Micrognathia

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

What is Micrognathia?

Micrognathia (sometimes called mandibular hypoplasia) is a development disorder in which the lower jaw is undersized. Affected individuals have an abnormally small chin compared to the rest of their face. The typical characteristics of micrognathia are visible when looking at the profile of the face. In this profile view, the chin is retracted, which is referred to as retrognathia.

Sometimes micrognathia is mild and the only abnormality in the child. However, micrognathia can also be part of a general condition (such as a skeletal or muscular disease) or a syndrome. A cleft palate (hole in the roof of the mouth) commonly occurs in association with micrognathia. Sometimes the tongue is moved backwards, due to lack of space, and blocks the pharynx. In this situation, micrognathia interferes with your child's feeding and breathing after birth, which may be life-threatening. The combination of micrognathia, a recessed tongue with airway obstruction is often named Pierre Robin sequence.

How does micrognathia happen?

Micrognathia appears as a birth defect in many conditions. These conditions can be inherited (passed on through genes of the parents) or caused by a mutation of a gen that newly occurred in the baby. However, there are cases in which the cause of micrognathia remains unknown.

Should I have more tests done?

You should ask if a specialised ultrasound of the baby during the pregnancy can be performed to detect other anomalies. Sometimes MRI (magnetic resonance imaging) can be performed. MRI is a way to obtain different images of a fetus inside the womb, which is safe to perform during pregnancy.

Micrognathia may be part of a general disease or a syndrome. If other anomalies are visible, then the risk that micrognathia is part of general disease or syndrome is high. Especially in this situation, it is important to have a consultation with a genetic specialist. Probably invasive testing (e.g., amniocentesis) will be offered to you to detect or exclude some of these conditions. Amniocentesis involves an amniotic fluid puncture and carries a small risk of miscarriage. Therefore, some women choose not to have it for fear of losing their baby. It must be kept in mind that not all syndromes can be diagnosed by genetic investigations such as an amniocentesis and not all anomalies are visible on ultrasound examination.

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What are the things to watch for during pregnancy?

Sometimes babies with micrognathia will accumulate extra amniotic fluid during the pregnancy, 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 examinations at least every 4 weeks.

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

The outcome varies from child to child and depends especially on the presence of an underlying condition, but also how quickly it was diagnosed (preferably before birth) and on the medical care after birth. The outcome for a child with an isolated micrognathia and prenatal detection is generally good.

Some babies experience severe breathing problems immediately after birth. Therefore, delivery in a center specialised in airway emergencies is recommended in most cases. If micrognathia is part of a general disease or syndrome then the baby may have more problems after birth.

In case of a serious condition, the baby may even live for only a short time. Additional surgeries or orthodontic and speech therapy later in life may be necessary. Most children are monitored until they reach skeletal maturity.

Will it happen again?

The risk that isolated micrognathia will happen again is very low. If micrognathia is part of a general disease or syndrome, then the risk that it will happen again depends on the type of the underlying condition and a consultation with a genetic specialist may be helpful to help to sort this out.

What other questions should I ask?

- Does this look like a severe form of micrognathia?
- 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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