NHS

Screening Programmes

The importance of taking a family history

A two-day old baby died of medium chain acyl-CoA dehydrogenase deficiency (MCADD).

An article in the Telegraph on 21st October 2010 told the tragic story of a two-day old baby who died of medium chain acyl-CoA dehydrogenase deficiency (MCADD). The parents warned staff there was a strong family history of MCADD but despite this the baby was not screened early for the condition or given any advice before being discharged from hospital on the day the baby was born. The hospital has admitted liability.

http://www.telegraph.co.uk/health/healthnews/8076850/Hospital-neglect-led-to-baby-death.html

What is MCADD?

During long periods between eating, the body breaks down its own fat stores to produce energy. People with this inherited condition 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 there is a need to break down fats quickly, for example, when there is an extended period of not eating or when there is 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.

Family history

If a mother or her partner has a family history of MCADD you should make a referral to a paediatrician or genetic counsellor for advice. This advice should form the basis of a birth plan (making sure the birth plan is written in the mother's notes).

Depending on the risk of the baby having MCADD, the parents may be advised that their baby needs early screening for MCADD. A sample of blood should be collected 24 - 48 hours after birth on a blood spot card marked 'Family history of MCADD'. The parents will also be given information about any special treatment required after the birth of their baby.

Management (*Prior to results)*

It is essential to ensure that the baby maintains a good milk intake. A term baby should be fed every 4 hours and a preterm baby every 3 hours. Exclusively breast fed babies are particularly at risk in the first 72 hours when the supply of breast milk is poor; top up feeds of expressed breast or formula milk may be necessary in the first 48-72 hours until a good milk supply is established. If oral feeds are not tolerated or if the baby is unwell in any way, urgent referral should be made to a paediatrician for review and consideration on nasogastric tube feeds or commencing intravenous glucose (If MCADD confirmed: follow the standard MCADD clinical and dietary management guidelines).

Key messages:

- Establish if there is a family history of MCADD
- Refer parents antenatally to a paediatrician or genetic counsellor for advice
- Make an informed birth plan with the parents
- Ensure colleagues are aware of the risk of MCADD and the care required after birth.