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University Health Board

Newborn Bloodspot Screening for MCADD

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Flowchart

The offer of urgent Newborn Bloodspot Screening for MCADD (Medium-chain acyl-CoA dehydrogenase deficiency)

Antenatal Booking Visit
All pregnant women to be asked about any inherited conditions that may prompt further action

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Maternal History of MCADD	**Family history of MCADD**
Arrange urgent antenatal appointment in Antenatal / Medical Clinic	**(including maternal carrier status)**

Community midwife to record MCADD history in:

- Hand Held Maternity Record in "Special Considerations" and "Postnatal Care Plan"

A hospital birth is advised. If a women requests a home birth, an appropriate care plan must be in place.

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Offer Urgent Newborn Bloodspot Screening

- Provide mother with verbal and All Wales Information for parents leaflet to obtain informed consent
- When consent given - rapid testing to be taken 24 to 48 hours after birth using bloodspot card (using the four circles). Bloodspot card to be marked **"Family history of MCADD". Inform paediatrician.**
- Transport bloodspot card to the Biochemistry Department who will arrange urgent transport to Cardiff Newborn Screening Lab (ensure bloodspot card is air-dried before inserted into envelope).
  To request urgent transport to the Cardiff newborn screening laboratory. telephone:
  01792 703036 (Singleton, NPT, POW samples) or 01792 703052 (Swansea /NPT), 01656 752333 (POW)
  Refer to Cardiff & Vale *MCADD Family History and Diagnostic Samples Protocol* (appendix 1)
- Inform Cardiff Newborn Screening Laboratory of urgent sample:
  - **02920 744032 or 02920 743561**

Newborn bloodspot screening to be repeated when baby is 5 - 8 days old (counting day 0 as day of birth) for other conditions.

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Infant Feeding

British Inherited Metabolic Diseases Group (BMIDG) 2008)
Ensure infant has a good milk intake. A term baby should be fed every four hours from birth and a preterm infant every three hours. There is a risk of problems in the first 72 hours in breast fed infants, since the supply of breast milk may be insufficient. These infants may require supplement feeds for the first few days until a good milk supply is established:
Further guidance:
BMIDG: http://www.bimdg.org.uk/store/protocols/docs/Mcad-babyv1-2-743946-24-06-2008.pdf

ABM University Health Board (2011): Guideline for the reluctant breastfeeding babies
http://howis.wales.nhs.uk/sites3/Documents/790/Reluctant%20Breastfeeding%20Babies_ABMU%20Guideline%202010.pdf

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Concerns re infant condition

- Urgent referral to paediatrician

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Informing parents of MCADD Result

- If MCADD not identified – inform parents of result. Bloodspot sample to be repeated when baby 5 - 8 days old to test for other conditions (counting day of birth as day 0)
- If MCADD suspected, Cardiff newborn screening lab will contact local paediatrician to arrange urgent referral
- MCADD carrier status will not normally be detected. If MCADD carrier status is detected, offer parents referral to genetic counselling services.

National Patient Safety Agency October 2011

"Keeping newborn babies with a family history of MCADD safe in the first hours and days of life"

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 etc) usually accompanied by hypoglycaemia, that can result in coma or sudden death. Fortunately, such catastrophes can be prevented by simple treatment (primarily a regular intake of glucose during illnesses), allowing affected individuals to live a normal healthy life.

The disease affects about one in 10,000 babies born in the UK. Newborn bloodspot screening throughout England identifies around 60 cases each year. Around one in 80 healthy people is a carrier of MCADD, but will not have any symptoms. However, if both parents are MCADD carriers, there is a one-in-four chance of their child being born with MCADD. Routine newborn bloodspot screening for MCADD will commence in Wales in June 2012.

Newborn bloodspot screening will detect cases of MCADD that were previously missed. This will result in more families being aware that they could be carriers of MCADD.

Patterson and Henderson (2010) reported 5 cases of newborn babies who died from MCADD between 2 - 4 days of life within an 11 year period and highlighted the risks for babies with MCADD in the first 3 days of life especially if breast fed. The diagnoses were made after death in all cases by acyl-carnitine analysis. In one baby the cause of death had been attributed to an overwhelming infection. The clinical pictures prior to death showed minimal prodromal signs and the infants generally succumbed rapidly and without warning. During the study period, the Yorkshire region had prevalence for early MCADD death of 1:100,000. This estimate highlights that these deaths are exceptional and underscore the importance of vigilance in babies with a family history in whom such deaths can almost always be avoided.

Reference

Patterson AL. Henderson MJ. (2010) 5 Deaths. *Journal of Inherited Metabolic Disease,* 33:S59.

Further information fro health professionals available at:

Newborn Bloodspot Screening Wales (Public Health Wales)
http://www.newbornbloodspotscreening.wales.nhs.uk/sitesplus/documents/1008/MCADD%20Professionals%20Information.pdf

MCADD Family History and Diagnostic Samples Protocol

Background

Newborn bloodspot screening for Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD) was introduced in Wales on 1st June 2012. Routine newborn screening samples are collected by midwives between days 5-8 of life. These screening samples are sent via 1st class post directly to the Newborn Screening laboratory.

MCADD is a fatty acid oxidation disorder which can result in sudden infant death if the condition is undiagnosed. Therefore it is essential that diagnosis is not delayed. If there is a family history of MCADD (in first degree relatives) the baby is at higher risk of inheriting the condition. Any baby with a family history of MCADD should have a newborn screening card taken early at 24-48 hours of life (day 1-2). This sample will be tested for MCADD only; therefore a routine newborn screening card taken at day 5-8 of life is still required.

Sample Collection

It is the responsibility of the local paediatrician at the hospital in which the baby was born to organise a newborn bloodspot sample to be collected for early screening before the baby is discharged from hospital.

Requirements

Due to unreliability of the postal service, newborn screening cards from babies with a family history of MCADD should be sent to the Newborn Screening laboratory via the local Hospital laboratory where the baby was born.

Samples should arrive at the Newborn Screening Laboratory at UHW within 1 working day of collection.

- Samples can be analysed on the day of sample collection if the sample is received by 12pm lunchtime Monday - Friday.
- Samples are not analysed at the weekend, therefore samples collected on a Thursday or on a Friday morning should arrive in the laboratory by 12pm lunchtime on Friday so the sample can be analysed before the weekend.
- Samples collected at the weekend or those samples collected on a Friday that are unable to reach the laboratory by 12pm should be received at UHW by 12pm Monday.

The Newborn Screening laboratory ***MUST be informed*** that they are expecting a sample to ensure that the run is not processed until the sample has been received (Contact details are at the end of this information sheet).

Please send these samples to the Newborn Screening Laboratory in an ***envelope clearly marked as URGENT for Newborn Screening.***

Sending Samples by Taxi

If a sample is sent to the lab by taxi the driver should go to:
Pathology Reception (Medical Biochemistry and Immunlogy)
Upper Ground Floor, B Block, University Hospital of Wales

Diagnostic Samples for Any MCADD Screen Positive Babies

Babies who screen positive for MCADD will have to have diagnostic samples sent urgently to UHW, via the local laboratory. These samples are as follows:

- 3 dried bloodspots with non-anticoagulated blood (these samples will be for acylcarnitines, common mutation analysis and extended mutation analysis).
- 1 Urine sample – for organic acids and (if required) quantitative hexanoyl glycine*.
- Lithium heparin sample for acylcarnitines

*Hexanoyl glycine analysis is only required if hexanoyl glycine is not detected in the qualitative organic acid profile.

These samples should be received at UHW on the same day of sample collection and should be marked as urgent for Newborn Screening.

Family History of PKU

As untreated PKU can cause severe learning disability, which can worsen with diagnostic delay, the Newborn Screening Programme Centre has recommended that babies with a family history of PKU should also have a sample collected early at 24-48 hours of life. These should also be sent via the local laboratory to the Newborn Screening Laboratory within two working days of sample collection.

Laboratory Communication

Receipt of the sample and results of the test should be communicated to the requesting laboratory. This will be done by the Clinical Biochemist or Senior in the laboratory.

Contacts

If there are any queries regarding this protocol please contact:
Roanna George on: 02920 743561 or roanna.george@wales.nhs.uk.

To inform the laboratory of babies with a family history of MCADD:
Roanna George: 02920 743561 or Newborn Screening Office: 20920 744032

Directorate of Women & Child Health

Checklist for Clinical Guidelines being Submitted for Approval by Quality & Safety Group

Title of Guideline:	Flowchart for babies with a family history of MCADD
Name(s) of Author:	Wendy Sunderland-Evans
Chair of Group or Committee supporting submission:	Antenatal Forum
Issue / Version No:	1
Next Review / Guideline Expiry:	August 2016
Details of persons included in consultation process:	D. Apsee Lead for Midwife for CDS S. Banergee, Consultant Neonatologist (Singleton) M. Bonduelle Clinical Director A. Cookson, Consultant Chemical Pathologist K. Creese, Consultant Neonatologist (POW) M. Dey Consultant Obstetrician (POW) J. F. Doran, Consultant Chemical Pathologist C. Dowling Head of Midwifery H. Durrant, Consultant Chemical Pathologist A. Gunneberg, Consultant Chemical Pathologist S. Hancock, Clinical Scientist (Pathology Department) S. Hemmadi Consultant Obstetrician T. Hildebrandt, Consultant Paediatricain (POW). J. Hillborne Consultant Obstetrician P. Hopkins, Process Service Manager (POW Pathology) H. James, Lead Nurse (Neonatology) E. Kevelighan Consultant Obstetrician V. Langford Sister ANC Singleton R. Llewelyn Consultant Obstetrician F. Majoko Consultant Obstetrician J. Matthes, Consultant Neonatologist (Singleton) I. Millington (GP - LMC / member of HB A/ N Forum G. Morris Consultant Neonatologist (Singleton) M. Morgan Consultant Obstetrician Sara Morgan Lead Midwifery Singleton P. Morris Consultant Obstetrician M. Moselhi Consultant Obstetrician R. Nagrani Consultant Obstetrician Neath Port Talbot Community Midwifery Teams L. Owen Senior Midwife Community Singleton S. Passey, Head of Nursing (Neonates and Paediatrics) K. Phillips, Practice Dev. Nurse (Neonatology) H. Rees Thomas NPT ANC Sister

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Brief outline giving reasons for document being submitted for ratification	Flowchart which describes the process to follow in relation to newborn bloodspot screening when there is a family history of MCADD
Name of Pharmacist (mandatory if drugs involved):	N/A
Please list any policies/guidelines this document will supercede:	None
Keywords linked to document:	MCADD / Newborn bloodspot screening
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* To be completed by Author and submitted with document for ratification to Clinical Governance Facilitator