Coloboma

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

What is a coloboma?

It is a rare malformation of the eye. It happens when part of the tissue that makes up the eye is missing. It is an important cause of visual impairment in children.

How does coloboma happen?

Coloboma is due to an absence of closure of the early tissues of the eye particularly at the choroid fissure. In some cases, particularly when it affects both eyes, a genetic disorder must be considered.

When is a coloboma suspected?

A coloboma may be suspected when there is a family history of eye disease or eye syndrome, in cases where the fetal eyes may be found on ultrasound to be too small (microphthalmia) or when cysts are seen at the level of eyeballs at prenatal ultrasound. Sometimes colobomas can be detected after a routine examination of the fetal eyes during pregnancy.

How is a suspected diagnosis of coloboma established?

It is necessary to perform a complete examination of the fetus by US and MRI to confirm that coloboma is isolated. Genetic tests are usually offered to complete the assessment.

What is the prognosis of coloboma?

If coloboma is involving one eye only (unilateral) with a normal examination of the other eye and no other fetal defects or genetic disorder, the child's vision in the involved eye can be impaired, the consequences may range from reduced vision in the involved eye to unilateral blindness.

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

It is important that the baby have a thorough ophthalmologic evaluation after birth.
If coloboma is involving both eyes (bilateral) it's likely that the baby will have severe visual impairment, including blindness.

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