Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

GIG CYMRU NHS WALES
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Public Health Wales

Newborn Bloodspot Screening Programme

Your baby's screening result

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is suspected

Your baby's screening result

The result of your baby's 'heel prick' screening blood test suggests they might have medium-chain acyl-CoA dehydrogenase deficiency (MCADD). Your baby now needs further tests to check whether he or she has MCADD or not.

This leaflet gives you some information about MCADD and explains what happens next.

What is MCADD?

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is a rare but treatable inherited disorder.

Babies with MCADD inherit two faulty copies of the gene for MCADD, one from each parent.

Babies with MCADD have a problem breaking down fats quickly enough to produce energy.

MCADD only causes problems when fats need to be broken down quickly, for example when a baby has not eaten for a long period or when they are unwell. If this happens, MCADD can cause low blood sugar and a build-up of certain fats. This can make toxic substances that can lead to serious symptoms.

Babies with MCADD benefit significantly from early treatment and can live healthy and active lives.

Without early diagnosis and treatment, MCADD can lead to serious illness and possibly death.

Treatment

Babies with MCADD should feed regularly, eat a healthy diet and be treated like any other child. They do not need special medications. They do not need a special diet but should avoid long periods without feeding.

Children with MCADD will be seen regularly by a specialist metabolic team.

If your baby is unwell in any way it is important to follow medical advice. When they are unwell they may need to go into hospital for treatment. Take any information you have about MCADD with you.

What happens next?

You will be given an appointment to see a specialist metabolic team who will:

- discuss the screening test result with you
- arrange blood and urine tests for your baby, and
- explain how these tests can confirm if your baby has MCADD.

If MCADD is confirmed, your specialist metabolic team will:

- explain how to use an emergency feed during illness
- explain what to do if your baby is not feeding well
- let your GP know about your baby's tests and MCADD
- give you written information about MCADD for you to share with your family, GP and local hospital
- answer any questions you may have, and
- arrange a follow-up appointment to discuss the test results.

Until you see the specialist team, you can breastfeed your baby or bottle-feed them with normal infant formula. You should feed your baby every three to four hours, day and night, or more often if demanded. If you are concerned about poor feeding or if your baby is unwell, contact your specialist metabolic team.

Your questions answered

How will I know if my baby is ill and what should I do?

Babies who have MCADD can become ill within the first few days of life. If they become ill they may:

- feed poorly
- vomit or have diarrhoea, or
- become drowsy, irritable or not respond normally.

You should not ignore these symptoms. If left untreated, babies with MCADD can have fits and slip into a coma, which can be life-threatening. If you are worried that your baby is ill, contact a member of your specialist metabolic team. If you cannot contact your specialist metabolic team you should take your baby to your local accident and emergency department as soon as possible.

Take any information that you have been given about MCADD to the hospital with you.

More information and support

- Metabolic Support UK (The National Information Centre for Metabolic Diseases) provides information and support for people with MCADD and their families:
  www.metabolicsupportuk.org

- Newborn Bloodspot Screening Wales:
  phw.nhs.wales/newborn-bloodspot-screening

Contact details for your specialist metabolic team

Specialist centre	
Consultant	
Metabolic dietitian	
Clinical specialist nurse	
Ward (if this applies)	

Notes

You can write down any notes here from discussions with your health professionals.

Using your information

For us to contact you as part of the programme, we will need to handle some of your and your baby's personal information. If you need more information about this, you can:

- visit the website: phw.nhs.wales/use-of-site/privacy-notice
- email PHW.InformationGovernance@wales.nhs.uk, or
- phone 029 2010 4307.

We also keep personal details to make sure that the standard of our service is as high as possible. This includes checking your baby's records if your baby is found to have a condition after having a screening test which showed a 'not suspected' result.

We only ever publish information as statistics and we never publish personal details. We pass on your personal information to health professionals or organisations that need it, including your GP, health visitor and consultant paediatrician. These professionals must protect the personal information in the same way as we do.

Information courtesy of Public Health England

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