# Joint Trust Guideline for the Management of Newborns with a Family History of MCADD (Medium-Chain Acyl-CoA Dehydrogenase Deficiency)

**Document Control:**

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|---|---|---|---|
| **For Use In:** | Norfolk and Norwich University Hospitals (NNUH), James Paget University Hospitals (JPUH) | | |
| | Blakeney Ward, NICU, Delivery Suite, Antenatal clinic | | |
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| **Document Owner:** | Women's and Children's (NNUH) | | |
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| **Ratified By:** | Clinical Safety and Effectiveness Sub-board (NNUH)<br>Clinical Governance Committee (JPUH) | | |
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**Version History:**

| Version | Date | Author | Reason/Change |
|---|---|---|---|
| 2.1 | July 2019 | Consultant Neonatologist | Reviewed and no clinical changes were needed |
| 3 | May 2023 | Consultant Neonatologist | Transfer of document to new Trust Procedural Document template. |

**Previous Titles for this Document:**

| Previous Title/Amalgamated Titles | Date Revised |
|---|---|
| None | Not applicable |

Note which Trust, where applicable.

**Distribution Control**

Printed copies of this document should be considered out of date. The most up to date version is available from the Trust Intranet.

## Consultation

The following were consulted during the development of this document:

- Dr P Ambadkar Consultant Neonatologist JPUH
- Dr O Tayo Neonatal Lead JPUH
- The Neonatal Unit Guidelines Review Forum

## Monitoring and Review of Procedural Document

The document owner is responsible for monitoring and reviewing the effectiveness of this Procedural Document. This review is continuous however as a minimum will be achieved at the point this procedural document requires a review e.g. changes in legislation, findings from incidents or document expiry.

## Relationship of this document to other procedural documents

This document is a clinical guideline applicable to Acute Collaborative: NNUH and JPUH; please refer to local Trust's procedural documents for further guidance.

## Guidance Note

This guideline has been approved by the Trust's Clinical Guidelines Assessment Panel as an aid to the diagnosis and management of relevant patients and clinical circumstances. Not every patient or situation fits neatly into a standard guideline scenario and the guideline must be interpreted and applied in practice in the light of prevailing clinical circumstances, the diagnostic and treatment options available and the professional judgement, knowledge and expertise of relevant clinicians. It is advised that the rationale for any departure from relevant guidance should be documented in the patient's case notes.

The Trust's guidelines are made publicly available as part of the collective endeavour to continuously improve the quality of healthcare through sharing medical experience and knowledge. The Trust accepts no responsibility for any misunderstanding or misapplication of this document**.**

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## Joint Trust Guideline for the Management of Newborns with a Family History of MCADD (Medium-chain acyl-CoA dehydrogenase deficiency)

## Quick reference guideline

**Family History of MCADD (i.e. affected first degree relative)**

- When the mother is admitted in labour inform the neonatal intensive care unit (NICU)
- When the baby is born inform NICU

Admit baby to Transitional Care or NICU – **no** admissions to postnatal ward
Ensure baby has a good milk intake and offer a milk feed ***as soon as possible after birth***

Then:

Term baby: feed every 4 hours
Preterm baby: feed every 3 hours

**Breast-fed babies**

Top up feeds with standard infant formula for 1st 72 hours of life
Discontinue top-up feeds *only* when good breast milk transfer is well established

Do not rely on blood glucose testing to guide adequacy of feeding: Hypoglycaemia only occurs at a relatively late stage so it is not safe to base the management on monitoring of blood glucose.

**Inadequate feed volume**

If the baby is not taking adequate volumes orally, transfer to NICU and feed by nasogastric tube.

**Does not tolerate oral feeding**

If enteral feeds are not tolerated, start an intravenous infusion of 10% glucose at 100 ml/kg/day.

Change to 10% glucose/0.18% NaCl on day 2
If there is no oral intake, the volume should be increased over 3 days to 150 ml/kg/d.

Monitor blood glucose and plasma electrolytes but base treatment on the clinical state (since hypoglycaemia occurs at a late stage).

**Unwell Baby**

If the baby seems drowsy or unwell in any other way, transfer to the neonatal unit urgently and give an intravenous bolus of 2 ml/kg 10% glucose followed by an infusion of glucose 10% at 100 ml/kg/day.

Change to 10% glucose/0.18% NaCl on day 2
If there is no oral intake, the volume should be increased over 3 days to 150 ml/kg/d. Monitor blood glucose and plasma electrolytes but base treatment on the clinical state (since hypoglycaemia occurs at a late stage)

***Hypoxic ischaemic encephalopathy***: Do not delay starting 10% dextrose IV – babies with MCADD are much less tolerant of energy stress

**Diagnosis**: when the baby is between 24 and 48 hours old

- Collect blood spot samples on the 'Guthrie card' – newborn bloodspot testing
- Mark the card 'family history of MCADD' and send to the newborn screening laboratory
- Discuss urgent testing with the screening laboratory – telephone 01223 217160

**Joint Trust Guideline for the Management of Newborns with a Family History of MCADD (Medium-chain acyl-CoA dehydrogenase deficiency)**

# 1. Introduction

## 1.1. Rationale

5 cases of newborn babies who died from MCADD between 2-4 days of life within an 11 year period have recently been highlighted in a National Patient Safety Agency (NPSA) rapid response report [1]. The diagnosis was made after death in all cases. The clinical pictures prior to death showed minimal prodromal signs and the infants generally succumbed rapidly and without warning.

Such deaths can almost always be avoided.

## 1.2. Objectives

Babies with MCADD are at risk of encephalopathy and death in the first few days of life. Recognition of the risk, in a baby with a family history of MCADD, followed by simple measures to ensure an adequate supply of milk or alternative sources of glucose, can prevent morbidity and mortality.

## 1.3. Scope

Medium chain acyl-CoA dehydrogenase deficiency (MCADD) is a rare inherited disorder where the body cannot metabolise fat properly. Individuals with undiagnosed MCADD commonly present with an episode of encephalopathy (drowsiness, seizures) usually accompanied by hypoglycaemia, that can result in coma or sudden death. Newborns with MCADD are especially vulnerable to sudden death in the first few days of life, as a regular feeding pattern may not have been established and the breast milk supply may be limited.

Since February 2009, all babies born in England, Scotland and Northern Ireland are offered screening for MCADD. This is done on blood spots as one of the battery of new born screening tests. Blood spots are collected at 5-8 days of age and abnormal metabolites can be detected in patients with MCADD. The diagnosis is confirmed by mutation analysis and / or urine organic acid analysis. Simple dietary management can then prevent adverse outcomes. Because the national screening programme is detecting cases of MCADD that were previously missed, there will be more families who are aware they could be carriers of MCADD. If both parents are MCADD carriers, there is a one-in-four chance of their child being born with MCADD.

Therefore babies born to families with a history of the disease need a special feeding regimen and observation from the moment of birth, rather than waiting until they have a positive test result. All such babies must be admitted to transitional care or to NICU, depending on clinical need. These babies should not be routinely admitted to Blakeney postnatal ward.

For the purposes of this guideline, a family history of MCADD is defined as a confirmed diagnosis of the condition in a first degree relative of the newborn (ie a sibling or parent). The autosomal recessive nature of the disease means babies with more distant family history of MCADD remain at low risk.

### 1.4. Glossary

The following terms and abbreviations have been used within this document:

| Term | Definition |
|---|---|
| NNUH | Norfolk and Norwich University Hospitals |
| JPUH | James Paget University Hospitals |
| MCADD | Medium-Chain Acyl-CoA Dehydrogenase Deficiency |
| NPSA | National Patient Safety Agency |
| BIMDG | British Inherited Metabolic Disease Group |
| EIA | Equality Impact Assessment |

## 2. Responsibilities

- Dr David Booth – Consultant neonataologist, NNUH: author and reviewer
- Dr Fauzia Akhtar – Specialty trainee, NNUH - reviewer

## 3. Processes to be followed

Key components of management of newborn babies with a family history of MCADD include:

### 3.1. Diagnosis

Consultant paediatrician responsible for metabolic diseases should be informed antenatally; urgent testing for MCADD should be discussed, and a clear plan of care for after delivery established.

### 3.2. Management

Do not wait for test results but ensure baby has a good milk intake, with term baby fed every 4 hours and preterm baby every 3 hours. Particular risks have been reported in breast-fed babies in the first 72 hours due to the quantity and composition of milk from the mother during this period. It is therefore recommended that these babies receive top-ups of formula milk until good milk supply from the mother is established.

### 3.3. Treatment

Babies not taking adequate volumes orally may be transferred to neonatal units and fed by nasogastric tube. If enteral feed is not tolerated, intravenous infusion is recommended.

### 3.4. Testing

Hypoglycaemia only occurs at a relatively late stage so it is not safe to base the management on monitoring of blood glucose. With a family history of MCADD, the 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’ and the screening laboratory informed.
- Send urine for organic acids

- Send standard day 5 blood spot screening and mark 'Family history of MCADD'

### 3.5. Parent information

An information booklet for parents or carers is available from the British Inherited Metabolic Disease Group (BIMDG) and is available via this link: MCADD-Parents-Info-sheets-April_2015_263953_12052015.pdf (bimdg.org.uk)

## 4. References

(1) National Patient Safety Agency 2011 – Rapid response report - Keeping newborn babies with a family history of MCADD safe in the first hours and days of life NPSA/2011/RRR002 (access here)

British Inherited Metabolic Disease Group 2008 – MCAD deficiency: management of newborn babies with a family history - (access here)

National Patient Safety Agency 2011 – Clinical briefing sheet: keeping newborn babies with a family history of MCADD safe (access here)

National Patient Safety Agency 2011 – Rapid response report – Supporting information NPSA/2011/RRR002 – (access here)

British Inherited Metabolic Disease Group: http://www.bimdg.org.uk/guidelines.asp
Newborn bloodspot screening: http://newbornbloodspot.screening.nhs.uk/mcadd

## 5. Monitoring Compliance

All pregnant women with a family history of MCADD should have a neonatal alert raised, to inform neonatal services of the problem and allow timely referral to a metabolic paediatrician.

All babies with a family history of MCADD should have a feeding plan prior to delivery, as part of the neonatal alert process, and should be fed as soon as possible after delivery and no later than 4 hours for term babies and 3 hours for preterm babies.

Babies at risk of MCADD who cannot tolerate oral feeding should be managed with appropriate IV glucose as outlined in the quick reference algorithm.

All babies at risk of MCADD should have early blood spot testing, between 24 and 48 hours of age.

Compliance with the process will be monitored through the following:

| Key elements | Process for Monitoring | By Whom (Individual / group /committee) | Responsible Governance Committee /dept | Frequency of monitoring |
|---|---|---|---|---|
| MCADD is rare – an annual review of data from NICU looking for any births with a family history of MCADD | Annual case review – have any babies been born in the past year with a family history of MCADD?<br>If any cases – audit against the compliance monitoring standards above | David Booth – NNUH Neonatal paediatrician - JPUH | NICU governance at NNUH Paediatric governance at JPUH | Annual |
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The audit results are to be discussed at the NICU departmental audit and guidelines monthly meeting to review the results and recommendations for further action. Then sent to Women's and Children's governance and directorate who will ensure that the actions and recommendations are suitable and sufficient.

## 6. Appendices

There are no appendices for this document.

## 7. Equality Impact Assessment (EIA)

| **Type of function or policy** | Existing |
|---|---|

| **Division** | Women's and Children's | **Department** | NICU |
|---|---|---|---|
| **Name of person completing form** | David Booth | **Date** | 22.05.23 |

| **Equality Area** | **Potential Negative Impact** | **Impact Positive Impact** | **Which groups are affected** | **Full Impact Assessment Required YES/NO** |
|---|---|---|---|---|
| Race | | | | No |
| Pregnancy & Maternity | | | | No |
| Disability | | | | No |
| Religion and beliefs | | | | No |
| Sex | | | | No |
| Gender reassignment | | | | No |
| Sexual Orientation | | | | No |
| Age | | | | No |
| Marriage & Civil Partnership | | | | No |
| **EDS2 – How does this change impact the Equality and Diversity Strategic plan (contact HR or see EDS2 plan)?** | No effect | | | |

- **A full assessment will only be required if: The impact is potentially discriminatory under the general equality duty**
- **Any groups of patients/staff/visitors or communities could be potentially disadvantaged by the policy or function/service**
- **The policy or function/service is assessed to be of high significance**

**IF IN DOUBT A FULL IMPACT ASSESSMENT FORM IS REQUIRED**

**The review of the existing policy re-affirms the rights of all groups and clarifies the individual, managerial and organisational responsibilities in line with statutory and best practice guidance.**