



Northern Ireland Newborn Bloodspot Screening Programme

**Revised Screening Programme
Introduction**

3rd AUGUST 2009



This 20 minute e-learning session has been prepared for the following groups of staff :

- antenatal clinic midwifery;**
- community midwifery;**
- inpatient midwifery;**
- neonatal nursing;**
- inpatient paediatric nursing;**
- health visiting; and,**
- community paediatric nursing.**



Trust Records of Staff Access

ALL staff accessing this e-learning session MUST ensure that they:

- **Sign the training list located at the PC they have accessed, or**
- **Inform their line manager (using the certificate provided at the end of this presentation, if requested), or**
- **Inform the Trust Lead for MCADD/Revised CF Implementation (see final slides for contact details)**

IT IS IMPORTANT THAT EMPLOYING TRUSTS MAINTAIN A RECORD OF ALL STAFF TRAINED.



This session should be complemented by:

- **the N Ireland MCADD and Revised CF Implementation Professional Handout (issued April 2009); and,**
- **your Trust's written Repeat/2nd Test Request Protocol (Appendix 3).**

N.B. your Trust's existing professional guidance on newborn bloodspot screening continues to apply unless where otherwise stated in the Professional Handout document.



Aims of the session (1)

By the end of this session you will know about:

- **NSC recommendations and the strategic context of newborn bloodspot screening;**
- **The introduction of the revised bloodspot screening programme across N Ireland and transition arrangements;**
- **The new bloodspot consent policy and the new parental information leaflet;**
- **New reporting codes being introduced by the laboratory.**



Aims of session (2)

You will have a basic understanding of:

- the MCADD condition, its incidence, the benefits of screening, how suspected MCADD cases are referred and clinically managed;**
- the extended CF screening programme, its benefits, CF incidence, the expected incidence of CF gene carriers and clinical referral of infants who need further investigation.**



Aims of the session (3)

You should also know about important factors relating to:

- **obtaining good quality bloodspot samples and reducing avoidable repeat tests;**
- **sharing results with parents; and,**
- **follow-up of infants with outstanding results.**

In addition you will know where to access further information relating to all of the topics covered in the session.



Revised Newborn Bloodspot Screening Introduction

The newborn bloodspot test, offered to all Day 5 infants in Northern Ireland will be revised from

3rd August 2009 to include:

- phenylketonuria (*PKU*);
- congenital hypothyroidism (*CHT*);
- homocystinuria (*N Ireland only*);
- tyrosinaemia (*N Ireland only*);
- revised cystic fibrosis (*CF*); and,
- medium chain acyl coA dehydrogenase deficiency (*MCADD*).

Bloodspot - Strategic Context



- **Screening for PKU, CHT, MCADD and sickle cell disorders (SCD) should be offered as part of the screening programme for all newborn infants (National Screening Committee - NSC).**
- **NSC also determined that universal newborn screening for CF should be implemented.**
- **In August 2008, the Minister directed that the existing programme for CF in N Ireland should be extended to include genetic testing, in line with protocols in the rest of the UK.**
- **Priority for Action targets to have MCADD and Revised CF screening implemented in 2009 and SCD implemented in 2010 are in place in N Ireland.**



What is MCADD?

MCADD is a rare inherited enzyme deficiency, which reduces the body's ability to metabolise fat into energy.

Infants with this condition are at risk from hypoglycaemia, coma and death with fasting and particularly during intercurrent illnesses when the demand for energy increases and calorie intake is often reduced.

(refer to pages 15-20 of the Professional Handout for further information about MCADD)



Why screen for MCADD?

Screening for MCADD, which affects 1 in 10,000 infants born (*around two cases per year in N Ireland*), will mean that, once diagnosed, infants with this condition can usually, through straightforward dietary management, lead healthy normal lives.



Referring babies to MCADD clinical services

- **Where it is suspected that an infant has MCADD arrangements are made (via the GP) for the family to see the MCADD Specialist Team at RBHSC within 24 hours of the presumed positive result becoming available;**
- **The MCADD Specialist Team carries out diagnostic tests and provides the family with extensive advice on dietary management including what to do if the infant becomes unwell.**



How will CF screening be extended?

The existing CF screening programme will be extended to include genetic testing for CF, which will be carried out for a small group of infants with high IRT screening test results (*around 125 per year in Northern Ireland*).

Genetic testing, where necessary, is carried out on the first sample collected at Day 5.

(refer to pages 21-27 of Professional Handout for full information on extended CF screening)



Why revise CF screening? (1)

- **‘Two-step’ screening will mean that the majority of infants with CF (*between 12 and 15 cases per year in Northern Ireland*) will be detected through Day 5 blood spot screening.**

This will allow for earlier and appropriate clinical referral to the Paediatric CF Service at RBHSC and earlier interventions for babies with CF;



Why revise CF screening? (2)

- Two-step screening will also significantly reduce the number of repeat tests currently required, the majority of which are negative;
- The new protocol will also identify a few more cases of CF that would not have been detected by the existing biochemical IRT test alone.

(Around 250 repeat IRT equivocal tests are carried out in N Ireland every year, this will reduce to ~35, once two-step screening is introduced)



What about CF carriers identified?

It is estimated that between 15 and 30 healthy carriers of the CF gene will be identified in Northern Ireland every year as a result of the introduction of two-step screening.

Genetic counselling will be available through the Paediatric CF Service for the families of infants identified as healthy carriers.

Referring babies to CF clinical services



- **The CF specialist will contact the HV with:**
 - the appointment details for the parents (within 24hrs)
 - what to say to the parents
 - contact number for the parents/HV should further support be required
- **The HV will confirm contact with the parents and their attendance at the appointment back to CF services**
- **HV services will be guided and supported by the CF Specialist through this process**



Key points for Professionals

Offering Screening and Taking Samples



Who offers and takes the test?

- Most tests are offered and taken by CMW services.
- Neonatal and paediatric nursing services undertake screening for sick infants who remain in hospital at Day 5.
- HVs and community paediatric nursing services largely carry out tests for movement in babies and those infants requiring tests who are no longer within the neonatal period, i.e. those infants older than 28 days of age.



Transition Arrangements for screening

- The *revised* newborn bloodspot screening test should be offered on 3rd August 2009 and thereafter.
- This should include all repeat insufficient / unsuitable tests offered from this date.

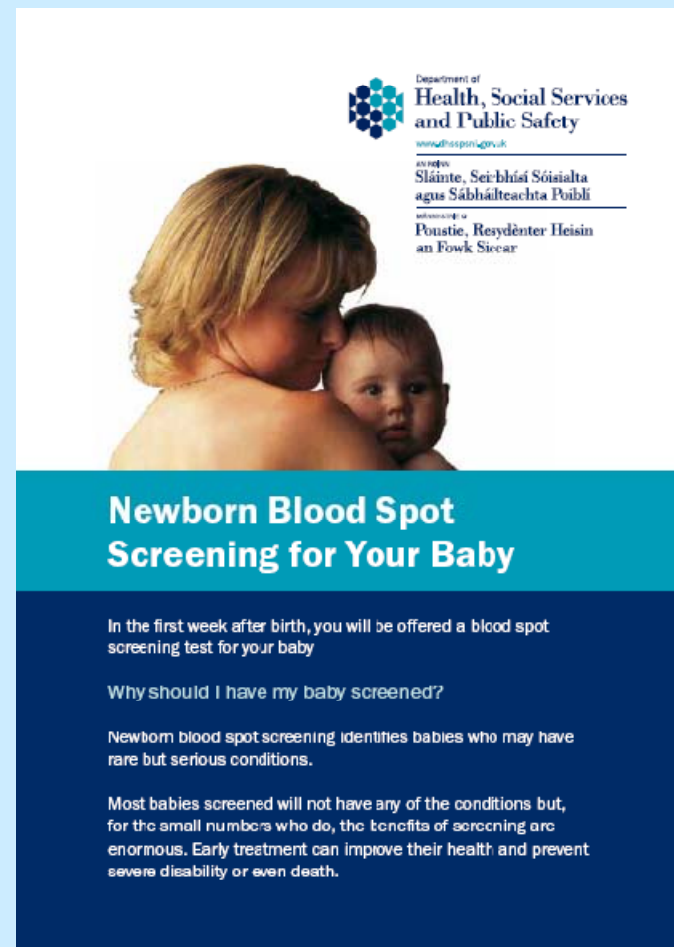
(refer to pages 3-4 of the Professional Handout for further information on transition arrangements)



Following the new bloodspot screening consent policy (1)

- Issue the *new* Northern Ireland parental information leaflet in the antenatal period (28-wk assessment);
- Ensure the parent has a copy of the leaflet and has read and understands the procedure at least 24 hours before taking the bloodspot sample;

(for leaflet see Appendix 1 of the Professional Handout)





Following the new bloodspot screening consent policy (2)

- **Do NOT issue the new leaflet before 3 August 2009;**
- **As per national guidance, parents should NOT be asked to provide written consent or decline;**
- **Parents may decline all or part of the bloodspot screening programme;**
- **For CF screening, consent is to the approved NI Protocol, which in 0.5% of cases will involved genetic testing. No alternative CF screening protocol can be offered;**



Following the new bloodspot screening consent policy (3)

- Record parental consent / decline in both the PCHR (page 18) and professional record;
- Notify the laboratory of ALL declined screening tests (*recording the detail on the top right hand corner of the bloodspot card*) and inform GPs about declined screens **AS SOON AS POSSIBLE;**

(refer to pages 5-8 of the Professional Handout for N Ireland Bloodspot Consent Policy)



Taking bloodspot samples (1)

“Repeat samples requested because of poor quality (avoidable repeats) cause delay in identification and treatment of screen positive babies, anxiety to parents, distress to babies and wastes healthcare resources.”



Taking bloodspot samples (2)

AVOIDABLE REPEATS ACCOUNTED FOR ALMOST 2% (400) OF ALL BLOOD SAMPLES TAKEN IN NORTHERN IRELAND DURING 2007/8!



Taking bloodspot samples (3)

AVOIDABLE REPEATS INCLUDE SAMPLES TAKEN:

- **Too young (*on or before Day 4*);**
- **Too soon after blood transfusion, i.e. less than 72 hrs after blood transfusion;**
- **Unsuitable specimens, e.g. where the:**
 - **specimen is contaminated with urine or faeces;**
 - **specimen has been over-layered;**
 - **bloodspot card that has gone past expiry;**
 - **bloodspot card does not contain full information; or,**
 - **transit time of the bloodspot card is in excess of 14 days.**
- **Insufficient specimens, i.e. 4 bloodspots have not soaked through to the back of the card.**



Taking bloodspot samples (4)

- All bloodspot samples should be taken on Day 5, irrespective of medical condition, prematurity, or feeding status (*count DOB as Day 0*);
- Always take Day 5 samples before planned blood transfusion or delay sample collection for 72 hours post transfusion. Where transfusion is ongoing take the sample at Day 8;
- Do NOT use bloodspot cards that have gone past expiry;

Taking bloodspot samples (5)

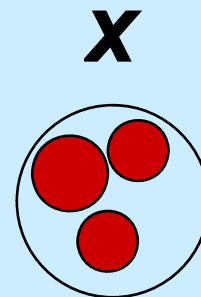
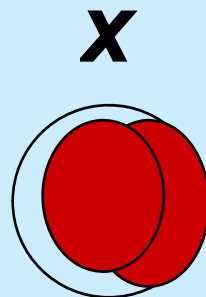
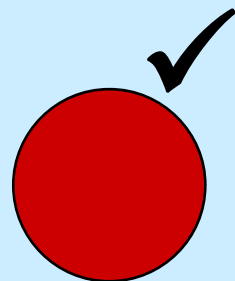


- **Take time to complete all areas of the bloodspot card. Remember where you omit to record the date that the bloodspot sample is taken the laboratory will request a repeat test;**
- **Ensure that the infant's heel is thoroughly cleaned, either by washing with plain water or disinfecting with an alcohol swab, and allow to dry before taking the sample;**



Taking bloodspot samples (6)

- Wait for blood to flow;
- Allow blood to fill each circle and soak completely through;
- Do not add layers of small blood spots;
- Allow the card to air dry (*away from direct sunlight*) before inserting into glassine sleeve;



(refer to pages 9-10 of the Professional Handout for full guidance)



Taking bloodspot samples (7)

- Inform parent when and how they will find out about results (*HV feedback by 8-week assessment*);
- Record details of consent and date that sample was taken and despatched in PCHR and professional record;
- Always use the yellow prepaid addressed bloodspot envelopes;
- Despatch cards on the day the sample is taken. **DO NOT** batch post bloodspot cards (*only 1 card per envelope*);
- **DO NOT** use internal mail to access either Royal Mail or Trust transport systems, such as blood runs.



Repeat Bloodspot Requests (1)

- Carry out repeat tests within 72 hours of request, or as requested by lab, e.g. all Repeat - CF Inconclusive (equivocal) tests should be taken at Day 21 and NOT Day 28;
- Inform parents why the repeat test is necessary and how they will find out about results (*from HV in approximately 2 weeks*);



Repeat Bloodspot Requests (2)

- Carry out a Preterm CHT test where the first sample was taken at less than 35 weeks and 7 days equivalent gestational age;
- Follow your local Repeat and 2nd Test Protocol to effect fast-track sampling (*see Appendix 3*).

(refer to page 12 of the Professional Handout for full guidance)



Key points for Professionals

Sharing Results with Parents and Following Up Outstanding Results



Who provides results to parents?

- HV services are responsible for sharing 'normal' results as well as ensuring that all results are inserted into the PCHR;
- Professionals with responsibility for taking repeat tests (*as defined in Appendix 3*) are responsible for sharing initial test results where a repeat is necessary;
- GPs are responsible for sharing 'abnormal' results as part of the clinical referral protocols established for each condition (*excluding CF referrals*).



New Screening Status Codes

These are the new status/reporting codes which will be used by the laboratory:

- 01 – Specimen received in laboratory
- 02 – Declined
- 03 – Repeat (see Reasons for Repeat Test)
- 04 – Not suspected ('normal')
- 05 – Carrier (CF)
- 07 – Not suspected (other disorder follow-up – 'normal')
- 08 – Suspected ('abnormal')
- 09 – Not screened / screen incomplete



Repeat Codes

Where a repeat is necessary the reason is reported:

- A – Raised Tyrosine
- B – Too young for reliable screening (< Day 5)
- C – Too soon after blood transfusion
- D – Unsuitable sample, e.g. over-layered or contaminated sample, card past expiry or outstanding information, excessive transit time (>14 days)
- E – Insufficient sample
- F – Unsatisfactory analysis
- G – Borderline Result (PKU/CHT/homocystinuria)
- H – Inconclusive CF
- K – Movement In, reason unknown

(G and H historically referred to as 'equivocal')



Giving results to parents

- Ensure that parents are informed about results by the 8-week assessment;
- Avoid use of ‘negative’ or ‘positive’ terminology – ‘condition not suspected’ or ‘condition suspected’ is less confusing for parents;
- Insert the extra copy of the laboratory result report into the PCHR (*after page 18*).

(refer to pages 13-14 of the Professional Handout for full guidance)



Following up outstanding results (1)

- **New CHS failsafe reports will be introduced and professionals will continue to be responsible for follow up of outstanding results for all livebirths and movement in infants**

(aged from day 11 to 1 year);

- **These infants need to be followed up as a matter of priority and according to the Trust's local Failsafe protocol;**

N.B. professional staff will be advised if there are any changes to their local protocol by their line manager / CHS manager.



Following up outstanding results (2)

- Do remember that bloodspot sample cards can get lost in transit and that it is important to follow-up test results **EVEN WHERE** there is evidence that a sample has already been taken;
- Where there is no documented evidence of screening test results available for movement in infants reported in failsafe **DO NOT DELAY** in offering bloodspot screening tests, as the follow-up of previous screening test results can take considerable time and resource and is often non-productive.

(refer to pages 28-29 of the Professional Handout for full guidance)



For more information

UK Newborn Screening Programme Centre
www.newbornbloodspot.screening.nhs.uk

About regional screening programmes:

A DHSSPSNI link will be available for information leaflets

Regional newborn screening lab contact person:

Department of Clinical Biochemistry

Kelvin Building

Royal Victoria Hospital

Grosvenor Road

Belfast

BT12 6BB

Telephone No.

Local contact points -Trust Leads (1)



Belfast HSCT

Northern HSCT

Southern HSCT

Local contact points - Trust Leads (2)



South Eastern HSCT

Western HSCT



Notification of Access to E-Learning Session for MCADD and Revised CF Implementation

***For the purposes of maintaining Trust training records
I have undertaken and achieved the identified aims of
this E-learning session and have access to the
complementary guidance and protocols discussed
during the session.***

SIGNED: _____ **DATE:** _____

PRINT NAME: _____

***(if requested, please print, complete and return this page to your
line manager as soon as possible and keep a copy for your own
records)***