

# Eye Anomalies

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

## **What kind of structural differences in the position or size of the baby's eyes can develop?**

During early fetal life the eye sockets and the eyeballs develop. Normally the distance between the eyes is equivalent to the size of a single eye. If something goes wrong with development of the face of the baby the position and/or the size of the eyes may change (eye anomaly). The eyes can be too close to each other (hypotelorism) or too far apart (hypertelorism).

It can also occur that the eyes are fused together (synophthalmia or cyclopia). In some cases, one or both eyes are too small, and this can mean that there is a severe visual impairment in one or both eyes (microphthalmia/anophthalmia).

## **How do these eye anomalies happen?**

Usually, these anomalies are part of a chromosomal or genetic problem interfering with the normal development of the eyes/face.

## **How are chromosomes relevant to these anomalies?**

Eye position and size anomalies are found in association with some chromosomal anomalies such as an extra chromosome 13 (trisomy 13) or other rare genetic anomalies.

## **Should I have more tests done?**

When an eye anomaly is suspected at ultrasound investigation, the baby will be thoroughly checked for the presence of other anomalies as usually eyes anomalies are associated with brain anomalies or other organ's anomalies. The doctor may suggest doing an amniocentesis (take some fluid from around the baby) to check the chromosomes and its genetic material. Depending on the diagnosis, the doctor may bring you in contact with other specialists, such as a geneticist or an eye doctor or other specialists.

## What are the things to watch for during the pregnancy?

The pregnancy usually progresses normally, although the baby may not grow well, depending on whether a genetic anomaly is found or not.

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

This will depend on the exact diagnosis. If genetic analysis is normal and the baby has one or both eyes that are too small, this may mean that the baby may not be able to see or have a severely impaired vision. Plastic surgery can correct the esthetic problem, but there is no real cure for blindness.

## Will it happen again?

This will again depend on the exact diagnosis. If there is a familiar problem the geneticist will inform you on the chance that the problem comes back in a future pregnancy. If the anomaly was associated with a chromosomal anomaly your chance in a following pregnancy will be slightly increased, but an early test can tell you if this is the case. The eyes can also be seen by ultrasound at as early as 12-13 weeks of pregnancy. This means that you can be reassured or get a diagnosis of recurrence already very early in pregnancy.

## What other questions should I ask?

- Does this look like a severe case of eye anomaly?
- Is it possible for me to terminate the pregnancy?
- Where should I deliver?
- Where will the baby receive the best care after it is born?

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