Microtia

Patient Information Series – What you should know, what you should ask.

What is microtia?

Microtia is when one or both ears are abnormally small. More severe forms can also include abnormal shape and location of the outer ear. The most severe form, anotia, is when the ear is completely missing. In addition, microtia can be associated with hearing loss.

How does microtia happen?

Microtia can occur for many reasons. Certain medications and drugs have been associated including accutane, retinoic acid, thalidomide and alcohol. There are also several associated genetic syndromes. Genetic syndromes are disease caused by mutations or differences in genetic information. Genetic syndromes often cause multiple anomalies.

However, many cases of microtia are sporadic meaning the development of the condition is random and has no known cause. There are also cases of microtia that are familial, or run in the family, without an associated genetic syndrome.

Should I have more tests done?

You should ask if a specialised/advanced ultrasound of the baby during the pregnancy can be performed to see if the baby has other anomalies or if the microtia is the only issue. If there are additional anomalies, a consultation with a genetic specialist is usually recommended to determine if there may be a causative genetic syndrome. You may be offered genetic testing to see if certain genetic changes are the cause for the microtia. Amniocentesis is the most common method used for genetic testing. It is done using a thin needle that is inserted into the pregnancy to collect amniotic fluid in the sac around the baby.

There is a small risk of miscarriage (approximately 1 miscarriage for every 900 amniocentesis done) with this procedure. Not all patients choose to undergo this kind of testing. You should discuss with your doctor to determine what decision is right for you and your family. Not all conditions can be diagnosed by amniocentesis, and not all anomalies can be seen on ultrasound. However, if there are no other anomalies seen on ultrasound and invasive testing gives normal results, then the microtia is more likely to be isolated and unrelated to a genetic cause.

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What are the things to watch for during the pregnancy?

If there are no other anomalies, then you can have your pregnancy checks as normally planned. If there are other anomalies, changes in your care will depend on what they are and what they mean.

What does it mean for my baby after it is born?

Children born with microtia need to be evaluated for possible hearing loss. This will frequently include audiologic testing and may also include imaging to try to detect any abnormalities that may exist with the external auditory canal, middle, and inner ear. Often a team of specialists will be involved including geneticists, pediatric audiologists, pediatricians, and plastic surgery. Often when the child is older, cosmetic reconstruction of the ear can be considered by plastic surgery.

Will it happen again?

Patients who have a child with microtia have an increased chance of having another child with microtia, around 1 in 20. However, these odds may be different depending on the specific cause of your child's microtia. You should talk to a geneticist to see if you have an increased chance of having a child with microtia in your next pregnancy.

What other questions should I ask?

- Are there any other abnormalities on the ultrasound?
- What kind of genetic screening or testing should I consider?
- How often should I have ultrasound examinations?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?

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