

Iniencephaly

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

What is Iniencephaly?

This defect forms part of a category of neurological disorders related to spinal cord malformations called neural tube defects. Iniencephaly affects the bone at the base of the skull and the cervical (neck) and thoracic (chest) regions of the spine resulting in a severe backward bending of the head, absence of the neck, and severe distortion of the spine.

How does Iniencephaly happen?

The exact cause of iniencephaly is unclear, but it has been associated with genetic and environmental factors. Iniencephaly is thought to occur very early in pregnancy, often before a woman knows she is pregnant as a result of a failed development of the bone at the base of the skull and the cervical and thoracic regions of the spine.

Almost 80% of babies with Iniencephaly will have other anomalies. The most frequent associated malformations affect the brain, heart, digestive and urinary systems. Many women will also accumulate extra amniotic fluid around the baby due to abnormal fetal swallowing; this condition is called polyhydramnios.

How is Iniencephaly diagnosed?

Iniencephaly is usually diagnosed before the baby is born through a detailed ultrasound. Although usually there is no need to have more tests done, when there are other anomalies, it could be advised to have more tests done to know more about the baby's condition. The additional tests offered may include:

- An **MRI scan** can sometimes be done to provide more information on the condition of the baby. This scan uses strong magnetic fields and radio waves to create detailed images of the inside of the body.
- An **amniocentesis** to look for problems with the number of chromosomes and some of the problems within the chromosomes. This is done by removing a small amount of amniotic fluid surrounding the fetus.

What does it mean for my baby to be diagnosed with Iniencephaly?

Unfortunately, babies with Iniencephaly usually die in the womb or die shortly after birth, and there are only rare examples of survival in infrequent mild cases.

What are the things to watch for during the pregnancy?

If you are pregnant with a baby with Iniencephaly, you will not experience any related symptoms; however, if your pregnancy is complicated by polyhydramnios (too much amniotic fluid), you could experience breathlessness, heartburn, constipation, and swollen ankles and feet.

Due to the poor prognosis, termination of pregnancy is common in countries where it is legal. Where termination of pregnancy is prohibited or when parents choose to continue with the pregnancy, most specialists will recommend an early induction of labor to avoid obstructed labor and maternal complications due to the distortion of the baby's body.

Will it happen again?

The risk of Iniencephaly happening again is less than 1 in 100; however, if there is a family history of other neural tube defects, this risk may be higher. Notably, the risk of neural tube defects such as Iniencephaly can be reduced with folic acid supplementation (e.g., 4 mg/day).

What other questions should I ask?

- Are there other associated anomalies?
- How often will I have ultrasound examinations done?
- Where should I deliver?
- Can I meet the team of doctors that will be assisting me and my baby when it is born in advance?

Last updated August 2022