# Megalencephaly

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

## What is megalencephaly?

Macrocephaly is a term used to describe an enlarged head circumference. This may be due to multiple different conditions, including enlarged fluid-filled spaces within the brain, a thickened skull, or enlargement of the actual brain tissue. Megalencephaly is the term used to describe the latter, where the brain itself is the major contributor to an enlarged head. Most cases of megalencephaly are diagnosed after birth. Of those diagnosed before birth, late diagnosis in the third trimester is the most common. Megalencephaly can occur as an isolated condition, or it may be associated with an underlying genetic condition.

## How common is megalencephaly?

Macrocephaly, by definition, includes approximately 2% of all pregnancies. In these cases, most of the fetuses will be normal. The degree of macrocephaly is important, and the more enlarged the head circumference is, the greater the likelihood of a poor developmental outcome. The prevalence of megalencephaly, a subset of macrocephaly, at birth is unknown.

## How are chromosomes relevant to megalencephaly?

Chromosomes are the packaging of our DNA. Abnormalities in the chromosomes can result in a genetic syndrome. If an underlying cause of megalencephaly is found, then this would be referred to as syndromic megalencephaly. This is more likely the diagnosis when the head circumference is significantly increased, it appears earlier in pregnancy, (late second to early third trimester) and is associated with additional brain and non-brain anomalies.

## Should I have more tests done?

Many women will choose to have more tests done to know more about the condition and possible outcomes of their baby. Tests to ask about include:

- 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 and sending it to the lab for analysis.

- A fetal brain **MRI scan**, if available. This scan uses strong magnetic fields and radio waves to create detailed images of the inside of the body. It is safe for both mom and baby, and like ultrasound, does not involve any radiation. If can provide useful information regarding additional brain abnormalities existing with megalencephaly. That can help direct more detailed genetic investigations and assist in counseling regarding developmental outcomes.

- **Fetal echo**. This is a specialised ultrasound of the baby's heart during the pregnancy that can detect if there are heart abnormalities, which may co-exist with megalencephaly.

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

Babies with megalencephaly are not at increased risk during the pregnancy. However, if the head circumference is significantly enlarged, you will be at risk for a Cesarean delivery since the enlarged head may not engage in the pelvis or descend beyond the narrowest part of the pelvis during labor. Most specialists will recommend regular ultrasound examinations at least every 3-4 weeks. The ultrasound will help identify the rate of growth of the head circumference and the presence of other anomalies in or outside the brain.

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

Babies with megalencephaly with head circumference of less than +2.5 standard deviations above the mean, no other abnormalities, and normal genetic investigations may have a condition named benign familial megalencephaly. As the name implies, this is a fairly benign condition with normal neurodevelopmental outcomes. Generally, when there is an underlying genetic cause for the megalencephaly the head circumference will be greater than +2.5 standard deviations above the mean and the outcome will depend on the underlying genetic disorder.

General symptoms of megalencephaly include seizures, developmental delay, and motor dysfunction. As such, delivery should be at a tertiary center with an experienced neonatology service and neuroimaging service. The baby will need a head ultrasound after delivery, an MRI usually within a few months of age, and serial neurologic and pediatric developmental check-ups.

## Will it happen again?

If no other genetic reason is found to explain the megalencephaly, the risk of this happening again is extremely low. If there is a genetic reason, genetic testing will be offered to you and the father of the baby and/or future pregnancies. The results of this testing will determine the recurrence risk. In future pregnancies a targeted assessment of the fetal brain is recommended.

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