



Information for GPs Confirmation of telephone call and initial appointment

Dear Doctor **[insert name and address of registered GP]**

Re: **[insert full name and date of birth and address of child]**

[Name of child] has had a positive (abnormal) newborn screening test for medium-chain acyl-CoA dehydrogenase deficiency (MCADD). This is a rare inherited enzyme deficiency which impairs the metabolism of fatty acids to produce energy. A child with this condition may become lethargic, or may vomit or have seizures and is at risk of coma, hypoglycaemia and death during prolonged fasting, especially in the course of infectious illnesses.

The child's parents or carers have been contacted by the MCADD Designated Team. The positive test is a screening test, and it is essential that the family are counselled about the present findings and have a confirmed diagnosis. This will entail blood and urine tests. The child and carers are to attend their regional centre at **[Appointment location]** at **[appointment time]**. If they would like to discuss any matters prior to this appointment **[Name of clinician]** may be contacted on **[contact number]**.

We will arrange a review appointment within a week of the first assessment to discuss the results of the diagnostic tests. If the child is unwell at any time during this interval urgent assessment at the regional centre or the local hospital should be arranged. A copy of an Emergency Department letter for infants awaiting the results of diagnostic tests accompanies this letter.

A patient with MCADD requires no special treatment when well but must avoid prolonged fasting. Breastfeeding may be encouraged but it is important to ensure that the infant is feeding adequately. Formula feeds rich in medium chain triglycerides (MCT) should be avoided.

The family will be shown how to follow an Emergency Regimen (ER) during intercurrent illness. This aims to supply enough calories as glucose to prevent metabolic decompensation. Standard Oral Rehydration Therapy (ORT) solutions do not contain sufficient glucose to avoid decompensation, and if used require fortifying with glucose polymer.

If the ER is not tolerated, or the child's condition deteriorates, then the child must be referred to hospital for immediate assessment.

The long-term prognosis for MCADD is very good with appropriate management. Immunisations may be undertaken as normal. General medical care is unaltered. The condition is inherited in an autosomal recessive fashion, so present or future siblings have a 1 in 4 risk of also being affected. Once the diagnosis has been confirmed, screening of any present siblings will be offered.

We will contact you as soon as possible once the results of the diagnostic investigations are available. If you have any further questions, please do not hesitate to contact **[name and contact details]**.

Yours sincerely

[Delete as appropriate - Relevant Regional Metabolic Team]

Dr Peter H Robinson

Telephone 0141 201 0000, page 8500 or E-mail: peter.robinson3@nhs.net

or

Dr Alison Cozens

Telephone 0131 536 0441 or Email: alison.cozens@luht.scot.nhs.uk

EMERGENCY CONTACT NUMBERS

DAYTIME:

[Relevant Regional Metabolic Team]

DAYTIME:

Glasgow

Dr Peter Robinson, Consultant, Paediatric Inherited Metabolic Disease

Dr Bernd Schwahn, Consultant, Paediatric Inherited Metabolic Disease

07699 646865 (aircall for the on-call consultant)

or

0141 201 0000 (switchboard) and ask for page 8500 (Dr Robinson) or page 8341 (Dr Schwahn)

Edinburgh

Dr Alison Cozens, Consultant Paediatrician in Inherited Metabolic Disease

07895 713600

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EVENINGS and WEEKENDS and PUBLIC HOLIDAYS

There is a consultant in paediatric metabolic medicine on call at all times. Switchboards at RHSC Edinburgh and Glasgow will have the on-call rota and contact details