Nose Arhinia

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

What is Arhinia?

Arhinia is an extremely rare developmental disorder of the nose. There is a complete or partial (hemi arrhinia) absence of the nose at birth. The nasal area is flat and firm on palpation although small ridges, or rudimentary blind ending nostrils may be present.

In the vast majority of cases the upper jaw is small and the lacrimal duct system and sinuses are missing or mal developed. Arhinia is often associated with eye or ear anomalies or can be associated with other facial, skull or brain abnormalities.

How does this syndrome happen?

Because of the limited number of cases it is difficult to confirm the exact cause of this rare disorder. Most cases occur sporadically and happen by change.

Arhinia can coexist with holoprosencephaly, a disorder in which the brain fails to separate into two hemispheres. In this condition the prognosis is usually very poor and genetic aberrations are common.

Arhinia can also be part of Bosma arhinia microphtalmia syndrome. In this condition, arrhinia is associated with abnormal small or absent eyes and hypogonadism (underdeveloped reproductive tissues and a reduced production of hormones that direct sexual development). This syndrome is caused by a gen mutation on chromosome 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, consultation with a genetic specialist is recommended. MRI may be helpful to confirm or better define facial, skull or brain anomalies.

It may be that invasive testing (amniotic fluid puncture) will be offered to you to detect or exclude some genetic aberrations. Amniocentesis carries a small risk of miscarriage and some women choose not to have it performed out of fear for losing their baby. Not all syndromes can be diagnosed by genetic investigations such as an amniocentesis and not all anomalies are visible on ultrasound examination. However when there are no other anomalies visible on ultrasound and invasive testing gives normal results, then arhinia is very likely to be isolated.

What are the things to watch for during the pregnancy?

Sometimes babies with arrhinia accumulate extra amniotic fluid during the pregnancy, probably due to swallowing problems of the baby. This condition is called polyhydramnios. It can stretch the uterus too much and cause premature labour 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.

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

The prognosis of babies born with isolated arhinia is generally good. Some babies experience breathing or drinking problems immediately after birth because the nasal passage is absent. These babies may need oxygen or the placement of tubes to assist breathing. It may therefore be advisable to discuss the place of delivery with your caregiver.

Feeding via a tube is usually necessary as simultaneous sucking and breathing is difficult for these babies. Furthermore, vision and hearing assessments should be performed. The cosmetic result of nasal reconstruction will vary per person. Orthodontic and/or speech therapy is necessary in many cases.

When arrhinia is part of another syndrome, then the baby may have more problems after birth. The prognosis for children with holoprosencephaly is generally poor. When arrhinia is part of Bosma arhinia microphtalmia syndrome, then the children require hormone therapy by a pediatric endocrinologist (doctor specialised in hormonal disorders) in order to go through puberty. If there are no abnormalities of the brain, then intelligence is usually normal.

Will it happen again?

The risk that isolated arrhinia will happen again is very low. If arrhinia is part of a syndrome, then the risk that it will happen again depends on the type of underlying condition 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 arrhinia (complete or partial)?
- Are there other anomalies visible?
- Is invasive testing recommended?
- How much is the amount of 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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