Arhinia

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

What is Arhinia ?

Arhinia is an extremely rare problem of the nose. Babies with arhinia miss one half (hemi-arhinia) or their entire nose at birth. The nose area is flat and firm on palpation although small ridges or small and non-functional nostrils may be present. In the vast majority of cases, the upper jaw is small and the tear ducts at the base of the eyes and the sinuses are missing or mal developed. Furthermore, arhinia is often associated with eye or ear anomalies or can be associated with other problems with the face, the skull or the brain.

How does arhinia happen?

As there are limited number of babies born with arhinia, it is difficult to know the cause of this rare condition. When it is the only anomaly present in the baby, it usually happens by chance.

Arhinia can be found in babies with holoprosencephaly. Holoprosencephaly is a problem in which the brain fails to separate into two hemispheres. When holoprosencephaly and arhinia are present together, babies are usually not doing well due to the brain problem or a genetic problem which is the cause of both problems.

Arhinia can also be part of a genetic disease of which Bosma arhinia microphtalmia syndrome is the most common. Babies with this condition have arhinia, abnormally small or absent eyes and an underdeveloped reproductive system with abnormally low level of related hormones. This disease is caused by a change in the genetic information contained in the chromosomes of the baby. We usually have 46 chromosomes matched in pairs: 23 come from one parent and the other 23 come from the other parent. Babies with Bosma arhinia microphtalmia syndrome have a change in the information in one of the chromosome number 18.

Should I have more tests done?

You should ask if a specialised/advanced ultrasound of the baby during the pregnancy can be performed to detect other anomalies. If there are other anomalies, especially of the brain, a consultation with a genetic specialist is recommended.

A MRI may be helpful to have a better view of the face, the skull or the brain. MRI uses magnetic fields to create images of the inside of the body. It is safe to use in pregnancy.

You should also ask if an amniocentesis should be done. An amniocentesis can detect some genetic disorders like Bosma arhinia microphtalmia syndrome and genetic diseases related to holoprosencephaly. This is done by removing a small amount of amniotic fluid from the womb surrounding the fetus. There is a small risk of causing a miscarriage when doing an amniocentesis (less than 1 miscarriage for every 200 amniocentesis performed) and some women choose not to have it.

Not all genetic diseases can be identified by an amniocentesis and not all anomalies are visible on an ultrasound examination. However, when there are no other anomalies visible on

ultrasound and the amniocentesis gives normal results, then arhinia is very likely to be the only problem for the baby.

What are the things to watch for during the pregnancy?

Sometimes babies with arhinia accumulate extra amniotic fluid around them during the pregnancy because they have problem swallowing. This condition is called polyhydramnios. It can stretch the uterus too much and cause labor well before the due date. Knowing this in advance, your doctor or midwife can help with decreasing the risk of an early birth. That is why most specialists will recommend regular ultrasound examination at least every 4 weeks. If you feel that your belly is becoming big rapidly, you should contact your doctor or midwife to have this checked.

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

Babies born with arhinia as their only problem usually do well. Some babies have breathing or swallowing problems right after birth because the nostrils are absent or not functional. These babies may need oxygen or the placement of tubes to help their breathing. Because of this, it is advisable to discuss the best place for your delivery with your doctor or midwife. Feeding via a tube is usually necessary as sucking and breathing at the same time is difficult for these babies. A good eye and ear exam should be performed to check on the hearing and vision of the baby. The final result of the surgery or surgeries to create a nose will vary from one baby to the other. Orthodontic and/or speech therapy is necessary in many cases.

If there are no abnormalities of the brain, then intelligence is usually normal. When arhinia is part of another syndrome or disease, the baby may have more problems after birth. How well the baby will do depends on those other problems. When brain problems are significant, babies will have developmental delay and/or frequent seizures. When arhinia is part of Bosma arhinia microphtalmia syndrome, children require hormone therapy in order to go through puberty.

Will it happen again?

The risk that isolated arhinia will happen again is very low. If arhinia is part of a syndrome, then the risk that it will happen again depends on the type of syndrome and a consultation with a genetic specialist may be helpful in sorting this out.

What other questions should I ask?

- Does this look like a severe form of arhinia (complete or partial)?
- Are there other anomalies visible?
- Is an amniocentesis recommended?
- Is there too much amniotic fluid?
- How often will I have ultrasound examinations done?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet in advance the team of doctors that will be looking after my baby when it is born?

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