Cheshire and Wirral Partnership NHS
NHS Foundation Trust

Document level: Clinical Service Unit (CSU)
Code:CC33
Issue number: 1

Newborn Blood Spot Screening Guidelines

Lead executive	Director of Nursing Therapies Patient Partnership
Authors details	Clare Smalley, Health Visitor, 01829 733686, Peter Hardy

Type of document	Guidance
Target audience	All community staff Health Visiting service
Document purpose	Newborn Blood Spot guidance

Approving meeting	West Locality and Governance meeting	21/06/16
Implementation date	21-Jun-16	

CWP documents to be read in conjunction with	
HR6	Trust-wide learning and development requirements including the training needs analysis (TNA)

Document change history	
What is different?	
Appendices / electronic forms	Have appendices been added or changed since the last issue, if so explain the reasons why?
What is the impact of change?	Will this new document change the way we do things currently

Training requirements	Yes / No - Training requirements for this policy are in accordance with the CWP Training Needs Analysis (TNA) with Learning and Development (L&D)

Financial resource implications	No

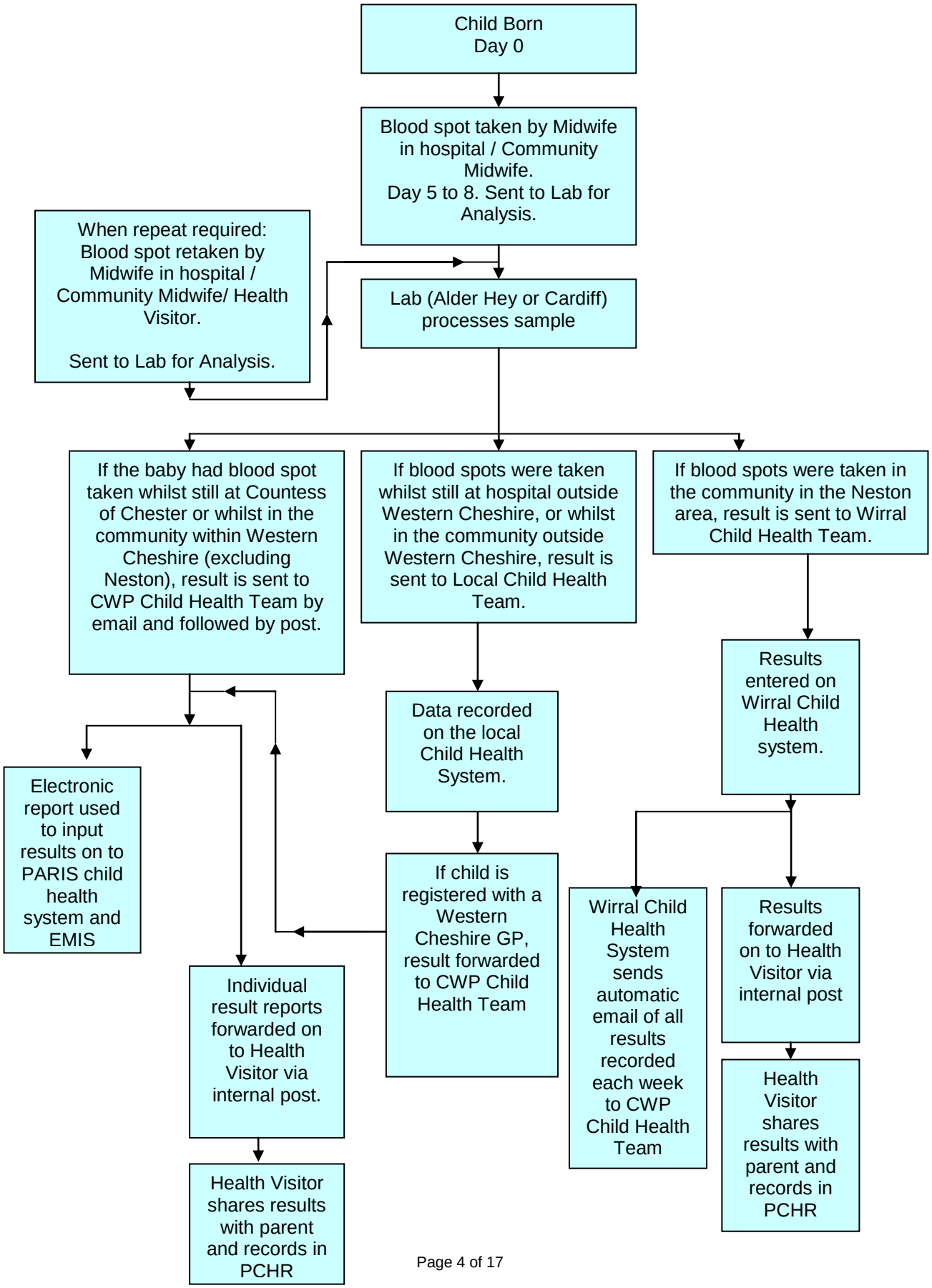
External references

Equality Impact Assessment (EIA) - Initial assessment	**Yes/No**	**Comments**
Does this document affect one group less or more favourably than another on the basis of:		
- Race	No	
- Ethnic origins (including gypsies and travellers)	No	
- Nationality	No	
- Gender	No	
- Culture	No	
- Religion or belief	No	
- Sexual orientation including lesbian, gay and bisexual people	No	
- Age	No	
- Disability - learning disabilities, physical disability, sensory impairment and mental health problems	No	
Is there any evidence that some groups are affected differently?	No	
If you have identified potential discrimination, are there any exceptions valid, legal and/or justifiable?		

Equality Impact Assessment (EIA) - Initial assessment	Yes/No	Comments
Is the impact of the document likely to be negative? - If so can the impact be avoided? - What alternatives are there to achieving the document without the impact? - Can we reduce the impact by taking different action?	No N/A N/A N/A	
Where an adverse or negative impact on equality group(s) has been identified during the initial screening process a full EIA assessment should be conducted. If you have identified a potential discriminatory impact of this procedural document, please refer it to the human resource department together with any suggestions as to the action required to avoid / reduce this impact. For advice in respect of answering the above questions, please contact the human resource department.		
Was a full impact assessment required?	No	
What is the level of impact?	Low	

Content

Quick reference flowchart - Overview of Newborn Blood Spot Screening Process for Western Cheshire.



1. Introduction

Newborn screening aims to identify babies who are at high risk of having certain serious but rare conditions before they develop symptoms. Screening is not the same as diagnosis: instead it identifies which babies need to go on to have more diagnostic tests to determine whether or not they do have a condition. By detecting these conditions early it is possible to treat them and to reduce their severity. Newborn blood spot screening is a crucial part of a national child public health programme and is offered to all babies in the United Kingdom.

There are currently nine conditions screened for in Western Cheshire:

- Congenital Hypothyroidism (CHT)
- Phenylketonuria (PKU)
- Sickle Cell Disorder
- Cystic Fibrosis (CF)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
- Homocystinuria (HCU)
- Maple Syrup Urine Disease (MSUD)
- Glutaric Aciduria Type 1 (GA1)
- Isovaleric Acidaemia (IVA)

HCU, MSUD, GA1 and IVA are collectively known as Inherited Metabolic Disorders (IMD)

1.1 Congenital Hypothyroidism (Cht)

Congenital Hypothyroidism is a condition where there is decreased or no thyroid hormone production. Early diagnosis is important as the effects of hypothyroidism are easy to reverse. If left untreated it can lead to the development of severe learning disabilities and delayed growth. If identified early the baby can be treated and can lead a healthy life.

1.2 Phenylketonuria (PKU)

Phenylketonuria (PKU) is an inherited metabolic condition where there is a defect in phenylalanine hydroxylase. This enzyme normally converts the phenylalanine in the body into tyrosine. Where there is an enzyme block the phenylalanine accumulates in the body tissues and affects the normal development of the brain causing learning difficulties. If untreated this leads to poor brain development. Early identification allows the baby to be put on a special diet and the brain can develop normally.

1.3 Sickle Cell Disorders

Sickle cell anaemia is an inherited condition where by a haemoglobin S gene is inherited from both parents. If the baby inherits one S gene from one parent this results in the baby being a carrier (The child would have no symptoms).

Sickle cell anaemia is a condition that affects the normal oxygen-carrying capacity of the red blood cells. When the cells are de-oxygenated and under stress in sickle cell conditions, they can change from round flexible disc-like cells to elongated sickle or crescent moon shape. The effect of these changes is that the cells do not pass freely through small capillaries and form clusters, which block the blood vessels. The blockage prevents oxygenation of the tissues in the affected areas resulting in tissue hypoxia and consequent pain. Sickle cell disorders are lifelong conditions.

Babies who are healthy carriers of sickle cell trait are also identified through the screening process. Although an infant with sickle cell trait does not have the problems of sickle cell disorder, if his or her parents are both carriers they could have another child who does have a sickle cell disorder.

1.4 Cystic Fibrosis (CF)

Cystic Fibrosis is a common inherited disease; around 1:25 are carriers of the faulty gene in the United Kingdom. The faulty gene that causes CF is recessive which means that an individual with the disorder has to have acquired a faulty gene from each parent. Carriers do not have the disease but their children may inherit the condition. CF is a condition that affects certain organs in the body, especially the lungs and pancreas, by clogging them with thick sticky mucus. The thick secretions in these organs cause digestive problems and chest infections. Early diagnosis is important as babies with CF are treated vigorously as soon as they are first diagnosed. Treatment of children with CF aims to do two things:

- Improve nutrition by providing supplements containing enzymes to help digestion
- Reduce chest infections with frequent physiotherapy and either occasional or continuous antibiotics.

1.5 Medium Chain Acyl Coa Dehydrogenase Deficiency (MCADD)

MCADD is an inherited metabolic disorder where there is lack of an enzyme required to convert stored fat to energy. If MCADD is not identified at an early age, up to a quarter of affected children may die from the condition, with one third of surviving children sustaining significant neurological damage. Treatment involves ensuring that children do not go for long periods without food although periods can increase as the child grows.

1.6 Homocystinuria

HCU is an inherited metabolic disorder affecting 1:144,000 babies born in the UK, in which the body has difficulty in breaking down the amino acid homocysteine resulting in an accumulation of this amino acid in the blood. Without treatment most children develop learning difficulties, eye problems, osteoporosis, blood clots and stroke.
Treatment is with Vitamin B6 medication or with a low protein diet and supplements.

1.7 Maple Syrup Urine Disease

MSU is a rare inherited metabolic disorder affecting about 1:116,000 babies born in the UK, in which the body has difficulty breaking down the amino acids leucine, isoleucine and valine, resulting in harmful accumulation in the blood.
Untreated babies can become unwell within a few days of birth with poor feeding, vomiting and excessive sleepiness, which can lead to coma, brain damage or death.
Treatment is with a reduced protein diet and supplements with an Emergency Regimen during illness.

1.8 Glutaric Aciduria Type 1

GA1 is an inherited metabolic disorder affecting 1:110,000 babies born in the UK, in which the body has difficulty breaking down the amino acids lysine and tryptophan. Early signs within days of birth may be vomiting, irritability, sleepiness and breathing difficulties.
Without treatment babies may go into a coma with permanent brain damage affecting the ability to control muscles and movements.
Treatment is with a low protein diet and Carnitine and an Emergency Regimen during illness.

1.9 Isovaleric Acidaemia

IVA is an inherited metabolic disorder affecting 1:155,000 babies born in the UK, in which the body is unable to break down the amino acid leucine. Young babies can become severely unwell within days of birth resulting in coma and death.
IVA may vary in severity and may also affect older children.
Treatment is with a low protein diet and supplements with an Emergency Regimen during illness.

2. Aims of This Guideline

- To ensure that CWP Health Visiting Service complies with the UK Newborn Screening Programme Centre policies and standards, and with the National Service Specification for the Newborn Blood Spot Screening Programme
- To provide clear guidelines to the Screening Link Health Visitors (SLHV), members of the Health Visiting Teams and the Child Health Department
- To support the delivery of a safe and effective screening service for repeat samples
- To support the delivery of 'suspected' results, 'not suspected' results and carrier results to families in a timely manner
- To support the detection of unscreened babies moving into Western Cheshire

3. Outcomes

- Screening Link Health Visitors (SLHV) and members of the Health Visiting Teams and Child Health will comply with the UK Newborn Screening Programme Centre policies and standards, and Service Specification to reduce the likelihood of late or missed diagnosis.
- All Screening Link Health Visitors will be aware of procedures for handling repeat blood spot samples.
- All Screening Link Health Visitors will have clear guidelines for dealing with babies who move into the area who do not have any record of bloodspot screening recorded.
- Improved communication between professionals and between professionals and parents
- Screening Link Health Visitors and Health Visitors will have clearly defined roles and responsibilities.
- The Child Health Team will have clear guidelines for the recording and management of results
- Parents and carers will receive results in a timely manner.

4. Target Group

All members of the health visiting teams designated screening link health visitors and child health employees employed by CWP are required to follow this procedure.

5. Related Policies And Documents

This list is not exhaustive; it highlights the most relevant policies for the safety of patients and staff:

- CWP Record keeping Policy
- UK Newborn Screening Programme Centre (2013) Standards for Newborn Blood Spot Screening
- NHS Public Health Functions Agreement 2015-16, Service Specification no.19, NHS Newborn Blood Spot Screening Programme
- UK Newborn Screening Programme Centre (2012) Guidelines for Newborn Blood Spot Sampling
- UK Newborn Screening Programme Centre (2008) Standards and Guidelines for Newborn Blood Spot Screening

6. Screening Link Health Visitor Responsibilities

The screening link health visitor, who has received additional training in screening, is responsible for:

- Following the Newborn Blood Spot Screening Guidelines (CWP 2015).
- Undertaking newborn blood spot screening on children moving into the area, under the age of one, when there is no evidence of screening results.
- Undertaking newborn blood spot screening on children who have not been screened and are over 28 days old.

- Following up referrals for repeat blood spot screening from Alder Hey laboratory. A repeat sample following raised IRT levels (possible CF) should ideally be taken on day 21.
- Visiting the family when a child has a 'suspected' blood spot result for CF. The laboratory in conjunction with the hospital will contact the SLHV and notify them of the time and place for an appointment for the family to attend. The SLHV will arrange to visit the family in the late afternoon the day before the appointment. Preferably with the Health Visitor. The SLHV will inform the parent of the 'suspected' screening result, discuss and leave the leaflet 'Cystic Fibrosis is suspected' and give the date, time and venue for the sweat test and appointment with the Consultant.
- Visiting the family when a child has a 'suspected' blood spot test for MCADD. The laboratory in conjunction with the hospital will contact the SLHV and notify them of the time and place for an appointment for the family to attend. The SLHV will arrange to visit the family in the late afternoon of that same day, preferably with the Health Visitor, and inform the parents of the 'suspected' screening result. The SLHV will inform the parents of the hospital appointment, and leave the leaflet 'MCADD suspected'.
- Visiting the family when a child has been identified as having a carrier status for CF or Sickle Cell Disorder. Discussing the results with parents and leaving the appropriate leaflets.
- The accurate documentation of all initial and repeat screening, and the sharing of all 'suspected' and carrier results on the communication sheet (see Appendix 4) and in the Personal Child Health Record (PCHR). The communication sheet should be sent to the Health Visitor and GP, and scanned into EMIS records.
- NB. When a child has a 'suspected' result for MSUD, IVA, GA1 or HCU the SLHV will be contacted by Alder Hey for information sharing only. The parents will be contacted by a Manchester or Alder Hey Consultant Paediatrician for urgent assessment and to initiate referral.

7. Health Visitor Responsibilities

- Health Visitors to assess at birth visit whether baby has been screened. Untested babies are defined as those babies who do not have evidence of screening or a decline notification for each of the conditions for which screening is offered. Any baby identified at the birth visit who has not been screened should be referred to the Community Midwife for timely screening to be arranged.
- On identification of untested babies, the screening process should be 'fast- tracked' for parents wishing to have their child screened and the pre-testing leaflet provided. (Newborn Blood Spot Screening for Your Baby NHS 2015)
- If the baby is under 28 days old and has not been screened, the Health Visitor should contact the Community Midwife team to arrange a sample to be taken. Over 28 days old, the Health Visitor should contact the SLHV via referral form. (See Appendix 5).
- The Health Visitor will discuss with parents/carers at the birth visit when they will receive the results of the screening tests.
- All 'not suspected' results received from Child Health will be discussed with the parents/carers at the 6-8 week contact. Results will be recorded in the Personal Child Health Record (PCHR) and in the Birth Book. Health Visitors should also record that results have been shared with parents on the 6-8 week EMIS template. This will be subject to local audit.
- If results have not been received by the 6-8 week contact and there has been no contact from any other health professional to indicate that the results are abnormal, the Health Visitor will need to contact the Child Health Department or screening laboratory at Alder Hey Children's Hospital to follow up the results.
- For Health Visitors seeing babies under one year of age who have moved into the area and are reported to have been screened, evidence of testing is required. This may take the form of a faxed or written confirmation of the results from child health or regional screening laboratory. The Newborn Screening Laboratories Network (www.newbornscreening.org) has details of UK screening laboratories. Screening results need to be sent to the Child Health Department within 21 days of a transfer being notified to them. Where the Health Visitor finds there is not proof of testing available, it should be assumed that the baby is untested and retesting

discussed with the parents and arranged by contacting the SLHV. If the baby is older than 8 weeks, blood spot screening should be offered for CHT, PKU, Sickle Cell Disorders, MCADD, MSUD, IVA, GA1 and HCU; parent/carer should be informed that screening will not pick up CF as serum levels return to normal after 8 weeks. If the parents do not want testing/retesting then this should be fed back to Child Health that parents have been offered and refused testing so that it can be recorded on the Child Health system.

- Health Visitors receiving results that the baby is a healthy carrier for Sickle Cell Disease or Cystic Fibrosis will need to contact the SLHV to discuss results with parents.

8. Looked After Children

For any child who is looked after by the local authority, consent must be obtained by the person who has parental responsibility i.e. social work manager, and fully documented in the child's health records.

9. Parents/Carers Who Decline

If parents decline testing, the reason for their decision should be explored and further information offered. However, parents should not be unduly pressured, although they need to be making an informed choice.

The SLHV or HV must:

- Document that the parent has declined a test, including reasons for decision, on the Newborn Blood Spot Screening Communication Form (Appendix 4) scanned onto EMIS and Personal Child Health Record.
- Confirm the parents understand the risks of the baby not being screened.
- Offer further information and who to contact if they change their minds.
- Inform the General Practitioner, Child Health Department and Alder Hey Laboratory.

10. Performing the Procedure

Taking the blood spot involves balancing the need to collect sufficient blood, with the potential for discomfort for the baby and unease for the parents. The procedure should be carried out in accordance with the UK Newborn Screening Programme Centre (2012) Guidelines for Newborn Blood Spot Sampling.

11. Clinical Incidents

Any related incidents arising from carrying out these procedures, which may involve clinical error or near miss, must be reported following the CWP incident reporting policy.

12. Child Health Team

Cheshire and Wirral Partnership NHS Foundation Trust's Child Health Team cover all children with a Western Cheshire GP, except Willaston Surgery, Neston Medical Practice and Neston Surgery; Wirral Child Health Team covers these three practices.

As per the National Screening Guidelines, CWP's Child Health Team are responsible for recording the newborn blood spot screening results on the Child Health System and also for running a daily search of the Child Health System for children who are between the age of 14 days and 1 year, who do not have any newborn blood spot results recorded.

The Child Health Team receives the newborn blood spot results daily from the Newborn Screening laboratory by email, which is followed up by an individual report per child which arrives in the internal post.

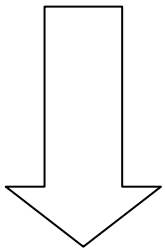
The Child Health Team records the newborn blood spot results within 1 working day of receipt of the email. Once these have been recorded the email is retained within the Child Health Team's email account for two years in line with the Trust's document retention policy.

When the individual reports arrive from the laboratory the Child Health Team check that the result is recorded on the Child Health System and scans it into the EMIS record. The result is then forwarded to the appropriate Health Visitor within one working day.

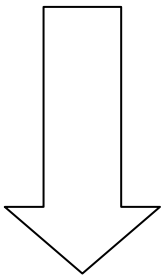
A report is run daily by the Child Health Team from the Child Health System of all children who are between fourteen days and one year of age and who do not have any newborn blood spot results (or a refusal) recorded. The Child Health Team then contacts the appropriate laboratory to request results. If the laboratory has not received the screening sample, the Child Health Team informs the Community Midwives Office or Health Visitor to arrange for the child to be screened (see Appendix 7). All phone calls/and emails in relation to the daily check are stored with the daily check in the Child Health Office for one year.

Appendix 1 - Flowchart for Repeat Newborn Blood Spot Screening

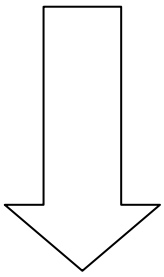
Laboratory will contact Screening Link Health Visitor to request further sample. If Health Visitor directly contacted by laboratory or midwife for repeat, Health visitor to contact Screening Link Health Visitor.



Screening Link Health Visitor will contact Health Visitor for family details and arrange visit informing family of the reason for the request.

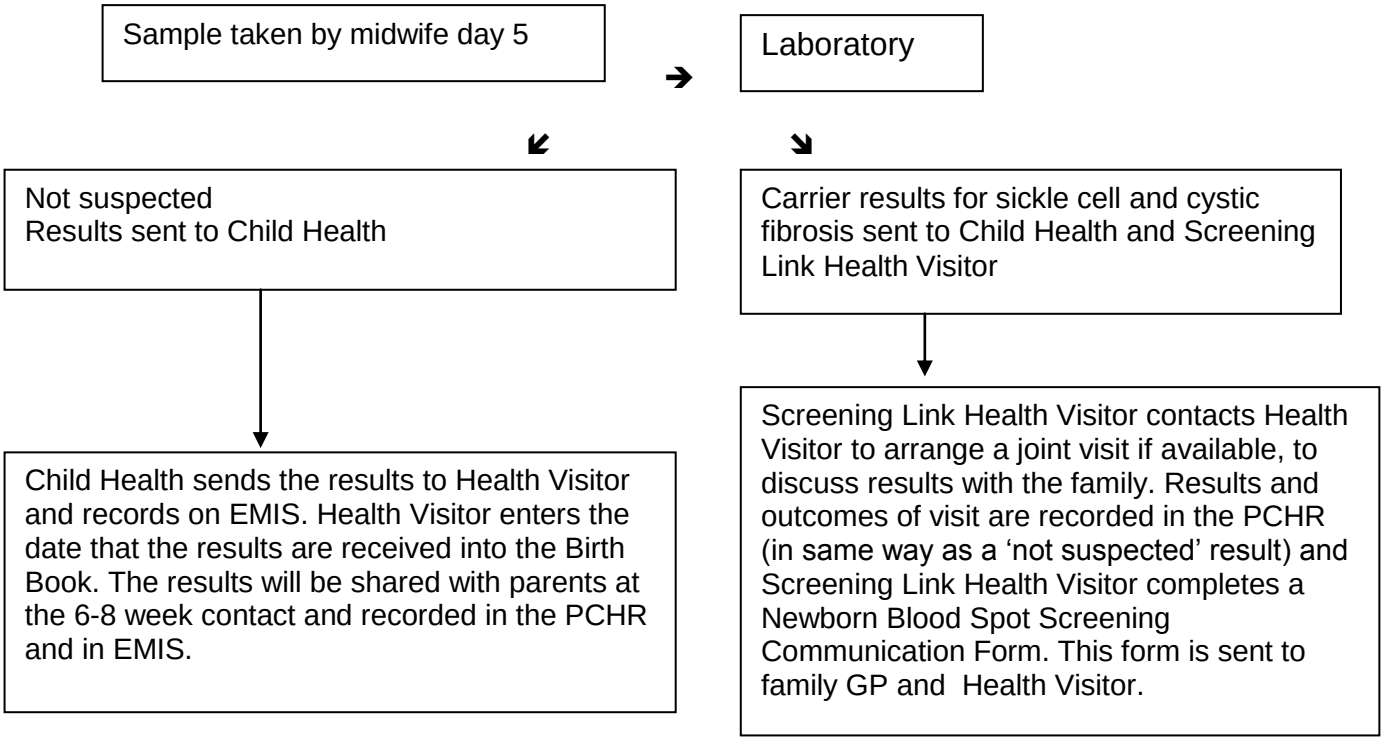


Informed consent for blood sample must be obtained and recorded in PCHR. Newborn Blood Spot screening procedure for performing heel prick followed.



Screening Link Health Visitor to record sample taken on Newborn Blood Spot Communication Form (appendix 4) and send sample off by post on the same day as taking sample.

Appendix 2 - Overview of Screening Pathway For Not Suspected And Carrier Status Results



Any queries about results received from Child Health, please phone Alder Hey lab on

0151 252 5489

Appendix 3 - Overview Of Screening Pathway For Suspected Results

PKU and CHT

Families will be informed of results by specialist teams from Alder Hey or COCH.

This does not involve either the SLHV or Family Health Visitor

Sickle Cell or other Haemoglobinopathy

Families will be referred to the haemoglobinopathy counsellor/specialist nurse at Alder Hey by the lab and offered genetic counselling at Alder Hey or Countess of Chester Hospital

MCADD

If a child has a MCADD suspected screening result s/he has to be seen the next day for a repeat test in hospital.

The hospital will contact a Screening Link Health Visitor and notify them of the time and place for an appointment for the family to attend the following day.

The Screening Link Health Visitor will then contact the Health Visitor (or team) and arrange to go out to visit the family in the late afternoon of that same day. The Screening Link Health Visitor will inform the parents of the hospital appointment and ascertain whether the child is feeding well. The leaflet ‘MCADD is suspected’ is given.

Cystic Fibrosis

If a child has a suspected CF screening result s/he has to be seen in the next day or two for a sweat test.

The Laboratory and hospital will contact a Screening Link Health Visitor and notify the time and place for an appointment for the family to attend.

The Screening Link Health Visitor will then contact the Health Visitor (or team) and arrange to go out to visit the family in the late afternoon the day before the appointment. The Screening Link Health Visitor will inform the parents of the CF suspected screening result, discuss and leave the leaflet ‘Cystic Fibrosis is suspected’ and give the date, time and venue for the appointment with the Consultant.

Inherited Metabolic Disorders (IMD)

If a child has a suspected result for an IMD they will be contacted directly by a Consultant Paediatrician from Manchester or Alder Hey for immediate assessment and to initiate hospital referral. The SLHV will be notified

Appendix 4 - Newborn Blood Spot Screening Communication Form

Child's Name	
Date of Birth	
Child's NHS Number	
Family address	
Family telephone	
Family GP	
HV base and telephone no.	
Communication difficulties	
Relevant clinical information	
Relevant family history	

Results discussed for Sickle Cell Trait	
Results discussed for Cystic Fibrosis Trait	
Hospital Appointment Cystic Fibrosis suspected discussed	Yes/No
Hospital Appointment MCADD suspected discussed	Yes/No

Neonatal Blood Sample

	Date sample taken	SLHV signature
PKU		
CHT		
Haemoglobinopathies		
Cystic Fibrosis		
MCADD plus IMD's		

Further information, i.e. informed consent obtained, consent declined, leaflets given: **Family Health Visitor**: Please ensure results discussed with family and document in PCHR and EMIS

Name of HV Bloodspot Screener	
Distribution	**Date**
Completed form sent to family GP	
Completed form sent to family Health Visitor	

Appendix 5 - Health Visitor Newborn Screening Referral Form

Name of Child	
Previous Surname (if different at birth)	
Sex (M/F)	
Date of Birth	
NHS Number	
Family address	
Family GP	
Family Telephone Number	
Family Health Visitor	
Health Visitor base/contact number	

Referral Details

Reason for Referral	
Has referral been discussed with family?	

Please forward to Screening Link Health Visitor:

Jan Reeves janreeves@nhs.net
Mercury House,Tattenhall
01829 771823
07717 513452

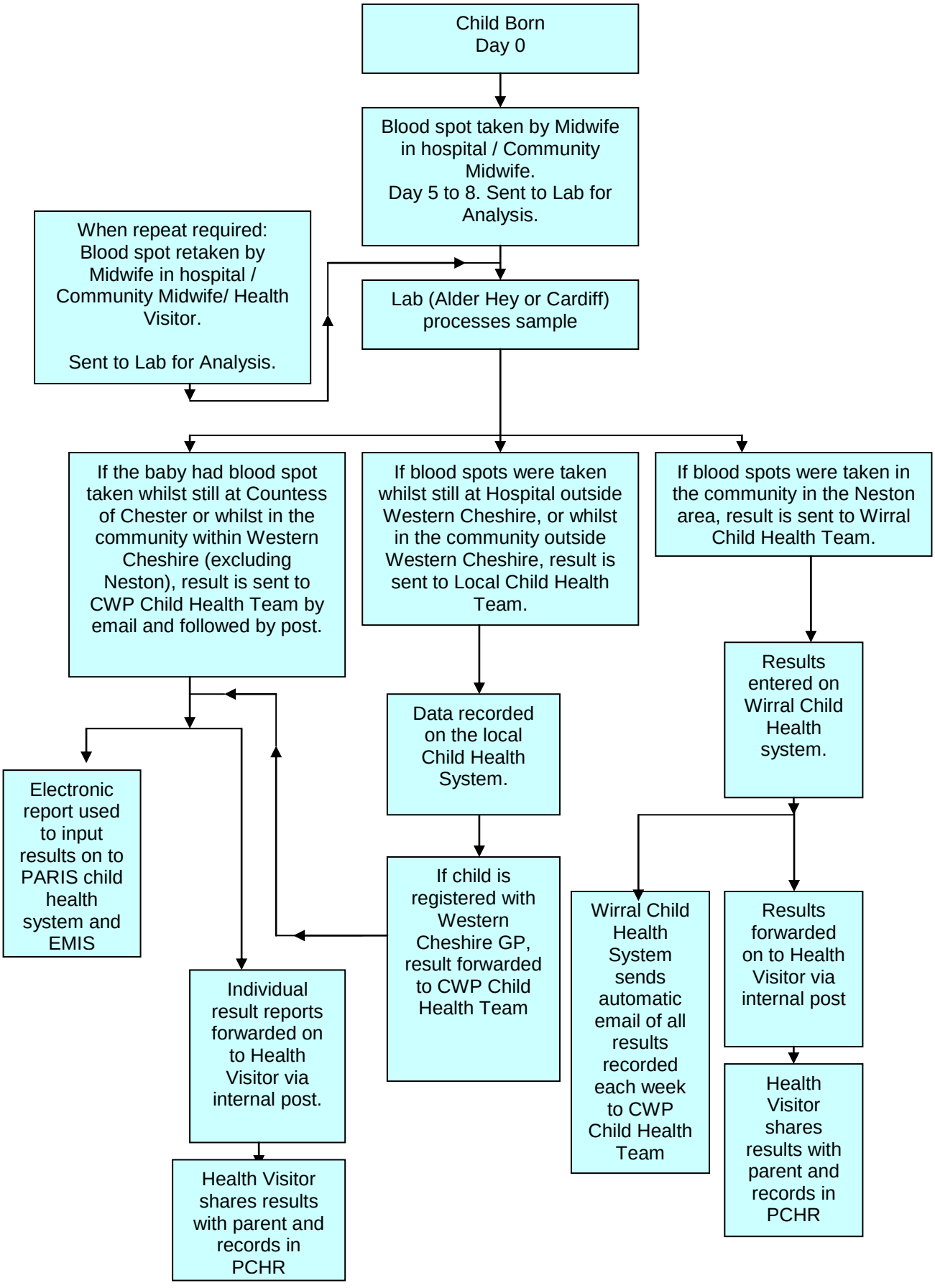
Becky Marsh rebeccamarsh@nhs.net
Tarporley Health Centre
01829 733686
07717 535050

Donna Hardcastle donna.hardcastle@nhs.net
Upton Village Surgery
01244 398010
07557 172419

Rebecca Mayne rebeccamayne@nhs.net
Stanney Lane Clinic
0151 350 3325
07557 172416

Clare Smalley csmalley@nhs.net
Tarporley Health Centre
01829 733686
07717 535158

Appendix 6 - Overview of Newborn Blood Spot Screening Process for Western Cheshire.



Appendix 7 - Child Health Team's Daily Sweep for Children with outstanding Blood Spot screening results between 14 and 365 days of age.