brain and abdominal ultrasound, cardiological exam including heart ultrasound, ophthalmologic examination and spine X-rays. OAE testing of the left ear was normal. ENT specialist recommended follow-up and planned external ear reconstruction at the age of 10 years. Head MSCT showed the absence of EAC and auditory ossicles of the middle ear on the right side while the right inner ear was morphologically normal and no structural abnormalities of the auditory structures were present on the left side *(figure 3)*. Brain and abdominal ultrasound were normal. Cardiological exam revealed only a patent foramen ovale with a structurally healthy heart. No bulbar dermoid cysts, microphtalmia or other anomalies were found on eye examination. Spine X-rays did not show any structural abnormalities of the vertebrae.

Hospital stay was uncomplicated, there was no newborn jaundice, meals were well tolerated, stools were of normal colour and consistency and diuresis was regular, newborn cried in regular frequency.

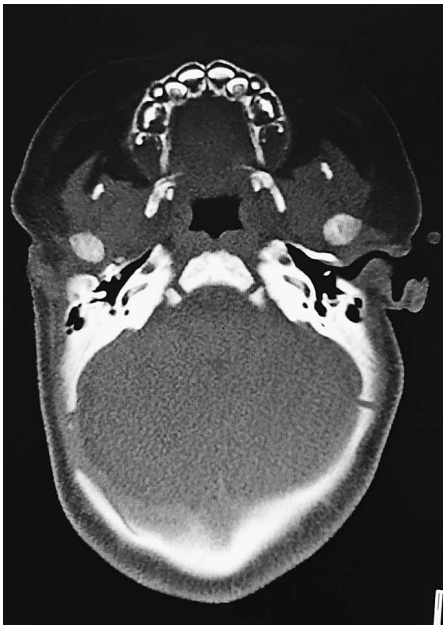

Figure 3. Head MSCT: the absence of EAC and auditory ossicles of the middle ear on the right side and no structural abnormalities of the auditory structures on the left side

Discussion and conclusions

Taking into account the bilateral ear malformations and the relatively high probability of ear malformations being associated with genetic syndromes, we considered possible malformation syndromes that might be the cause. Goldenhar syndrome (oculoauriculo-vertebral dysplasia) was the primary consideration. Goldenhar syndrome is part of the hemifacial microsomia

spectrum with facial hypoplasia, ear anomalies (especially microtia and accessory skin tags), eye malformations (bulbar dermoid cyst and microphtalmia), hemivertebrae and parotid gland dysfunction. It can cause conductive or mixed hearing loss. The underlying cause of this syndrome is still largely unknown, although a genetic cause is presumed with autosomal dominant inheritance or sporadic occurrence.[7]

However, microtia or other ear abnormalities can be found in a wide spectrum of other disorders, most commonly craniofacial microsomia and Treacher Collins. Multiple other syndromes or genetic causes have been identified and are associated with microtia in less than 50% of cases: Auriculo-condylar, Bixler (hypertelorism-microtia-clefting), Bosley-Salih-Alorainy, Branchio-oculo-facial, Branchio-oto-renal/branchio-otic, CHARGE syndrome (coloboma, heart defects, atresia choanae, growth retardation, genital abnormalities, and ear abnormalities), Fraser, Kabuki, Klippel-Feil, Labyrinthine aplasia, Meier-Gorlin, Miller, Nager, Oculo-auricular, Pallister-Hall, Townes-Brocks, DiGeorge deletion syndrome, Wildervanck (cervico-oculo-acoustic).[1,8] A wide range of diagnostic modalities was applied to uncover other abnormalities that might be present in this infant.

Many of the above mentioned multiple congenital anomaly syndromes present with diverse facial malformations, vertebral anomalies and a higher incidence of other organ abnormalities, especially kidneys and heart. A newborn with auricular deformity must undergo a detailed investigation of the craniofacial structures. A thorough examination of the skull, the face and the neck is mandatory and special attention should be given to the configuration, symmetry, facial proportions, masticatory apparatus, occlusion, hair and skin condition, sensory function, speech, voice and deglutition.[9] Other than bilateral ear malformations, there was no additional visible external malformations in this case.

Imaging techniques like high resolution computer tomography (HRCT) and magnetic resonance imaging (MRI) can be used to evaluate the structures of the ear.[10] Because of the good representation of bony structures, HRCT is more suitable for displaying the changes of the outer ear, the external auditory canal, the middle ear and the mastoid as well as osteogenic diseases, which is why we decided to use it in this case and confirmed the absence of the EAC and auditory ossicles on the right side. Hearing tests remain the most important functional investigation in patients with ear malformations. Early examination of the hearing in the apparently normal contralateral ear is important in order to detect or exclude bilateral hearing loss in newborns with unilateral atresia auris congenita.[11] Our patient displayed normal OAE testing result on the left side.

Children with outer ear anomalies should go through diagnostic audiological assessment because hearing problems at a young age may cause learning difficulties and seriously hamper language development. If contralateral hearing is adequate, leaving microtia untreated will not cause any functional problems but it may cause psychological and cosmetic issues for the developing child.[12]

It is important for the surgeon and the family to generate a cohesive plan that includes management of the ear and hearing and, ideally, involve the child in any decision-making regarding reconstructive treatment options. The hearing surgery needs to be carried out with reconstructive surgeon consultation so that future surgical options are not jeopardized.

The formation of a new auditory canal and meatus (atresiaplasty) does not lead to any significant long-term improvement in the hearing and is not routinely carried out as it can be quite problematic postoperatively, with ongoing care needed.[13] Surgical reconstruction of the external ear can be achieved either using a prosthesis (alloplastic), the patient's own tissue (autologous) or a combination of the two.[13,14] Plastic surgery of low-grade auricular dysplasia begins at the age of 5–6 years. Most of the authors recommend plastic reconstructions from the age of 8 years or 10 years because the contralateral ear is near full size, the costochondral cartilage is of adequate size and the patient is able to understand the reconstructive options.[14]

Creating an autologous construct using the patient's own costochondral cartilage is currently the most popular reconstructive technique. This operation is dependent on the patient's size. It is usually performed around the age of 10 years because it corresponds to a chest circumference of 60 cm at the inferior portion of the sternum and it tends to mean that there is enough cartilage to harvest for the reconstruction. As an alternative to this, allogenic materials (e.g. porous polyethylene) can be used in reconstruction. These materials permit reconstruction in one or two steps and can be successfully implemented at age 3 and above.[15] Ear, nose and throat (ENT) specialist was consulted on this case and recommended external ear reconstruction around the age of 10 years.

In addition, there has been recent advancement in recreating the ossicular chain using 3-D printing technology, namely 3-D printing of incudal replacements with polylactic acid (PLA)[16] as well as novel methods of recreating earlobes by using in vitro engineered patient-specific ear-shaped cartilage. This has proven to be fast and efficient, but the method is still in development.[17]

In recent years, the technological advancement of ultrasound has enabled better visualization of different fetal abnormalities. As ultrasound screening of the external ear can be used as one of the indicators of prenatal diagnosis of fetal chromosomal abnormalities which might help in decreasing the birth defects it should ideally be performed in the period between 20 and 24 weeks of gestation.[18]

Furthermore, ultrasound of the kidneys, heart and brain can be performed to discover other anomalies, but

there are opposite opinions on this by different authors. Most of the studies suggest that ultrasound screening is not necessary if a minor and isolated anomaly of the ear is present. However, ultrasound screening, especially renal ultrasound is indicated if it is accompanied by other dysmorphic features, suggestive family history or known risk factors such as gestational diabetes. It was found that syndromic children with microtia demonstrate a higher crude rate of renal ultrasound abnormalities (22%) than children with isolated microtia (7%).[19,20] There were no abnormalities visible on the abdominal ultrasound in our case and the heart ultrasound showed only a patent foramen ovale. As well as microtia, our patient also had multiple preauricular accessory skin tags on the contralateral side, which is described as an associated anomaly in 30.5% of cases.[21]

In conclusion, multiple bilateral ear abnormalities require a thorough diagnostic work-up, but they are not necessarily part of a syndrome, as seen in this case where we found no additional anomalies during our evaluation. Given that our patient displayed normal hearing on the left side, the main problem for the child is mostly cosmetic in nature and the decision when to approach reconstruction should be carefully considered, with regard to psychological and developmental factors. Optimal care is provided by multiple disciplines, including a reconstructive surgeon, an otologic surgeon, an audiologist and a pediatrician.

References

1. Alasti F, Van Camp G. Genetics of microtia and associated syndromes. J Med Genet 2009;46(6):361–9.

2. Schloss MD. Congenital anomalies of the external auditory canal and the middle ear. New York: Oxford University Press; 1997;119–24.

3. Brent B. The pediatrician's role in caring for patients with congenital microtia and atresia. Pediatr Ann 1999;28:374–8.

4. Stoll C, Alembik Y, Ditt B, Roth MP. Associated anomalies in cases with anotia and microtia. Eur J Med Genet 2016; 59(12):607–14.

5. Braun T, Hempel JM, Berghaus A. Developmental disorders of the ear in children and adolescents: conservative and surgical treatment options. Dtch Artzebl Int 2014;111(6):92–8.

6. Shibazaki-Yortozuya R, Nagata S. Preferential associated malformation in patients with anotia and microtia. Journal of Craniofacial Surgery 2019;30(1):66–70.

7. Gendron C, Schwentker A, van Aalst JA. Genetic advances in the understanding of microtia. J Pediatr Genet 2016;5(4): 189–97.

8. Cox TC, Camci ED, Vora S, Luqueti DV, Turner EE. The genetics of suricular development and malformation: new findings in model systems driving future directions for microtia research. Eur J Med Genet 2014;57:394–401.

9. Klockars T, Rautio J. Embryology and epidemiology of microtia. Facial Plast Surg 2009;25(3):145–8.

10. Qin FH, Zhang TY, Dai P, Yang L. Anatomic variants on computed tomography in congenital aural atresia and stenosis. Clin Exp Otorhinolaryngol 2015;8(4):320–8.

11. Ohl C, Dornier L, Czajka C, Chobaut JC, Tavernier L. Newborn hearing screening on infant at risk. Int J Pediatr Otorhinolaryngol 2009;73(12):1691–5.

12. Mileshina NA, Osipenkov SS, Tavarkiladze GA. The management of patients with congenital malformations of the external and middle ear. Vestn Otorhinolaringol 2018;83(4): 51–5.

13. Jonathan J Cubitt, Ling-Yun Chang, Derek Liang, John Vandervord and Damian D Marucci. Auricular reconstruction. J Pediatr Child Health. 2019;55:512–7.

14. Bly RA, Bhrany AD, Murakami CS, Sie KC. Microtia reconstruction. Facial Plast Surg Clin North Am 2016;24(4): 577–91.

15. Bonilla A. Pediatric microtia reconstruction with autologous rib: personal experience and technique with 1000 pediatric patients with microtia. Facial Plast Surg Clin North Am 2018; 26(1):57–68.

16. Kamrava B, Gerstenhaber J, Amin M, Har-el Y, Roehm P. Preliminary model for the design of a custom middle ear prosthesis. Otol Neurotol 2017;38(6):839–45.

17. Zhou G, Jiang H, Yin Z, Liu Y, Zhang Q, Zhang C et al. In vitro regeneration of patient-specific ear-shaped cartilage and Iis first clinical application for auricular reconstruction. E Bio Medicine 2018;28:287–302.

18. Wei J, Ran S, Yang Z, Lin Y, Tang J, Ran H. Prenatal Ultrasound Screening for External Ear Abnormality in the Fetuses. BioMed Research International. 2014;2014:1–5.

19. Kosling S, Omenzetter M, Bartel-Friedrich S. Congenital malformations of the external and middle ear. Eur J Radiol 2009;69(2):269–79.

20. Koenig J, Amoils M, Grade M, Chang K, Truong M. Renal ultrasound abnormalities in children with syndromic and non-syndromic microtia. Int J Pediatr Otorhinolaryngol. 2018; 113:173–176.

21. Van Nunen DP, Kolodzynski MN, van den Boogaard MJ, Kon M, Breugern CC. Microtia in the Netherlands: clinical characteristics and associated anomalies. Int J Pediatr Otorhinolaryngol 2014;78(6):954–9.

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PRIROĐENA DESNOSTRANA MIKROTIJA I PRIDRUŽENI LIJEVOSTRANI PREAURIKULARNI PRIVJESCI, PRIKAZ SLUČAJA

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Prikaz slučaja

Ključne riječi: mikrotija, preaurikularni privjesci, prirođene malformacije, novorođenče

Sažetak. Kongenitalne malformacije vanjskog i srednjeg uha mogu imati genetske ili stečene uzroke. Oko 30% kongenitalnih malformacija povezano je sa sindromima koji uključuju druge pridružene malformacije. Također, mikrotija je povezana s prirođenim defektima vanjskog zvukovoda različitih stupnjeva. Prikaz slučaja. U donošenog i spontano rođenog novorođenčeta, odmah po rođenju zamijetili smo desnostranu mikrotiju i lijevostrane preaurikularne privjeske. Osim kliničkog pregleda, provedena je slikovna obrada kojom se s desne strane našla izolirana mikrotija lobularnog tipa i atrezija zvukovoda, a s lijeve strane formirana uška s preaurikularnim kožnim privjescima. Audiološki pregled pokazao je normalan sluh na lijevoj strani. Dijagnostičkom obradom isključene su anomalije drugih organskih sustava te je dijete uključeno u daljnje praćenje otorinolaringologa. Operativna korekcija desnostrane mikrotije predviđena je u dobi do 10 godina. Rasprava i zaključak. Kada je riječ o obostranim anomalijama vanjskog uha, s neonatološkog aspekta je važno tragati za pridruženim anomalijama, uzimajući u obzir relativno visoku povezanost malformacija uha s genetskim sindromima. Slikovne dijagnostičke metode slike neophodne su za planiranje i uspješan ishod operativne rekonstrukcije i rehabilitacijskih postupaka. U slučajnu urednog nalaza ispitivanja sluha na kontralateralnoj strani u novorođenčeta s izoliranom mikrotijom, očekuje se normalan rast i razvoj (uključujući razvoj govora). Obitelj treba uputiti u mogućnosti suvremenog liječenja.