(Slide 1) My name is Emily Tignor, I am one of the ENT interns. I want to start by thanking Dr Mukerji for the time and expertise she has lent to me in making this talk. Today I will be talking about microtia, which is the abnormal development of the external ear.

(Slide 2) Topics that I will be discussing today include embryology of the pinna, etiology of microtia, how microtia typically presents, anomalies associated with microtia, classification schemes for microtia and the management of the microtic ear.

(Slide 3) Let’s begin with auricular embryology. The external ear begins to form in week 5. This is after the inner ear has already started to take shape in week 3. The external ear comes from the 1st and 2nd branchial arches also known as mandibular (1st) and hyoid (2nd) branchial arches. These arches are mesoderm. This can also be classified as hillocks 1 through 6. The 1st through 3rd hillocks arise from branchial arch 1 where the 4th through 6th hillock come from the 2nd branchial arch. There are many theories that describe which hillock will become which final auricular structure. Today I will discuss the classic theory as well as mentioning another.

(Slide 4) The traditional theory states that the 1st hillock becomes the tragus, the 2nd and 3rd hillocks become the helix, the 4th and 5th hillocks become the anti helix and the 6th hillock becomes the anti tragus and lobule. Other theories state that the majority of the auricle arises from hillocks 4 through 6 (which is the 2nd branchial arch). These theories state that as much as 85% of the auricle can arise from the 2nd branchial arch where the 1st branchial arch really is concentrated in forming the tragus.

(Slide 5) Here is a drawing that I made of the hillocks and the traditional theory of what they become. The hillocks all start out in this upside down horseshoe shape and morph into the appropriate final structures as you can see on the picture.

(Slide 6) There is more to the embryology of the ear than simply the story of branchial arches. The ear does not begin in its final position. It must undergo a migration. The ear begins to form in an anterior position and must migrate dorsally and cephalically during weeks 8 through 12 to end up in the correct and final position at 20 weeks of gestation. This migration is happening with the formation of the mandible. This is key when we will later look at disorders associated with microtia and we will see that the mandible formation is closely linked with external ear formation. The external auditory canal
Microtia: The Abnormal Development of the External Ear

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(EAC) is also important to microtia. It forms from the 1st branchial cleft. Now this is no longer mesoderm, rather from ectoderm. The EAC will open in weeks 3 through 4 from an invagination of the otic placode. The meatus will later plug with epithelial nest cells in weeks 4 through 5. Recanalization will not happen until week 21 and will finish with the formation of the tympanic membrane in week 28.

(Slide 8) Now let’s move on and talk about what can happen in this complex embryology to cause a malformed ear. Many theories talk about etiologies of microtia and no one theory is completely accepted as the cause of microtia. Vascular etiologies are often blamed. This will describe, as we will discuss later, the often unilateral component of microtia. The stapedial artery gives the blood supply to the area during development and a stapedial artery insult (either through occlusion, constriction, or failure of the artery to properly develop) can lead to microtia. Teratogens can also be responsible for microtia. Some of these teratogens include retinoic acid inhibitors like isotretinoin. This is a famous teratogen that is used as an acne treatment. It can affect the external and inner ear as well as the EAC. The typical presentation of microtia in a isotretinoin case are malformed, low set, small ears. Other teratogens include thalidomide. This classically causes the flipper limb deformities. However, it can also affect the ear. This is especially damaging if taken in the 1st month of pregnancy. It can lead to external ear, middle ear, inner ear and EAC malformations. CellCept or mycophenolate mofetil is an immunosuppresive drug that can lead to teratogenic affects on the external ear. This drug typically only affects the external ear and the EAC leading to low set, microtic ears. Genetic causes can also be behind microtia. Chromosomal abnormalities including Turner syndrome, Down Syndrome, and other trisomies including 13, 15, 18, and 22 are associated with microtia. Other mutations can also include microtia. These are notably failure in neural crest cell migration mutations including Treacher Collins syndrome.

(Slide 9) Now that I’ve talked all around microtia, let’s define it completely. The definition of microtia is, “the abnormal development of the external ear that results in a malformed auricle.” It is relatively uncommon with an incidence of 1-20 in 10,000 live births. Microtia is also strongly associated with hearing loss. In fact, 80% of patient with microtia will have conductive hearing loss and 20% will have sensory neural hearing loss, according to one study. Now I will say, that this particular study was only looking at more severe forms of microtia. The real issue with microtia is a cosmetic and psychological issue. Children can find themselves with a stigma and can be ostracized for an abnormal ear which can be a big issue to a child and to a family. This becomes more of a concern as the child ages. This is always important for us to keep in mind.

(Slide 10) Microtia is more common in males with a male to female ration or 2.5 to 1. It is also more common in certain races including Japanese, Hispanic, and Native American. Interestingly, it is more common in higher altitudes. One study compared the incidence and prevalence of microtia in South American cities of high altitude and low altitude and found that both incidence and prevalence of microtia was higher in the high altitude cities. This is explained by the thought that these pregnant women are exposed to relative hypoxia which could affect the external ear development. It is also for women who have had more than 4 pregnancies to have a child with microtia than women with less than 4 pregnancies.

(Slide 11) Microtia is often mostly unilateral (90% of the time) leaving only 10% of cases to be bilateral. Right side is more commonly affected than the left (60% right sided). Hearing loss is commonly present in the affected ear, but 6% of cases show hearing loss in the non affected ear. In
summary of the presentation, the most common case would be a boy with right sided only microtia with ipsilateral hearing loss.

(Slide 12) Let’s move on to associated anomalies with microtia. 50% of cases of microtia are associated with some other anomaly. This is a large number. Therefore, it is important when you see a child like this in your clinic that you are on the lookout for any associated anomalies. The most common associated anomaly is congenital aural atresia (CAA). CAA is EAC atresia which come from a failure to recanalize during the EAC development. This is actually present in almost every case of severe microtia (97%). CAA is associated with cholesteatoma in 4-7% of cases. Typically in children with CAA a CT scan of the temporal bone will be done around age 4 to assess for cholesteatoma. Other associated anomalies with microtia include hemifacial microsomia (which is a failure of lower face development) and acrofacial dysostosis.

(Slide 13) Goldenhaar syndrome, also known as oculofaciovertebral dysplasia, as you can see in this picture, is associated with preauricular nodes, epibulbar dermoids and mandibulohypoplasia which is commonly unilateral.

(Slide 14) Treacher Collins Syndrome, which I mentioned earlier is a failure of neural crest cell migration having to do with a mutation on the TCOF1 gene which produces the treacle protein. This is also known as mandibulofacial dysostosis and is associated with cleft palate, conductive hearing loss, absent parotids, lack of air cells in mastoid cavity, hypoplastic orbital rims and hypoplastic facial bones.

(Slide 15) Microtia can be classified in many different ways. Many schemes exist which are most helpful for surgical approaches, but today I will talk mainly about the the traditional and original microtia classification. This is known as the Marx system and was developed in 1926. A new classification system has come out in 2009 known as the Hunter classification which is almost identical to the Marx. In this picture we can see a typical type 1 deformity. These are mild deformities with all structural components of the auricle present. This is often called a “near normal” ear. The changes that are present are typically helical or may just be a smaller ear.

(Slide 16) Type 2 microtia is also known as atypical microtia. Some auricular structures are still present and mainly helical changes have occurred. In type 2, no CAA is found but there may be some EAC stenosis. These deformities may have a large range from a mini ear deformity to cup ear deformities.

(Slide 17) Type 3 microtia is classic microtia. Few auricular structures are still remaining (none recognizable) and can be classified by what is left- conchal remnant or lobular remnant remaining. Lobular remaining is most common. In these cases, the lobule is typically anterior and may appear as a skin tag. These patients are where we see the 97% CAA associated and where we start to see higher incidences of hearing loss.

(Slide 18) The final stage of microtia is type 4, also known as anotia. Here there are no auricular structures left and always CAA.

(Slide 19) How do we manage patients with microtia? Important things to think about will be hearing status. The degree of microtia is associated with the degree of middle ear deformity and
hearing loss. Conductive hearing loss is common in the microtic ear, which we discussed earlier. 80-90% of patients with hearing loss and microtia will have hearing loss due to conductive means leaving 10-20% of hearing loss due to sensory neural causes. As stated earlier, the non-microtic ear can have hearing loss - this is common in Goldenhar syndrome. If hearing loss is present, in order to preserve normal speech and language development, it will be important to protect the normal hearing ear. This means lowering the thresholds for PE tubes. Bilateral hearing loss can be an indication for bone anchored hearing aid placement. Important to remember, children with microtia need to have their hearing evaluated and monitored.

(Slide 20) Dealing with the cosmetic portion of microtia is also important. With any surgical problem, there is always a non-surgical option in management. No exception with microtia. Observation can be a good option for patients - especially children with type 1 microtia. It is also always chosen for children up to the ages of 5-8 years. This will allow for the child to grow enough for reconstruction to be possible. The advantages of observation are in the lack of risk associated. There will always be a possibility for future reconstruction if desired. Disadvantages are cosmetic and psychosocial. It this is chosen, the main focus on the child needs to turn to hearing and speech.

(Slide 21) Prosthetics are also good choices for microtia management. Two different types of prosthetics exist - adhesive and magnetic. Adhesive prosthetics may require the removal or partial removal of the microtic ear so that the prosthesis will have a good surface on which to stick. Magnetic prosthetics do require a surgery to remove the microtic ear and to defat the area and to place magnetic anchors. This is a 2 step surgical procedure. Prosthetics are typically used if the patient has failed reconstruction or cannot have reconstruction done.

(Slide 22) The main advantage of prosthetics are that they are the best cosmetic result possible - these can be identical to a normal ear. Disadvantage of adhesive prosthetics are that future reconstruction can be difficult if part of the microtic ear was removed, dislodgment is possible (especially with sweating and physical activity - sports), the ear must be removed at night, and cost. The cost of a prosthetic ear can be around 8,000 dollars and the ears do not last forever. Some insurances will cover this, but many will not. Magnetic prosthetic disadvantages include no future reconstruction possible because of the removal of so much tissue to place the magnets. Every night the ear must be removed and cleaned. These ears are also as expensive as the adhesive ears and must be replaced. This also required 2 separate surgeries.

(Slide 23) The most common choice for severe microtia cosmetic management is reconstruction. Two different types of surgical reconstruction exist including autogenous rib (a 4 step procedure including donor site harvesting of rib - possibly 2 steps if steel suture is used, but this is highly uncommon) and Medpor (a porous polyethylene substance that is a more difficult procedure and requires typically a temporalis fascia flap overtop of the implant). Both surgeries are typically done around ages 5-8 because, in the case of autogenous rib procedure, the donor site (rib cage) will have grown to workable size at this time and in both procedures, the other ear will be approximately 90% of its adult size around age 6 and the normal ear is upon which the reconstruction is based.

(Slide 23) The advantages of reconstruction are cosmetic and in maintenance. The ear may even begin to grow with the body and does not require cleaning or replacing. Advantages to autogenous rib procedure include lower risk for extrusion or infection. Advantages to Medpor include
no donor site morbidity. Disadvantages to reconstruction are that major surgery is necessary, the skin flap may fail and significant scar contracture or hypertrophy may occur. Autogenous rib disadvantages are donor site morbidity and pneumothorax where disadvantages of Medpor include higher risk for extrusion or infection post op. This is especially prevalent in trauma or even a small blow to the area and often requires children to abstain from sports or other risky behaviors.

(Slide 24) Steps of the autogenous rib reconstruction include: 1- cartilage harvesting and implantation, 2- lobule transfer, 3- creation of post auricular sulcus, and 4- tragus reconstruction. Complications of the operation include hematoma which can lead to total flap failure and pneumothorax which commonly occurs during rib harvest. After the reconstruction is finished, it can be time to consider CAA repair. This happens 2 months after the microtia surgery is complete.

(Slide 25) This slide shows the different stages and results of Autogenous rib vs Medpor reconstruction.

(Slide 26) In summary, microtia is auricular failure of development. The external ear arises from branchial arches 1 and 2. The etiology of microtia is mainly sporadic and may be due to vascular insult, but also some genetic causes and syndromes are associated with microtia. Associated anomalies are present in 50% of cases of microtia and CAA is often found in microtia patients as well.

(Slide 27) Type 1 microtia is minimal deformity. Type 2 is mainly helical deformity and is known as atypical. Type 3 microtia is classic microtia with no recognizable auricular structures. Type 4 microtia is anotia- total lack of external ear. Management of microtia options include observation, prosthesis and reconstruction. While managing these patients, keeping in mind hearing status is the key to speech and language development.

(Slide 28) On an interesting note, there has some talk in the last 20 years about implanting ears that have been grown on mice. This picture was circulated around 2000 and contributed to a large craze about genetic engineering. However, this mouse was not genetically engineered to have an ear on its back. This is an immunodeficient mouse who cannot reject foreign tissue that has had a hydrogel scaffold in the shape of an ear implanted in its back. Before the scaffolding was implanted, human cells were distributed over it. The scaffold incubated on the back of the mouse provides a place for the cells to grow and eventually cartilage will take its place. This was started by Vacanti in 1997 and has not been widely used medically. Now research is being done to create the cartilage sans scaffold with simply 20,000-40,000 human cells. The other picture I have on this page is of an artist, Stelarc, who has had one of these scaffolds with his cells sprinkled on it implanted in his arm. His most recent surgery was in 2006. He has had many complications from the surgery but still plans to go ahead with implanting some of his own stem cells into it to form a more realistic lobule. He also hopes to implant a microphone in the ear so that he can whisper “sweet nothings” to the world.

Thank you for listening to my talk. I hope this has helped you to understand a little bit more about microtia.
Faculty Discussion: Shraddha Mukerji, MD

I think that was an excellent Grand Rounds presentation. I just want to reiterate a few things that Dr. Tignor said. I want everyone to understand that microtia is for the external ear only and that the grades of microtia are for the external ear only. There are different grading systems for the external auditory canal atresia. So that you will have to grade the microtia with or without external canal atresia or stenosis or whatever. There are two different things and the contemporary terms for the external ear and the external canal are all known as congenital aural dysplasias. Most of the time the external ear deformities will be associated with middle ear deformities. How much percentage of inner ear deformities do you get with microtia and middle ear deformities -- does anyone know? So it's about 9 to 20% and why is there such a low number? One reason is from different times of development and the other one is that they develop from different structures.

The other thing Dr. Tignor said is that when the ear is developing the mandible is developing as well, and if you have deformities of the external ear, the mandible will be affected as well, and the most important structure to be affected is the condyle because that’s the most anterior limit of the external ear.

Regarding surgery, though we see it as multiple stages it is important that as physicians to counsel the parents that there’s not going to be a single surgery but there may multidisciplinary surgery between Plastics, Otology and Neuro-otology, and they will require follow-up with Speech and Audiology. That's important because most of the time parents believe that one surgery will fix it all and as you know that's not the case.

References

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