Treacher Collins Syndrome

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

What is Treacher Collins Syndrome (TCS)?

A fetus, baby with TCS has several structural differences in the face (eyes, cheekbones, palate, mouth and jaws) and of the ear. The severity of these anomalies varies greatly per case from mild to severe.

The baby usually appears on ultrasound to have a small chin and occasionally a cleft of the palate. The other typical anomalies may also be recognized. A three-D ultrasound may be useful in revealing the typical aspect of the face, but this is especially possible late in pregnancy, after 25-28 weeks. In cases where the mouth and throat are severely abnormal, the baby may experience severe breathing difficulties at birth and this requires delivery in a specialized center where these emergencies, if present, can be managed properly.

How does TCS happen?

A change in a gene, called “mutation” is responsible for TCS.

How are chromosomes relevant to TCS?

Individuals with TCS have a normal number of chromosomes. The cause of the syndrome is a minuscule defect (mutation) within a gene that cannot be revealed by the conventional tests assessing the number of chromosomes. A specific test is needed to identify the mutation causing TCS, but the diagnosis is not always possible.

Should I have more tests done?

TCS is usually suspected after ultrasound examination of the baby’s face has revealed the typical anomalies of this syndrome. The diagnosis usually happens late in pregnancy because the anomalies may be more difficult to detect at early scans, even at 20 weeks. Usually, the ultrasound pictures are discussed with a geneticist (doctor specialized in diagnosing anomalies produced by an abnormal gene or chromosome). The doctor may suggest doing an amniocentesis (take some fluid from around the baby) to check for the specific defect (gene mutation) within the chromosomes of your baby, if this specific diagnosis is locally available.

What are the things to watch for during the pregnancy?

Sometimes your belly grows rapidly towards the end of the pregnancy because there is accumulation of a lot of amniotic fluid. This is because the baby cannot swallow the fluid, due to throat abnormalities and the fluid accumulates in the womb.

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

It is advisable that your baby is born in a center with experience to manage breathing difficulties, if these occur. It is possible to correct surgically the facial anomalies of TCS. A number of operations will be necessary, depending on the anomalies of the baby. For instance, the jaw anomalies, the cleft in the palate and the anomaly of the eye-socket can be corrected when the baby is about 5-7 years old. Unfortunately, the results are never perfect, but the aspect of the face can improve a lot. It is also important that the hearing of your baby is checked at about one year of age to detect possible deafness. Recent research indicates that in the future it may become possible to cure the anomalies more effectively, but this has only been tested on animals, thus far.

Will it happen again?

There are two types of TCS: 1) familial one where one of the parents has the affected gene. In that case the risk of each baby to inherit the syndrome is 50%. The other type, the most common (60% of the TCS cases), is caused by a new mutation (a new mistake in the gene) which has occurred during a very early stage in this pregnancy. In that case the risk of another baby to be affected is extremely low.

What other questions should I ask?

- Does this look like a severe case of Treacher Collins syndrome?
- Is it possible for me to terminate the pregnancy?
- Can a consultation with a geneticist be arranged for me to better understand what Treacher Collins syndrome means? (Which anomalies I should expect, how my baby will look like and what is the chance of my future children to have this syndrome?)
- 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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