



Screening Programmes

Pregnant with a family history of medium-chain acyl-CoA dehydrogenase deficiency (MCADD)

This leaflet gives you some information about developing a birth plan if you have a family history of MCADD.

What is MCADD?

During long periods between eating, the body breaks down its own fat stores to produce energy. People with MCADD lack one of the enzymes needed to do this. They can break down the stored fat partly but not completely. There is a hold up at the 'medium-chain fat' step where the enzyme needed to complete the breakdown is not working properly. This causes a build up of medium-chain fats.

Sometimes we need to break down fats quickly, for example, when we have not eaten for some while or when we have an infection. People with MCADD can't do this. The medium chain fats can build up and make toxic substances that may lead to serious symptoms.

If this condition is not diagnosed early or is ignored and not treated by following simple advice from a specialist medical team, it could lead to serious illness and possibly death. Fortunately – once diagnosed – MCADD is usually quite straightforward to manage and children with this condition usually lead healthy normal lives.

Before your baby is born

You should inform the health professional (obstetrician or midwife) looking after you that you or your partner have a family history of MCADD. You should ask for a referral to a paediatrician or genetic counsellor for advice then make a birth plan taking account of the information received (make sure the birth plan is written in your notes).

You **may** be advised that your baby needs early screening for MCADD. A sample of blood from your baby's heel will be collected 24 - 48 hours after birth on a blood spot card marked 'Family history of MCADD'. You will also be given information about anything special that you will need to do after your baby is born and you will be able to ask questions.