Malformations of cortical development (MCD)

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

What are malformations of cortical development?

Malformations of cortical development (MCD) are rare brain disorders that result from abnormal development of the brain's outer layer of nerve cells (known as the cortex) in the womb. MCD results for genetic, infectious or vascular causes. There are many types of MCD, which are classified according to the developmental step which is disrupted. For example, lissencephaly, or 'smooth brain', a disorder in which the brain does not properly form the gyri ("hills") and sulci ("clefts"), is a result of abnormal migration of nerve cells.

What does MCD cause?

The neonatal implications of MCD are broad and include epilepsy that does not respond to medication, cerebral palsy, feeding difficulties, intellectual disability, and other neurologic and behavioral abnormities.

How does MCD happen?

MCDs are a result of multifactorial causes involving different genes and environmental factors.

How are chromosomes relevant to MCD?

Chromosomes are the packaging of our DNA. Different chromosomes have different genes on them. In the past decade, a large and increasing number of genes have been identified that cause MCD, and their detection usually requires in-depth genetic testing (called whole exome sequencing).

Should I have more tests done if my baby is diagnosed with MCD?

Many women will choose to have more tests done to know more about the condition of the baby. The tests available depend on where you are. Tests to ask about include:

- An amniocentesis to look for problems within the chromosomes, such as single gene mutations. This is done by removing a small amount of amniotic fluid surrounding the fetus and analysing the fetal cells in the fluid.
- An MRI scan is very valuable in assessing the development of the cortex. Whenever an ultrasound raises a suspicion of an MCD, an MRI, if available, should be done to complement the findings of the ultrasound.
- A consultation with a fetal medicine specialist, a pediatric neurologist, and a geneticist is important to gain a better understanding of the potential implications this finding may have on the baby's development.

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

After delivery, the baby must be assessed by a pediatrician and a pediatric neurologist. A head MRI will be recommended to confirm the prenatally suspected findings. Since seizures are common with MCDs, anti-epileptic medication will likely be required. Long-term developmental follow-up will be needed as well to optimise the baby's development and address any developmental challenges (such as motor, language, and behavioral).

Will it happen again?

If the cause of the MCD is genetic, there is a chance for recurrence. That risk can be quantified by doing genetic tests for the mother and father of the baby, as well as brain MRIs for them if deemed necessary.

What other questions should I ask?

- Are other malformations present?
- What genetic testing is available?
- How often will I have ultrasound examinations done?
- Is surgery during pregnancy an option?
- Where is surgery after pregnancy available for the baby?
- What is the recommended method of delivery for my case?
- Where should I deliver?
- Where will the baby receive the best care after delivery?
- Can I meet in advance the team of doctors that will be looking after my baby following delivery?

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