Macrocephaly

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

This leaflet is to help you understand what macrocephaly is, how macrocephaly happens, what tests you need and the implication of having been diagnosed with macrocephaly for you, your baby, and your family.

What is macrocephaly?

The term macrocephaly means “large head.” Macrocephaly is diagnosed when the head circumference is above the 98th percentile, or greater than 2 standard deviations above the mean for gestational age. The most common variety is a benign condition running in families and without any clinical significance. However, in other cases, it can also be accompanied by many genetic disorders.

How does macrocephaly happen?

Macrocephaly may be caused by the enlargement of any of the head's compartments. As obvious causes of head enlargement (such as fluid on the brain), cerebral tumors, abnormal brain development etc. have been excluded, macrocephaly may be identified as megalencephaly.

The most common form of macrocephaly is benign familial macrocephaly. The large head is inherited and is similar to the head size of another family member. However, it could occur as part of a broad pattern of anomalies and dysmorphic features in many different syndromes, referred to as a syndromic macrocephaly.

How is a macrocephaly diagnosed?

The diagnosis of enlarged head can be made by ultrasound during pregnancy. However, the prenatal diagnosis of this condition is challenging because in the vast majority of the cases, it develops during the late stages of pregnancy, and it can be difficult to differentiate between the isolated benign and syndromic forms.

Should I have more tests done?

When the head measurements are larger than expected, there should be a careful search for intracranial abnormalities and other non-central nervous system abnormalities. Multidisciplinary counseling and investigation are important. Many women will choose to have more tests done to know more about the baby's condition.

MRI may add information regarding associated malformations and subtle structural differences in the brain that may be difficult to visualize by ultrasound. Amniocentesis for more detailed genetic testing should be considered when macrocephaly due to genetic causes is suspected.

What are the things to watch for during the pregnancy?

Pregnancy is monitored by ultrasound for evaluation of an enlarged fetal head and to rule out other structural differences. Babies with macrocephaly may be at higher risk of problems during delivery. The enlarged head can make the birth process difficult and Caesarean Section is more likely.

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

The baby's condition after birth depends on the underlying cause. Infants with familial benign macrocephaly usually require no treatment other than head size monitoring. If your child has macrocephaly due to an identifiable underlying cause, the outcome depends on the specific cause. Your doctor may prescribe specific and specialised treatments for your baby's symptoms, underlying disorders, or associated disabilities.

Will it happen again?

The chances of macrocephaly happening again in subsequent pregnancies depend on the underlying cause.

What other questions should I ask?

- How often will I have ultrasound examinations done?
- Can the treatment be started whilst my baby is still in the womb?
- Will my baby be delivered vaginally or by cesarean delivery?
- 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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