



National Newborn Bloodspot Screening Programme



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Lecture Outline



- Role of the Clinical Liaison Nurse
- Screen positive cases
- Reasons for repeat sample requests
- Abnormal/borderline results follow up
- Communication with parents when repeat samples are required
- Importance of filling out Newborn Screening Card (NSC) correctly
- Resources available to PHNs

What is the role of the Clinical Liaison Nurse (CLN)?

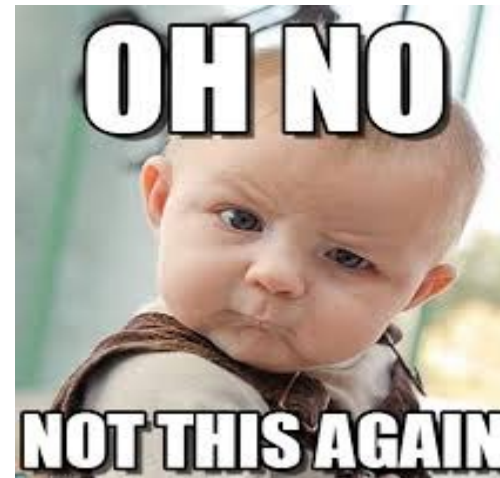
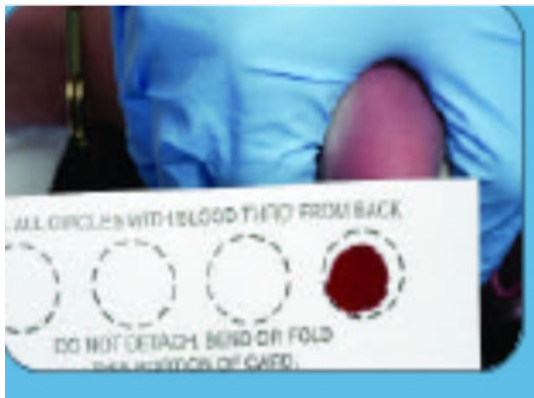
- Clinical coordination of the National Newborn Bloodspot Screening (NNBS) service, including the management and referral of 'screen positive' cases
- Ensuring urgent repeat samples are received and completing any additional follow up needed
- Educating sample takers
- Performing sweat tests as part of Cystic Fibrosis (CF) workup
- Performing audits to monitor programme performance and outcomes
- Developing parent and sample taker education material

What is the role of the Clinical Liaison Nurse (CLN)?

- Participating in process to expand the NNBSF programme
- Providing overview of NBS process to nursing and midwifery students
- Communication of results and follow up actions with Healthcare Professionals (HCP)s and parents
- Managing NNBSF databases and preparing case statistics and Key Performance Indicators (KPI)s for the NNBSF governance group
- Management and admin of eReports

Why repeat samples may be requested

- Insufficient blood on card
- Abnormal/borderline/equivocal result
- Unsatisfactory quality sample e.g. contaminated or diluted sample
- Baby <72 hours of age when sample was collected
- Baby not well established on full feeds when sample was collected
- Sample taken within 72 hours of an Red Blood Cell (RBC) transfusion
- Blood on card not dried properly before sending
- Sample taken on an expired NSC



Borderline/abnormal results

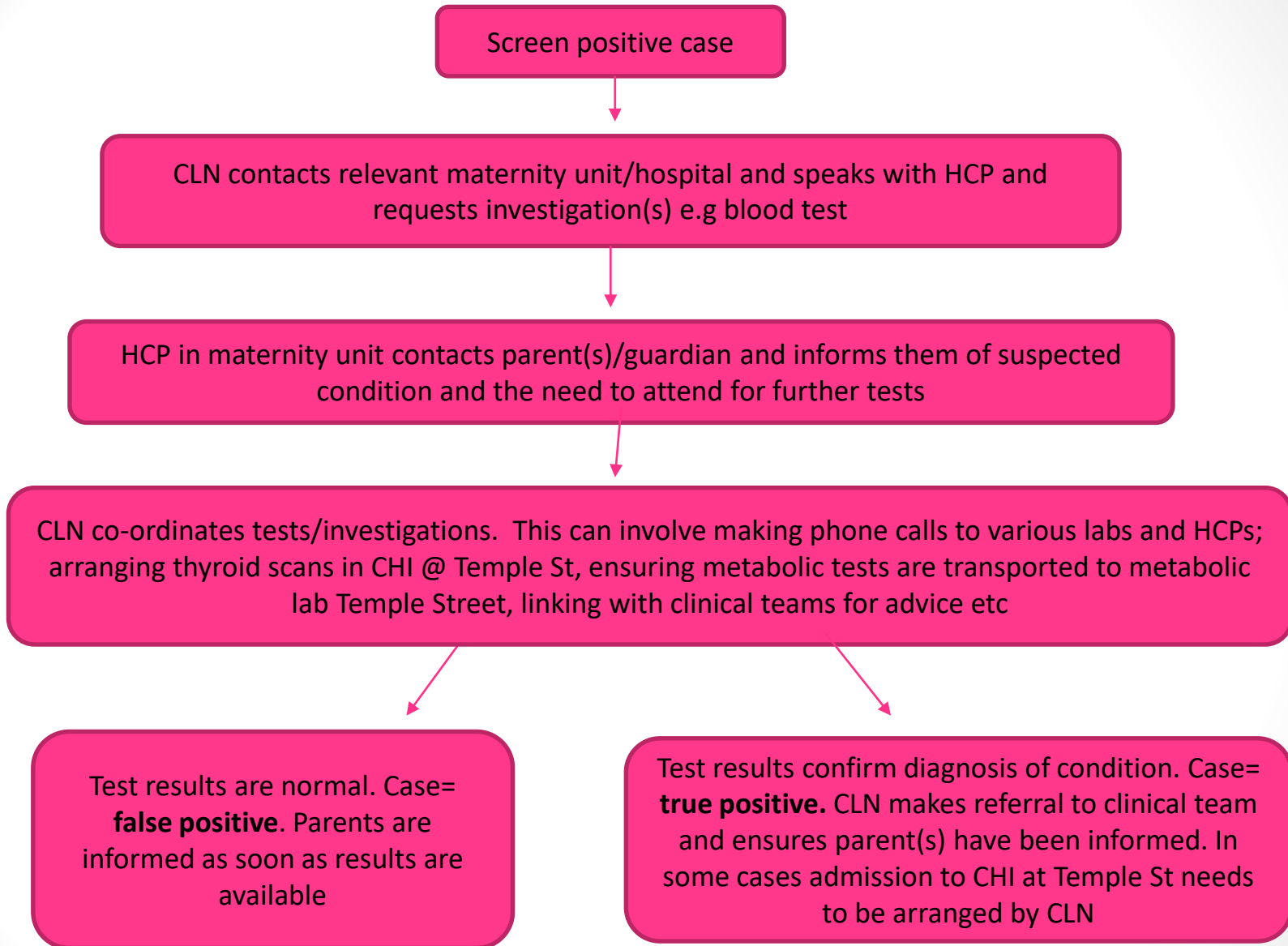
- There are different clinical pathways for each of the nine conditions currently screened for by the NNBSL.
- When a '**screen positive**' case is found, the CLN starts an 'action sheet' for that infant. (*see example on next slide*). Until a final diagnosis is reached (i.e true positive case or false positive), the infant is under the care of the NNBSL and it is the responsibility of the CLN to ensure the correct management/follow-up has occurred.
- Any cases with ongoing investigations that have not yet either been confirmed as either a false positive or true positive are known as 'open actions' within the NNBSL. In these cases we bypass a repeat sample and go straight to next tier testing.



Date: 10/04/23 Action by: A'OL Summary: ↑TSH
 PC ☐ DB ☐

Surname: <u>Doe</u>		Forename: <u>Jane</u>		UPI: <u>931-H312597</u>	
Lab No: <u>24-12345</u>		DOB: <u>04/04/23</u>		Hospital: <u>NMTH Holles St.</u>	
Location: <u>Community</u>					
Clinical Details		Contact Name and Number: <u>Nurse in NMTH baby clinic / neonate reg</u>			
Gest Age: <u>40</u>		FHX Thyroid disease: Y <input type="checkbox"/> N <input checked="" type="checkbox"/>		Surgery: Y <input type="checkbox"/> N <input checked="" type="checkbox"/>	
Wgt: <u>3.2kg</u>		Down Syndrome: Y <input type="checkbox"/> N <input checked="" type="checkbox"/>			
DBS results (µU/mL)		TSH	TSH repeat	Av	
1 st Card Date: <u>08/04/23</u>		<u>18</u>			
3 rd Card Date:					
2 nd Card Date:					
4 th Card Date:					
TFT Date	Processing lab:	TSH mU/L (>5.5)		Free FT4 pmol/L (ref c.)	
<u>10/4/23</u>	<u>NMTH</u>				
<p>NOTE: It is the birth hospital's responsibility to inform parents of a suspected case Any TSH > 5.5 mU/L requires Endocrine input regardless of external hospital range.</p>					
Date:	Notes:				Action Checklist: Inform:
<u>10/4/23</u>	<p><u>Called baby clinic in NMTH, spoke to nurse in charge, she will book Jane Doe for TFTs today.</u></p> <p><u>Bleeped reg to inform them. Spoke to Mary. She will phone parents and explain need to attend for TFT bloods. Advised Mary to pass on CLN number to parents in case they have further questions.</u></p>				<p>Scan 4566/4262 <u> </u>/<u> </u>/<u> </u></p> <p>Day ward 4806 <u> </u>:</p> <p>Endo Reg. bleep 968</p> <p><input checked="" type="checkbox"/> Bio Lab 4272 (dilution)</p> <p><input checked="" type="checkbox"/> Maternity Hospital</p>

Screen positive cases



Role of CLN in closing cases in NNBSL

Final diagnosis (false positive/true positive)



CLN:

- documents everything on patient's action sheet
- records final diagnosis electronically and on action sheet
- ensures parent(s)/guardian and all relevant HCPs are aware of final diagnosis
- ensures baby has been accepted under care of relevant clinical team in the case of a true positive
- add infant to relevant database for NNBSL case statistics
- files case away

URGENT

Repeat samples

-When a **borderline** result occurs, a repeat sample will be requested before an action sheet needs to be commenced. In most cases the medical scientist will report the borderline result in the baby's results report where it will clearly state which condition has had a borderline result and to repeat a sample as soon as possible. Any further instructions will be here also such as information to be put in the comment box of the NSC. *(see example on next slide).*

-If the team in NNBSL feel it needs to be acted on more urgently, they will link with the CLN and request that we phone the relevant PHN/hospital and request repeat sample. In this case the request on the baby's results report has probably not yet reached the unit/PHN.

*** In these cases it is essential to perform the repeat as soon as possible and communicate any issues/delays directly to the CLN or NNBSL ***

Sample taker name:

Contact number:

Location sample taken: Hospital place of birth


Screening results

Condition	Result	Result Codes	
		NNBSL	HSE
GA1 (Glutaric aciduria type 1)	Not Suspected	N	2
MCADD (Medium-chain acyl-CoA dehydrogenase deficiency)	Not Suspected	N	2
PKU (Phenylketonuria)	Not Suspected	N	2
HCU (Homocystinuria)	Refer to Text	RTT	6
MSUD (Maple Syrup Urine Disease)	Not Suspected	N	2
GAL (Classical Galactosaemia)	Not Suspected	N	2
CHT (Congenital Hypothyroidism)	Not Suspected	N	2
CF (Cystic Fibrosis)	Not Suspected	N	2
ADA-SCID (ADA Severe combined Immunodeficiency)	Not Suspected	N	2

Beutler Test: Activity Present

Classical Galactosaemia not suspected.

Note: Beutler result invalid if baby had red cell transfusion

 Baby not on full feeds (on TPN) when this card was taken - please take a repeat card when baby is on full-feeds.

NOTE: If baby not on full feeds a false normal for HCU in particular may be reported.

NOTE : Repeat screening card received, report to follow.

Communicating with parents about repeat samples

- All parents should be informed at the time of sampling that a repeat sample may be requested for multiple different reasons and this is not a reason for panic.
- Any baby that is not >36 weeks/not on full feeds/not 72 hours post RBC transfusion at time of initial NBS should be told that their baby will definitely require another sample.
- The reason for the repeat request will be stated on the baby's results report. This can be helpful when addressing parental questions e.g if it states '*insufficient for IRT please take repeat sample*' you can explain to the parent that there simply was not enough blood on the NSC to accurately screen for every condition.

If you have any queries about the reason noted, please contact CLN/NNBSL or refer to the practical guide for newborn screening for further information before going to take the sample

- If the report states that the first sample was borderline for one for the conditions, it is important to emphasise to the parents that **this is a borderline result, not a screen positive case**. If it was a very abnormal result, the baby would need to attend for bloods/further testing.
- **Remember:** If you are calm parents are more likely to be calm!



Communicating with parents about repeat samples

- Use your judgment to determine if parent(s)/guardian(s) need more or less information. Some parents are reassured by all the facts and figures and others find this information stressful.
- A basic knowledge of all nine conditions is important in order to address parental queries.
- Reassure parent(s)/guardian(s) of the importance of newborn screening and the benefit to their baby.
- Take all necessary steps to make the sampling process as straightforward as possible for the family and to minimise discomfort for the baby e.g have baby's heel kept warm in advance, allow mom to feed/hold baby during the procedure etc
- Use clear simple language when explaining a condition for example if you were describing CHT:
'The thyroid gland is located in the neck and makes a hormone called thyroxine. Thyroxine is needed for normal growth and development. Hypothyroidism is a condition that occurs when the thyroid gland is not able to produce enough thyroxine. Thyroxine (thyroid hormone) can be replaced easily with a medication given once a day. The thyroxine medication replaces the body's natural hormone which your baby's thyroid gland is not making'.

Remember: Direct parents to NNBSP website/HSE/advice them to contact CLN if they are worried. We are always happy to explain things to them in further detail.

Filling out the Newborn Screen Card (NSC)

- The NSC **must** be completely filled out with all information clear and legible. Errors made while filling out the card can lead to avoidable repeat samples being taken.
- Any relevant family history must be noted on the NSC. If the NNBSL is unaware of a positive family history, the correct high risk protocol won't be followed and additional testing won't be performed which could have a catastrophic impact on the infant.

Always ask when completing the NSC sample if there is any relevant family history- don't assume parents will tell you

- **Meconium Ileus** is a blockage in the small intestine of a newborn. There is an association between Cystic Fibrosis (CF) and babies with meconium ileus therefore it is essential to document meconium ileus on the newborn screening card. No other information regarding meconium needs to be specified on the card.

Filling out the Newborn Screen Card (NSC)

- All **feeds** must be clearly documented as certain feed types can influence the results of the NBS e.g If Soya-based feed not stated, test may give false negative result for Classical Galactosaemia
- **Parent's ethnicity** is important to note using the ethnicity codes on the back of the NSC. Certain ethnicities have higher incidences of some of the conditions screened for and it is therefore essential that the NNBSL is aware of.
- The date of baby's **first feed** must be noted so the NNBSL is aware how many days/hours baby has been feeding.
- Everything on the NSC is there for a reason, please leave no blanks.
- Take extra care with multiples when noting rank of the baby.
- Check the back to the NSC for guidance on filling out the card.

Remember: you can always contact CLN or NNBSL with questions regarding NSC

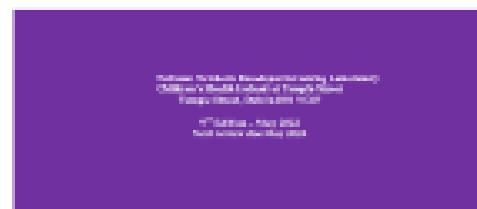


**DO IT ONCE.
DO IT RIGHT**

Resources:




**A Practical Guide to
Newborn Bloodspot Screening
in Ireland**
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hseland.ie

Heel prick screening

Every baby is offered newborn bloodspot screening, also known as the heel prick test. This screens for 9 rare conditions.

[About heel prick screening](#)

Learn about heel prick screening



[Conditions the heel prick screens for](#)

Heel prick screening checks for 9 rare but serious conditions in babies.



[Getting the heel prick screen](#)

Learn about the heel prick screen



[Getting your baby's heel prick results](#)

Learn about your baby's heel prick results



