

Craniosynostosis

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

What is craniosynostosis?

Craniosynostosis happens when the spaces between the bones of the head (skull) close prematurely.

How does craniosynostosis happen?

It is not clear why craniosynostosis occurs. It is a rare condition and happens in about 3.1 to 7.2 per 10,000 babies. Multiple proposed mechanisms explain this disease, such as gene or chromosome diseases, characteristics of the parents, and exposure to certain medications or toxic substances during pregnancy, among others. However, the precise mechanisms that cause this disease, particularly those not derived from genetic or chromosomal disorders, are still poorly understood.

How are chromosomes and genes relevant to craniosynostosis?

Chromosomes are where most of our genetic information is kept, while a gene is a short section of DNA. Our genes instruct the cells to make molecules called proteins. Proteins are essential to perform various functions to be healthy. Each gene carries the instructions that determine our features, such as hair color, height, etc. At least 25 to 30% of the babies with craniosynostosis are because of genetic disorders.

Should I have more tests done?

Many women will choose to have more tests done to know more about the baby's condition. The tests available will depend on where you live. Tests to ask about include:

- An **amniocentesis** to look for problems with the number of chromosomes and genes by removing a small amount of amniotic fluid surrounding the fetus. This procedure is indicated when craniosynostosis due to genetic causes is suspected.
- An **MRI scan** or **other imaging studies** can sometimes be done to provide additional information about the condition of the brain of the baby and additional information about the presence of other anomalies.

What are the things to watch for during the pregnancy?

Babies with craniosynostosis are at higher risk of problems during pregnancy, especially during delivery. The shape of the head can render the birth process difficult; likewise, these babies can have breathing complications and are more likely to require admission to the neonatal intensive care unit.

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

The brain developing inside the skull can be affected by the compression, which can limit brain growth and increase the pressure on the brain. Hence, there is a high risk of developmental problems, ophthalmic (eye) abnormalities, and respiratory complications.

Babies who also have had the information within their chromosomes or genes altered may have even more problems during pregnancy and after being born. This will depend on the type of genetic disorder.

At 6-12 months of age, a neurosurgeon could recommend performing one or more surgeries to attempt to fix the skull. The baby often requires complex care in a specialised hospital after the surgery to monitor for bleeding, infection, and swelling of the brain. It is possible that more than one surgical intervention is required when there are multiple craniosynostoses or in those attributable to genetic syndromes.

When they grow up, babies with craniosynostosis could have neurodevelopmental problems, vision loss, and other sensorial alterations. During the follow-up, psychological support is essential not only for children suffering from craniosynostosis but also for their families.

Will it happen again?

If no genetic reason is found to explain the craniosynostosis, the risk of this happening again depends on the suture involved, which is from ~2 to 10%. If there is a genetic reason, this will determine the risk, and a consultation with a geneticist is recommended.

What other questions should I ask?

- What sutures of the skull are involved in my baby's craniosynostosis?
- Is it one or more craniosynostosis?
- Does my baby have other alterations?
- How often will I have ultrasound examinations done?
- Will my baby be delivered vaginally or by cesarean delivery?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors assisting my baby and me during pregnancy and when it is born in advance?

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