1. **Aim/Purpose of this Guideline**

1.1 The purpose of screening as defined by The National Screening Committee is a process of identifying apparently healthy people who may be at increased risk of a disease or condition. They can then be offered information, further tests and appropriate treatment to reduce their risk and/or any complications arising from the disease or condition.

This document has been developed to provide clear pathways of care for women undergoing screening in the antenatal and new-born period. All of the screening programmes offered, comply with the standards set by the individual screening programmes from the National Screening Committee. One of the primary aims of screening is to enable parents to make informed choice concerning their pregnancy outcome.

This policy applies to all midwives, health visitors and medical staff involved in counselling women regarding their screening choices and any staff delivering the screening programmes, including laboratory staff and radiology department.

2. **The Guidance**

2.1. **Principles for Providing Screening Tests**

Women should be provided with written information in the form of “Screening Tests for You and Your Baby” in an appropriate language or format suitable to their needs, prior to their first booking appointment with a community midwife.

Women must be given the opportunity to discuss their options with health care professionals in a timely and non-directive manner to support informed decision making.

All screening test results should be reported to women in a timely and appropriate way.

- All negative results from infection screening and Sickle Cell & Thalassaemia screening, following the initial booking, will be recorded in the woman’s hand held notes at the 15 week/next appointment and signed and dated by the midwife.
- All low risk results from the Downs, Edwards and Patau’s Syndrome Screening will be sent to the women by post, to be received within 7 working days following the screen. This will then be recorded in the woman’s hand held notes at the 15 week/next appointment and signed and dated by the midwife.
• A copy of the 1st trimester and 18 – 20+6 week anomaly scan will be inserted into the hand held notes and a copy filed in the main health records

• All new-born blood spot screening results will be reported to the mother by 8 weeks and recorded in the Personal Child Health Record

• Results of the Newborn and Infant Physical examination will be recorded in the Personal Child Health Record

Precise and accurate records should be maintained in the women’s hand held notes to reflect choice and allow audit of the service.

2.2. Role of the Antenatal and Newborn Screening Governance Group

All of the Antenatal and Newborn Screening programmes will be monitored for their compliance against national standards by the Antenatal and Newborn Screening Governance Group (ANSGG). This group will be led by a Consultant in Public Health. Terms of reference for this group are attached at (Appendix 3).

The group will meet half yearily to review policy, ratify changes, implement and embed new practice and develop and monitor any changes to the screening programmes.

The group will monitor compliance with the approved documentation, which describes the process for ensuring that all appropriate maternal and new-born screening tests are offered, undertaken and reported on during the antenatal and post natal period. Evidence will be produced by audit and this will be presented to the group by the ANNSC which will demonstrate:

• Process for the review of the results has been followed as described. (Audited annually as part of the compliance monitoring process)

• Process for ensuring that women with screen positive test results are referred and managed within appropriate timescales as per individual policies. (This will be audited continually by way of data collection and within the annual report. This report will be reviewed by the ANSGG before being submitted to Trust and regional board.) Any deficiencies will be brought to the ANSGG and an action plan developed and monitored.

• Systems for ensuring that appropriate tests are undertaken within appropriate timescales. (This will be audited monthly as part of the Key Performance Indicators and annually as part of the Annual Report, both of which will be submitted both at Trust board and regional level).

• Systems for ensuring that appropriate tests are undertaken when women book late. (This will be audited monthly and reported to the Team Leaders forum).

• Process for reporting all results to women. (Audited annually as part of the compliance monitoring process).

• Process for reporting results to other relevant health care professionals. (This will be audited annually as part of the Annual Report, which will be submitted both at Trust board and regional level).
Where the monitoring has identified deficiencies, the ANSGG will develop action plans to address them. An Annual report will be produced by the screening co-ordinator which will be ratified by the ANSGG before being released to the Trust Board, the CCG and the Regional Screening team.

The ANSGG will also scrutinise the Key Performance Indicator data for each screening programme to ensure that standards are being met.

2.3. **Role of Individual Staff**

All midwifery staff are responsible for ensuring that women are offered screening in a timely manner as per the national standards. Midwifery staff will be responsible for ensuring that the “Did Not Attend” policy is put into process when women fail to attend for routine appointments. Following screen positive results, the responsibility for following women up that do not attend for referral appointments, will be with the referral unit and will include communication with the community midwifery team.

2.4. **Role of laboratory staff**

Laboratory staff involved in the screening process will be responsible for following the Standard Operating Procedure in place for each programme.

2.5. **Role of the Antenatal and Newborn Screening Co-ordinator**

The Antenatal and Newborn Screening Co-ordinator (ANSC) will be responsible for the co-ordination of all NHS antenatal and new-born screening programmes within the trust consistent with UK National Screening Committee recommendations (UK NSC) and national programme standards.

The ANSC will be a member of the multidisciplinary team supporting clients and staff involved in the delivery of antenatal and new-born screening programmes. She will be responsible for the development, implementation and local audit of policies, guidelines and pathways which will include performance management and quality assurance of the programmes. Education and training will be a key aspect of the role to ensure all staff is both competent and confident in the delivery and management of existing and new screening programmes.

The ANSC will have highly developed specialist knowledge and experience in the field of antenatal and new-born screening, the skills to impart this knowledge and to lead a team effectively at local level.

2.6. **Standards and Practice**

RCHT offers the following screening programmes as required by the National Screening Committee and the National Institute for Health and Clinical Excellence.

- Sickle Cell and Thalassaemia – all women will be offered screening for Thalassaemia and women with a positive Family Origin Questionnaire will be offered screening for Sickle Cell disorder. (See Appendix 4: RCHT Map of Medicine Pathway Document for Sickle Cell and Thalassaemia Screening)

- Infectious diseases including:
  - Syphilis
  - Hepatitis B
• HIV (Human Immunodeficiency Virus)  
  (See Appendix 5: RCHT Map of Medicine Pathway Document Screening for Infectious Diseases in Pregnancy)

• Down’s Syndrome, Edwards’ Syndrome and Patau’s syndrome which is supported by early pregnancy ultrasound scanning. (See Appendix 6: RCHT Map of Medicine Pathway Document for Antenatal Screening for Downs Syndrome)

• Early pregnancy ultrasound scan from 12 to 14 weeks (but can be up to 18 weeks), for women declining Down’s, Edwards’ and Patau’s Syndrome screening

• Chemical screening for Down’s Syndrome for women unable to complete first trimester screening by 14 weeks. This can be completed between 14-20+6 weeks.

• Fetal anomaly ultrasound scans between 18 and 20+6 weeks. (See Appendix 7: RCHT Map of Medicine Pathway Document for Fetal Anomaly Ultrasound at 18 – 20+6 weeks)

• Newborn Blood Spot Screening. (See Appendix 8: RCHT Map of Medicine Pathway Document for Newborn Blood Spot Screening)

• Newborn and Infant Physical Examination

2.7. **RCHT Policy Documents for the Screening Programme**

RCHT Map of medicine pathway documents for the individual screening programmes contain as a minimum the following clearly defined elements:

• Arrangements for giving pre-test information and offering the test
• Arrangements for provision of the test
• Arrangements for record keeping
• Arrangements to ensure timescale standards are met
• Arrangements for the handling of results
• Arrangements for specialist counselling and support when required
• Arrangements for necessary antenatal and immediate post natal care and follow-up
• Arrangements for referral to other agencies as required
• Failsafe processes to ensure that all screening has been completed

3. **Monitoring compliance and effectiveness**

<table>
<thead>
<tr>
<th>Element to be monitored</th>
<th>All of the Antenatal and Newborn Screening Programmes will be monitored for their compliance against national standards.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lead</td>
<td>Antenatal and Newborn Screening coordinator (ANNSC)</td>
</tr>
</tbody>
</table>
| Tool | Were all negative results from infection screening and Sickle Cell & Thalassaemia screening and all low risk results from the 1st trimester screening recorded in the woman's notes at the 15 week/next appointment and signed and dated by the midwife. 20 consecutive sets of notes of women who have delivered will be audited as per the screening audit programme and the results reviewed by the ANSGG.  
| Tool | Was a paper copy of the first trimester and anomaly scan filed in the main health records. 20 consecutive sets of notes of women who have delivered will be audited as per the screening audit programme and the results reviewed by the ANSGG.  
| Tool | Was the process for ensuring that women with screen positive test results are referred and managed within appropriate timescales as per individual policies completed (This will be audited continually by way of data collection and included in the annual report to the ANSGG). Any deficiencies will be brought to the ANSGG and an action plan developed and monitored.  
| Tool | Systems for ensuring that appropriate tests are undertaken within appropriate timescales. (This will be audited monthly as part of the Key performance indicators which are collected by the ANSC and annually as part of the Annual Report, both of which will be submitted both at Trust board and regional level quarterly, as well as to the ANSGG)  
| Tool | Systems for ensuring that appropriate tests are undertaken when women book late. (This will be audited monthly by the ANSC and reported to the Team Leaders forum)  
| Tool | Process for reporting results to other relevant health care professionals. (This will be audited annually as part of the Annual Report, by the ANSC, which will be submitted both at Trust board and regional level.)  

| Frequency | Frequency for audit is set out as above for each part of the Programme  
| Reporting arrangements | A formal report of the results will be received annually at the ANSGG  
| Reporting arrangements | During the process of the audit if noncompliance or other deficiencies are identified it will be highlighted at the next ANSGG and an action plan agreed  
| Acting on recommendations and Lead(s) | Any deficiencies identified in the annual report will be discussed at the ANSGG and an action plan developed  
| Acting on recommendations and Lead(s) | Action leads will be identified and a timeframe for completion identified  
| Acting on recommendations and Lead(s) | The action plan will be monitored by the ANSGG until all actions are complete  
| Change in practice and lessons to be shared | Required changes to practice will be identified and actioned within time frame identified by ANSG.  
| Change in practice and lessons to be shared | A lead member of the team will be identified to take change forward where appropriate.  
| Change in practice and lessons to be shared | Audit results included in the annual update day for midwives.
4. Equality and Diversity

4.3. This document complies with the Royal Cornwall Hospitals NHS Trust service Equality and Diversity statement which can be found in the 'Equality, Diversity & Human Rights Policy' or the Equality and Diversity website.

4.4. *Equality Impact Assessment*

The Initial Equality Impact Assessment Screening Form is at Appendix 2.
### Appendix 1. Governance Information

<table>
<thead>
<tr>
<th>Document Title</th>
<th>ANTENATAL AND NEWBORN SCREENING - CLINICAL GUIDELINE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Date Issued/Approved:</td>
<td>18th February 2016</td>
</tr>
<tr>
<td>Date Valid From:</td>
<td>1st March 2016</td>
</tr>
<tr>
<td>Date Valid To:</td>
<td>1st March 2019</td>
</tr>
<tr>
<td>Directorate / Department responsible (author/owner):</td>
<td>Jan Clarkson/Jenny Stevenson Antenatal and Newborn Screening Coordinator</td>
</tr>
<tr>
<td>Contact details:</td>
<td>01872 253092</td>
</tr>
</tbody>
</table>

**Brief summary of contents**

This document has been developed to provide clear pathways of care for women undergoing screening in the antenatal and newborn period. All of the screening Programmes offered comply with the standards set by the individual screening Programmes from the National Screening Committee.

**Suggested Keywords:** Screening, sickle, Thalassaemia, newborn, infectious, anomaly, Down's, Edwards' Patau's, Rubella

**Target Audience**

<table>
<thead>
<tr>
<th>RCHT</th>
<th>PCH</th>
<th>CFT</th>
<th>KCCG</th>
</tr>
</thead>
<tbody>
<tr>
<td>✓</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Executive Director responsible for Policy:** Medical Director

**Date revised:** 18th February 2016

**This document replaces (exact title of previous version):** Antenatal and Newborn Screening Policy

**Approval route (names of committees)/consultation:**

- Antenatal and Newborn Screening Governance Group (ANSGG)
- Maternity Guideline Group
- Obs & Gynae Directorate
- Divisional Board for noting

**Divisional Manager confirming approval processes**

Head of Midwifery

**Name and Post Title of additional signatories**

Not required
<table>
<thead>
<tr>
<th>Name and Signature of Divisional/Directorate Governance Lead confirming approval by specialty and divisional management meetings</th>
<th>{Original Copy Signed}</th>
</tr>
</thead>
<tbody>
<tr>
<td>Signature of Executive Director giving approval</td>
<td>{Original Copy Signed}</td>
</tr>
<tr>
<td>Publication Location (refer to Policy on Policies – Approvals and Ratification):</td>
<td>Internet &amp; Intranet ✓ Intranet Only</td>
</tr>
<tr>
<td>Document Library Folder/Sub Folder</td>
<td>Clinical/Midwifery and Obstetrics</td>
</tr>
<tr>
<td>Links to key external standards</td>
<td>National Antenatal and Newborn Screening Standards</td>
</tr>
</tbody>
</table>

**Related Documents:**
- Davies SC Oni L. Sickle Cell Disease Screening Programs: Integration Into Managed Care. Disease Management & Health Outcomes 2001; 9(6):295-304
- Davies SC Cronin E Gill M et al. Screening for sickle cell disease and thalassaemia: a systematic review with supplementary research. Health Technology Assessment 2000; 4(3).
• Olney RS. Preventing morbidity and mortality from sickle cell disease. A public health perspective.
• American Journal of Preventative Medicine 1999; 16(2):116-21
• Vichinsky E Hurst D Earles A et al. Newborn Screening for Sickle Cell Disease: Effect on Mortality.
• Paediatrics 1988; 81:749-55.
• The UK Collaborative Study of Newborn Screening for Medium chain acyl Co-A Dehydrogenase Deficiency (UKCSNS- MCADD): Overview and early findings http://newbornbloodspot.screening.nhs.uk/ mcadd_background [accessed 6th October 2010]

Training Need Identified? Annual training for all midwives via midwives update day

Version Control Table

<table>
<thead>
<tr>
<th>Date</th>
<th>Version No</th>
<th>Summary of Changes</th>
<th>Changes Made by (Name and Job Title)</th>
</tr>
</thead>
<tbody>
<tr>
<td>November 2011</td>
<td>V1.0</td>
<td>Initial document</td>
<td>Maggie Denholm Antenatal Screening Coordinator</td>
</tr>
<tr>
<td>September 2012</td>
<td>V1.1</td>
<td>Change to compliance monitoring only</td>
<td>Maggie Denholm Antenatal Screening Coordinator</td>
</tr>
<tr>
<td>18th February 2016</td>
<td>V1.2</td>
<td>Updated cessation of rubella Introduction of Edwards’ &amp; Patau’s Screening, T13/T18 Change of failsafe process for new-born blood spot screening</td>
<td>Jan Clarkson/Jenny Stevenson Antenatal and Newborn Screening Coordinator</td>
</tr>
</tbody>
</table>

All or part of this document can be released under the Freedom of Information Act 2000
## Appendix 2. Initial Equality Impact Assessment Form

<table>
<thead>
<tr>
<th>Name of Name of the strategy / policy / proposal / service function to be assessed (hereafter referred to as policy)</th>
<th>ANTENATAL AND NEWBORN SCREENING – CLINICAL GUIDELINE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Directorate and service area: Obs and Gynae Directorate</td>
<td>Is this a new or existing Policy? Existing</td>
</tr>
<tr>
<td>Name of individual completing assessment: Elizabeth Anderson</td>
<td>Telephone: 01872-252879</td>
</tr>
</tbody>
</table>

1. Policy Aim*  
Who is the strategy / policy / proposal / service function aimed at?  
To provide information and guidance regarding antenatal and newborn screening pathways

2. Policy Objectives*  
To ensure all women and babies receive screening in a timely manner and enter into care appropriately

3. Policy – intended Outcomes*  
All women and babies are offered screening and all that accept are screened in a timely manner

4. *How will you measure the outcome?  
Production of quarterly key performance indicators for each of the programmes and audit procedures as set out in the document

5. Who is intended to benefit from the policy?  
All pregnant women and babies

6a) Is consultation required with the workforce, equality groups, local interest groups etc. around this policy?  
No

b) If yes, have these *groups been consulted?  
N/A

C). Please list any groups who have been consulted about this procedure.  
N/A

7. The Impact  
Please complete the following table.

<table>
<thead>
<tr>
<th>Equality Strands:</th>
<th>Yes</th>
<th>No</th>
<th>Rationale for Assessment / Existing Evidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age</td>
<td>X</td>
<td>All pregnant women</td>
<td></td>
</tr>
<tr>
<td>Sex (male, female, transgender / gender reassignment)</td>
<td>X</td>
<td>All pregnant women</td>
<td></td>
</tr>
<tr>
<td>Race / Ethnic communities / groups</td>
<td>X</td>
<td>All pregnant women</td>
<td></td>
</tr>
</tbody>
</table>
Disability - Learning disability, physical disability, sensory impairment and mental health problems | X | All pregnant women

Religion / other beliefs | X | All pregnant women

Marriage and civil partnership | X | All pregnant women

Pregnancy and maternity | X | All pregnant women

Sexual Orientation, Bisexual, Gay, heterosexual, Lesbian | X | All pregnant women

You will need to continue to a full Equality Impact Assessment if the following have been highlighted:
- You have ticked “Yes” in any column above and
- No consultation or evidence of there being consultation - this excludes any policies which have been identified as not requiring consultation. or
- Major service redesign or development

8. Please indicate if a full equality analysis is recommended. | Yes | No | X

9. If you are not recommending a Full Impact assessment please explain why.
N/A

Signature of policy developer / lead manager / director | Date of completion and submission
Jan Clarkson and Jenny Stevenson | 18th February 2016

Names and signatures of members carrying out the Screening Assessment | 1. Jan Clarkson
2. Jenny Stevenson

Keep one copy and send a copy to the Human Rights, Equality and Inclusion Lead, c/o Royal Cornwall Hospitals NHS Trust, Human Resources Department, Knowledge Spa, Truro, Cornwall, TR1 3HD

A summary of the results will be published on the Trust’s web site.

Signed: Elizabeth Anderson

Date: 18th February 2016
Appendix 3.
Antenatal & Neonatal Screening Governance Board (ANSGB)

Terms of Reference

NHS Clinical Governance is a “framework through which NHS organisations are accountable for continuously improving the quality of their services and safeguarding high standards of care by creating an environment in which excellence in clinic care will flourish” (Dept of Health 1998).

The Cornwall Antenatal & Neonatal Screening Governance Board (ANSGB) endorse these principles and aims to ensure the development, implementation, delivery and monitoring of an antenatal & neonatal screening programme for Cornwall, in accordance with the guidance and requirements laid down by the NHS Antenatal & Neonatal Screening Programme.

The objective is to ensure a safe, accessible, appropriate, equitable and high-quality screening programme to improve a woman’s choice about her pregnancy and health outcomes for infants in the screened population, whilst complying with protocols and standards laid down by the National Antenatal & Neonatal Screening Programme (NBCSP).

The Cornwall & Isles of Scilly ANSGB is a multi-disciplinary committee chaired by the Director of Public Health (or her/her representative i.e. the Consultant in Public Health with the lead for screening) with representation from those involved in the delivery of the programme in primary and secondary care. Membership will include midwifery, health visiting, medical, laboratory, nursing and administrative staff from the acute trust, CCG, primary care and the South West NHS Antenatal & Neonatal Screening Programme Quality Assurance Centre. It will also include at least one lay representative from PALS or local Maternity Liaison Committee.

Specifically, the role of the Group is to:

1. Share information and decision-making, and to identify early and resolve any problems.
2. Oversee the development and implementation of an effective and efficient quality assurance framework.
3. Monitor, audit and evaluate the programmes quality assurance and performance procedures.
4. Address and resolve any complaints or clinical incidents and put in place mechanisms to reduce the likelihood of their reoccurrence.
5. Establish effective channels of communication to ensure the dissemination of good practice.
6. Co-ordinate and prioritize antenatal & neonatal screening developments and make recommendations to CCG on the use and targeting of resources for inclusion in local delivery plans
7. Agree and implement strategies and action plans to continually improve the local programme.
8. Write an annual report for the Royal Cornwall Hospitals Trust (RHT), the service Commissioner and the Director of Public Health for Cornwall & Isles of Scilly
9. Ensure full participation and collaboration of all stakeholders, including actively participating in the activities of the Regional Antenatal & neonatal Screening Group, the South West Antenatal & Neonatal Screening Quality Assurance Reference Centre and, where appropriate, will seek to influence policy and decision through the nominated representatives.
10. Commission time-limited task-groups as necessary to undertake specific pieces of work and to monitor their progress. Each task group will have terms of reference and a membership agreed by the Group and will report to it.

Frequency of Meetings

The group will meet half yearly.

Minute distribution

The minutes of the meetings will be recorded by the Antenatal & neonatal Screening Unit and circulated to:

Core members

<table>
<thead>
<tr>
<th>Area</th>
<th>Role or representation</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Midwifery</strong></td>
<td>Antenatal &amp; Neonatal Screening Coordinator, Midwifery Matron for community and outpatient services, Maternity risk manager</td>
</tr>
<tr>
<td><strong>Obstetrics</strong></td>
<td>Obstetric Fetal Medicine Lead Consultant</td>
</tr>
<tr>
<td><strong>Paediatrics</strong></td>
<td>Consultant Paediatrician</td>
</tr>
<tr>
<td><strong>Obstetric Ultrasound</strong></td>
<td>Superintendent Sonographer, Lead Screening Sonographer</td>
</tr>
<tr>
<td><strong>Pathology Services</strong></td>
<td>Biochemist (lead for Down’s, Edwards’ and Patau’s Syndrome Screening)</td>
</tr>
<tr>
<td></td>
<td>Haematologist (lead for SC&amp;T)</td>
</tr>
<tr>
<td></td>
<td>Microbiologist (lead for Infectious diseases)</td>
</tr>
<tr>
<td><strong>NHS Public Health England</strong></td>
<td>Screening &amp; Immunisation Team, Public Health Contracts Manager</td>
</tr>
<tr>
<td><strong>Support Services</strong></td>
<td>Child Health Records Department (Newborn Screening)</td>
</tr>
<tr>
<td><strong>Newborn Hearing Screening</strong></td>
<td>Newborn Hearing Screening Programme Manager</td>
</tr>
</tbody>
</table>
**Appendix 4:**

### Linked Sickle Cell and Thalassaemia Screening

**Identify eligible population**

- **High prevalence trust**
  - Take sample and send to laboratory with completed FOQ
  - Laboratory tests sample and reports results as per local arrangements
  - Nothing abnormal detected on screening
    - Offer screening to baby's father
      - Baby's father available and gives consent
        - Laboratory tests sample and reports results as per local arrangements
          - Nothing abnormal detected on screening of baby's father
            - Offer pre-natal diagnostic testing
              - Decline
                - Sample taken and sent to PND laboratory
                  - Result reported to referring physician
                    - Inform parents, community midwife and GP
                      - Carrier or normal result
                        - Continue pregnancy. Include results on newborn bloodspot
                          - Links to newborn SC screening covered by newborn Blood Spot Programme
                        - Baby affected by a major haemoglobin disorder
                          - Provide information and offer choice about continuation of pregnancy
                            - Discontinue with pregnancy
                            - Offer counselling and follow up support
                          - Offer pre-natal diagnostic testing
                            - Affected mother (sickle cell disease) - refer to consultant for clinical & obstetric management

  - Inconclusive (or incomplete) parental results
    - Carrier result
      - Offer pre-natal diagnostic testing
        - Baby's father available and gives consent
          - Laboratory tests sample and reports results as per local arrangements
            - Nothing abnormal detected on screening of baby's father
              - Offer screening to baby's father
                - Baby's father available and gives consent
                  - Laboratory tests sample and reports results as per local arrangements
                    - Affected mother (sickle cell disease) - refer to consultant for clinical & obstetric management

- **Low prevalence trust**
  - Laboratory tests sample as per national policy and reports results as per local arrangements
  - Nothing abnormal detected on screening of baby's father
    - Offer screening to baby's father
      - Baby's father available and gives consent
        - Laboratory tests sample and reports results as per local arrangements
          - Nothing abnormal detected on screening of baby's father
            - Offer pre-natal diagnostic testing
              - Decline
                - Sample taken and sent to PND laboratory
                  - Result reported to referring physician
                    - Inform parents, community midwife and GP
                      - Carrier or normal result
                        - Continue pregnancy. Include results on newborn bloodspot
                          - Links to newborn SC screening covered by newborn Blood Spot Programme
                        - Baby affected by a major haemoglobin disorder
                          - Provide information and offer choice about continuation of pregnancy
                            - Discontinue with pregnancy
                            - Offer counselling and follow up support
                      - Affected mother (sickle cell disease) - refer to consultant for clinical & obstetric management
            - Affected mother (sickle cell disease) - refer to consultant for clinical & obstetric management
Linked Sickle Cell and Thalassaemia Screening

1 Sickle Cell and Thalassaemia screening (linked antenatal / new-born programme)

Scope
The linked screening programme comprises two parts: sickle cell and Thalassaemia screening during pregnancy and sickle cell screening in the new-born period.
Newborn screening for sickle cell disease is offered to all babies in England and is integrated into the standard bloodspot test. Screening identifies approximately 350 babies a year.
The objective of the new-born screening programme is to detect infants at risk of sickle cell disorders within the neonatal period, in order to allow early diagnosis and to improve outcomes through early treatment and care. The analytical methods used at the moment will also detect most cases of β Thalassaemia major. The screening is offered at 5-8 days of age as part of the new-born dried blood spot screening programme. In the case of babies requiring a blood transfusion, a bloodspot sample should be taken prior to the transfusion.

The overall aim of the antenatal screening programme is to offer sickle cell and Thalassaemia screening to all women and couples by 8-10 weeks of pregnancy to allow for the option of early prenatal diagnosis.
All pregnant women in the antenatal population, irrespective of high/low prevalence status are offered screening for Thalassaemia using routine red blood cell indices.
In RCHT, the Family Origin Questionnaire is used to assess the risk of either the woman or her partner being a carrier for sickle cell and other haemoglobin variants.
The partner (baby’s father) of all women who are identified as carriers for either sickle cell or Thalassaemia are offered screening for sickle cell, other haemoglobin variants and Thalassaemia, irrespective of their family origin.
At risk couples are offered counselling and diagnostic tests for the baby.
Further information about screening can be found on the Sickle Cell and Thalassaemia website: sct.screening.nhs.uk.

Prevalence
• Sickle cell & Thalassaemia are among England's most commonly inherited genetic disorders.
• The programme identifies an estimated 350 affected babies, and approximately 33,000 carriers, consisting of babies, mothers and fathers, annually. Data shows that 1 in 29 pregnant women screened are carriers across the whole of England. This figure varies by family origin.
• Healthy people who are not affected by the disorders can be carriers of the sickle cell or Thalassaemia gene without knowing it.
• The highest prevalence of sickle cell is found among Black Caribbean, Black African and Black British communities.
• The highest prevalence of Thalassaemia is found among Cypriot, Pakistani, Indian, Bangladeshi and Chinese communities.

Importance of early screening
The aim of this part of the Programme is to offer reproductive choice, including the offer of termination of pregnancy to those found to have an affected pregnancy.
Early screening in pregnancy, before 8-10 weeks gestation, allows women/couples who are identified as being “at risk” of having a baby with a major haemoglobin disorder, an opportunity to make an informed choice about diagnostic tests and any other procedures required during the early stages of pregnancy.

Family Origin Questionnaire (FOQ)
The screening programme recommends that all areas use a family origin questionnaire (FOQ) as part of routine early antenatal risk assessment.

References
The National Institute for Health and Clinical Excellence (NICE) guidelines at: www.nice.org.uk/
sct.screening.nhs.uk/getdata.php?id=10756
Significant Haemoglobinopathies: guidelines for screening and diagnosis:
British Committee for Standards in Haematology
www.bcshguidelines.com/pdf/significant_final_sept09.pdf

Notes:
Cornwall is a low prevalence area and the FOQ is used to assess risk.

2 Identify eligible population
Quick info:

All pregnant women and couples are eligible for screening. Screening should be offered by 8-10 weeks.

**Testing women in Subsequent pregnancies**

Women need not be tested again in the same or a subsequent pregnancy provided that:

- the original result is unequivocal and well documented
- the red cell indices remain the same
- the patient identification has three or more matching data items, e.g.:
  - name, date of birth and hospital number
  - name, date of birth and address
  - hospital number, date of birth and address (if woman confirms name change)
  - name, date of birth and haemoglobinopathy card.

If a previous result is being used then this fact must be recorded in the woman’s notes for the current pregnancy.

If a woman has been previously tested you need to check if her baby’s father has changed since the previous pregnancy to assess the couple’s risk status in this pregnancy.

**Notes:**

Women are booked wherever possible before 10 weeks gestation.

If a previous result is used, this should be recorded in the hand held notes and clearly identified on the FBC request form if an FOQ is not required.

**Failsafe:**

Offer of screen and date taken will be recorded in the hand held record by CMW.

CMW documents in hand held notes and on stork if previous result is being used and father of the baby is the same as previous pregnancy.

Cohort of women identified by screening co-ordinator from monthly ECLIP report and cross referenced with monthly results report, issued from haematology lab. Missing results followed up by screening co-ordinator.

Any women moving into area having been screened prior to move should have screening results entered onto stork database with date screened, by CMW.

3 Provide information and take consent

Quick info:

Consent and reference to the provision of written information should be recorded in the maternity records.

**Notes:**

Written information regarding the screen is in the form of “Screening tests for you and your baby” which is given out before the booking appointment.

Information in accessible format via the National Screening Committee website (Information in other languages) may be printed and sent to women as required.

Information in other formats may be requested via screening co-ordinator.

**Failsafe:**

Any missing results followed up with CMW and team leader to ensure screen has been offered and performed.

Any samples with information missing on the FOQ, screening co-ordinator contacted from the lab and information obtained from stork and CMW.

4 High prevalence Trust

Quick info:
High prevalence trusts are those where Sickle Cell Diseases are estimated to affect over 1.5 pregnancies per 10,000 births, based on estimates from Census 2001 categories.

5 Low prevalence Trust

Quick info:

Low prevalence trusts are those where Sickle Cell Diseases are estimated to affect less than 1.5 pregnancies per 10,000 births. In low prevalence trusts, a family origin questionnaire is used to assess the risk of either the woman or the baby’s father of being a carrier for sickle cell and other haemoglobin variants. The Family Origin Questionnaire should be completed and sent to the laboratory with the sample. Obtaining a supply of FOQ forms: You can obtain a supply of Family Origin Questionnaire forms from Prolog. Telephone 0300 123 1002 Code ANSPFQ 07/07

6 Take sample and send to laboratory with completed FOQ

Quick info:

Midwives need to include information on family origins for all women to accompany the Full Blood Count (FBC) and haemoglobinopathy screen sample; complete all demographic information; and sign the FOQ form. Screening for sickle cell and thalassaemia should still be offered even if information about the baby’s father is unavailable.

• If woman declines screening, explore the reasons for this and document on the FOQ form.

The completion of the FOQ in all areas is essential as part of the screening for sickle cell and thalassaemia. In low prevalence trusts, if a sample is received in the laboratory without a completed FOQ there is a risk that the woman will not be screened for sickle cell and thalassaemia and a risk of missing an affected pregnancy resulting in the birth of an affected baby. There should be local policy in place to ensure that FOQ information on woman whose samples are received without an FOQ are obtained.

Notes:

• The FBC sample is sent to the lab in a plastic specimen bag with a completed FOQ.

7 Laboratory tests sample as per national policy and reports results as per local arrangements

Quick info:

All antenatal screening laboratories must work towards incorporating the screening status codes on their laboratory reports. This will assist when interfacing with the newborn screening laboratory and child health record systems to facilitate linking maternal, paternal and newborn screening results, and in future status codes will be used to facilitate communication of results from antenatal laboratories to primary care. Status codes are not meant to replace the full laboratory report, and are not designed to be used in detailed counselling of women and their partners. The outcome code is one code with different values and is designed with the ability to change in order to reflect the stage of each woman/couple’s journey along the screening pathway. It is anticipated that this code will give a picture of the couple’s outcome following screening and what the possibilities for the baby’s result are. The codes may change during the current pregnancy and could also be different in each pregnancy. Newborn status codes are led by the NHS Newborn Blood Spot Programme

Notes:

All results issued onto the Maxims system and recorded in hand held records at next appointment with community midwife.

Failsafe:
Antenatal samples cross referenced with booking information monthly.
Any samples with information missing on the FOQ, screening co-ordinator contacted from the lab and information obtained from stork and CMW.

8 Nothing abnormal detected on screening

Notes:
Normal result recorded in handheld notes at next appointment by community midwife.

9 Inconclusive (or incomplete) parental results

Quick info:
If the baby’s father is unavailable for testing or his haemoglobinopathy status is unknown, then a risk assessment should be done.
The Programme supports woman who are carriers, being offered prenatal diagnosis in this situation if she requests it.
Prenatal diagnosis for some genotypes of sickle cell disease can be undertaken without the baby’s father’s DNA.
Similarly, prenatal diagnosis for β thalassaemia can be undertaken without the baby’s father’s DNA, although the diagnosis cannot be given with as high a degree of certainty if samples from both parents are not known or tested.

Notes:
Pre-natal diagnosis should be offered to carrier women if father’s results are not available or risk assessment of inconclusive result indicates potential at risk couple.

Failsafe:
Any inconclusive or incomplete parental results will be informed to screening co-ordinator. Risk assessment will be performed and Prenatal Diagnosis (PND) offered if required. Discussion will be documented in hand held record.

10 Carrier result

Quick info:
A healthcare professional informs parents of carrier results in accordance with Programme Centre guidelines and local pathway.
The maternity unit should notify the new-born laboratory of carrier status.
The programme recommends the maternity units keep a log of and inform the new-born laboratory of all women who are carriers and affected, including PND results, and have systems in place to check the screening result of babies born to screen positive woman.

Notes:
Screening co-ordinator informed of carrier results and informs woman.

Failsafe:
Result details recorded on screening database for robust audit. This will include timing of appointments and verification of alert form sent.

12 Offer screening to baby’s father

Quick info:
The national programme recommends that maternity units work towards offering screening to the baby’s father before 11 weeks of pregnancy to identify ‘at risk couples’.

Notes:
Verbal consent to discuss result and need for partner sample will be gained by ANNCS. Appt arranged with couple for discussion and taking of sample by ANNCS at first available opportunity.
Written information regarding carrier status given to couple from APoGI website:
ANTENATAL AND NEWBORN SCREENING - CLINICAL GUIDELINE
www.chime.ucl.ac.uk/APoGI/

**Failsafe:**
Newborn Lab informed of carrier status via alert form from screening co-ordinator, even if partner status not known. Copy of alert form printed and filed with screening co-ordinator.
All results documented in hand held record following telephone contact and written confirmation of results, sent by screening co-ordinator.
Result details recorded on screening database for robust audit. This will include timing of appointments and verification of alert form sent.

13 Baby’s father available and gives consent

**Notes:**
Sample sent with FBC form completed with additional yellow partner sample alert sticker.

**Failsafe:**
Lab informed via e-mail to expect sample.

14 Baby’s father not available or declines consent (with conclusive maternal result)

**Quick info:**
See the information in the 'Inconclusive (or incomplete) parental results' box.

**Failsafe:**
Any inconclusive or incomplete parental results will be informed to screening co-ordinator. Risk assessment will be performed and PND offered if required. Discussion will be documented in hand held record.

15 Laboratory tests sample and reports results as per local arrangements

**Notes:**
If partner sample also carrier status, lab contacts screening co-ordinator via phone and e-mail.

16 Nothing abnormal detected on screening of baby’s father

**Quick info:**
Offer information and advice about the possibility of the child being a carrier based on the mother’s carrier result.

17 Confirmed carrier or affected result in both parents - refer at risk couple

**Quick info:**
Parents are referred for counselling by a Pegasus-trained health professional.
The Programme commissioned the Pegasus network to develop and deliver genetics and diversity training using sickle cell and Thalassaemia as a model.

**Notes:**
Couple seen by PEGASUS trained ANNSC or Consultant Haematologist and referred for pre-natal diagnosis if requested.
18 Offer pre-natal diagnostic testing

Quick info:

**Prenatal Diagnosis Test**

Always contact the PND laboratory beforehand to make arrangements for sending the sample off.

Fresh blood samples from both parents are to be sent to the DNA referral laboratory with appropriate consent for molecular analysis along with the Prenatal Diagnosis sample.

**Amniocentesis**

- Performed trans abdominally under continuous ultrasound guidance
- Not usually recommended before 15 weeks of pregnancy. It can also be performed later in pregnancy
- The risk of causing miscarriage is about 1%

**Chorionic Villus Sampling**

- Usually performed trans abdominally under continuous ultrasound guidance
- Can be undertaken between 10-13 weeks of pregnancy
- The total miscarriage rate following CVS is about 1%

**Notes:**

Laboratory used for PND is Bristol.

Miscarriage rate for CVS and Amnio both 1:100 (South West Data)

Sample taken in Fetal Medicine Department, and recorded in hand held notes.

Newborn lab informed by alert form as before

**Failsafe:**

Couples status, offer, acceptance/decline of PND, continuing of high risk pregnancy will be documented in the hand held record by health professional counselling/performing PND.

Newborn blood spot will have status recorded on card by health professional taking the sample.

19 Decline

Quick info:

Respect decision.

The programme recommends that maternity units have a robust system for recording information on at-risk couples declining PND testing, for example recording in maternity notes, on the bloodspot card and on alert forms to be sent to the newborn laboratory.

There should be a named person in every maternity unit with the responsibility to ensure that newborn screening laboratories are informed of carrier women whose pregnancy is ongoing.

**Notes:**

Result recorded in hand held notes, on Newborn Bloodspot card and alert form sent to Newborn lab.

The ANNSC is responsible for sending alert forms to the lab.

20 Accept - contact PND laboratory to make arrangements for analysis

Quick info:

For contact details of PND laboratories, see the information in the 'Offer pre-natal diagnostic testing' box.

22 Result reported to referring clinician

Quick info:

Maternity units to complete the short-term and long-term PND outcome form.

23 Inform parents, community midwife and GP

Quick info:

The responsibility for reporting results is as set out below:
Notes:
Results are issued to parent by ANNSC and referral to local Obstetric team is made.
ANNSC informs Community midwife.
Referral to Neonatologist made by Consultant Obstetrician.
GP informed from Consultant Obstetrician

24 Carrier or normal result

Quick info:
A designated healthcare professional informs parents of PND result in accordance with local policy and in a timely manner as per programme guidelines i.e. within 4 days of having the PND procedure.

25 Baby affected by a major haemoglobin disorder

Quick info:
Reporting of results to mother: see the information in the 'Carrier or normal result' box.
Counselling: Parents to be referred for counselling and follow up by Pegasus trained healthcare professionals.
Specialist Sickle Cell and Thalassaemia centres and Regional Genetic centres provide counselling services, information and advice for families with or 'at risk' of genetic conditions.
Alert form to be sent to Newborn Screening laboratory.

Notes:
Couple referred to Regional Genetics Centre (Exeter) and haematologist as per RCHT policy.
Alert form sent by screening co-ordinator.

27 Continue pregnancy, include results on blood spot card

Quick info:
Known High-Risk pregnancies
Local policies should be in place a liquid capillary blood specimen (not cord blood) to be taken from the baby for analysis soon after birth, if that is the parents' choice.
This is particularly important in cases identified antenatally that have declined prenatal diagnosis and decided to continue with the pregnancy.
This blood specimen should be sent to a specialist laboratory, which has expertise in haemoglobinopathy analysis in the newborn period. The screening laboratory will undertake the routine screen as normal. The fact that such a specimen has been taken should be noted by the midwife on the newborn screening bloodspot card.
Counselling and Follow-up
Parents to be offered advice and counselling about future pregnancies. See the information in the 'Baby affected by a major haemoglobin disorder’ box.
Impact to Child
Child will be referred when born to paediatrician / haematologist / specialist counsellor as per local policy and programme standards
i.e. registered with designated clinic by 8 weeks of age and attending local clinic by 3 months of age.
Maternity units should notify newborn laboratories of women continuing affected pregnancies and parental results to be included on newborn blood spot request card.

Failsafe:
All alert forms sent and copy retained by screening co-ordinator
Newborn screening lab notifies screening co-ordinator of any affected babies and antenatal screening history cross checked and lab informed for quality assurance checks.

30 Links to newborn sickle cell screening covered by Newborn Blood Spot Programme

Quick info:
ANTENATAL AND NEWBORN SCREENING - CLINICAL GUIDELINE
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For parents, the link between antenatal and new-born screening is obvious: the result of an antenatal genetic test will be related to the results of a new-born baby’s genetic test for the same condition, so the linked programme provides a natural failsafe check between the mother and baby result.

The antenatal report given to women should contain all antenatal screening results for communication to primary care and allow linkage to new-born screening results where relevant.

There should be systems in place to inform new-born screening laboratories of all antenatal screening and diagnostic results and there should be a named person in every maternity unit with responsibility to ensure that newborn screening laboratories are informed of carrier women whose pregnancy is ongoing.

Notes:
All antenatal screening results given to women as described previously
ANNSC is responsible for informing new-born lab of all carrier results and diagnostic results.
Known HIV and Hep B status

Offer of screening

Accept

Re-offer the test

Decline

Take sample and request screening test

Receipt of screening test result

Unacceptable samples and inconclusive test results

Negative results

Confidentiality

Managing positive screening results – HIV Hep B & Syphilis

Rubella susceptibility – offer of postnatal MMR for all women booked before 1st April 2016

Syphilis

HIV

Hepatitis B

Go to management of positive cases identified through screening

Rubella susceptible result until 1st April 2016

Hepatitis B screening policy

HIV screening policy

Syphilis screening policy

Rubella screening before 1st April 2016

HIV screening policy

Syphilis screening policy

Rubella screening before 1st April 2016

Hepatitis B screening policy

Note in text for Cornwall pathway

Secondary Care

Failsafe processes
1 Infectious Diseases in Pregnancy Screening Programme

Quick info:

The Infectious Diseases in Pregnancy Screening (IDPS) Programme is responsible for ensuring that women with hepatitis B, HIV and syphilis infection are identified early in pregnancy. The Programme is an essential component of strategies to prevent mother-to-child transmission of hepatitis B, HIV and syphilis.

Cessation of Rubella screening

From 1st April 2016 the offer of rubella screening in pregnancy will no longer be made. Any woman who was booked before 1st April 2016 and is rubella susceptible will continue to be offered postnatal MMR vaccination. For woman who book but decline rubella screening prior to 1st April 2016 will be reoffered screening as per the current pathway. All women will be informed that if they develop a rash during pregnancy they must contact their community midwife or their GP.

2 Hepatitis B screening policy

Quick info:

All pregnant women should be offered screening in each pregnancy regardless of immunisation history, unless they are already known to be hepatitis B positive. Hepatitis B screening should be recommended alongside the other antenatal booking blood tests. The aim of antenatal screening is to contribute to the reduction of perinatal hepatitis B infection.

The objectives of the screening programme are to:
• ensure that all hepatitis B positive women are identified,
• refer all hepatitis b positive women for assessment and management by an appropriate specialist (e.g. a Hepatologist / gastroenterologist / infectious diseases specialist) within 6 weeks of the screening test result being received by maternity services,
• ensure that the infant vaccination schedule is offered for their babies, that the first dose is administered within 24 hours of delivery and that arrangements for completion of the schedule are initiated.

3 HIV screening policy

Quick info:

All pregnant women should be offered HIV screening in each pregnancy, unless they are already known to be HIV positive. HIV screening should be recommended alongside the other antenatal booking blood tests. The aim of antenatal screening is to contribute to the reduction of paediatric HIV infection.

The objectives of the screening programme are to:
• identify all HIV positive women and those with positive test results,
• ensure the rapid referral of all HIV positive women for assessment and management within a multi-disciplinary team.

4 Syphilis screening policy

Quick info:

All pregnant women should be offered screening for syphilis early in each pregnancy regardless of the results of syphilis screening tests in previous pregnancies. Screening should be recommended to every pregnant woman alongside the other antenatal booking blood tests.

The aim of antenatal screening is to contribute to the reduction of congenital syphilis infection.

The objectives of the screening programme are to:
• identify all women with positive syphilis screening test results early in pregnancy,
• ensure their rapid assessment by an appropriate specialist, e.g. a Genitourinary Medicine (GUM) within a multi-disciplinary environment.

6 Offer of screening

Quick info:

Informed consent for screening must be given before a specimen is taken and tests requested. The woman should have seen the written information, Screening tests for you and your baby, or have had access to it in a format appropriate to their requirements.

In order for the woman to make an informed choice, the following points should be discussed:
• the three infections, their routes of transmission and the implications of a positive test,
- the benefits, to both mother and baby, to be gained from the identification and management of those with positive results,
- the results procedure, including the feedback of results and the possibility of a false negative or false positive result,
- all pregnant women should be advised that if they develop, or are exposed to, a rash during the pregnancy they should seek professional advice.

The process for informing the woman of the test results, both positive and negative, should be explained. For information on women who book late in pregnancy and women who arrive in labour unbooked for care refer directly to the Programme Standards.

**Notes:**
The offer of screening will be recorded in the hand held records along with the date offered and any screen declined recorded for each part of the screen, by the community midwife. Any previous positive screening results are to be recorded here and ANSC to be informed.

**Failsafe:**
Notes checked at next contact with CM and results recorded in hand held notes.

### 7 Decline

**Quick info:**
If the offer of screening for any infections is declined, the woman should be informed that it will be reoffered at a later stage of pregnancy. Advice on avoiding infection should be provided. Information should be provided on the availability of testing on request at any point during pregnancy should the woman consider herself to be at risk.

**Notes:**
If a woman declines part or all of the screen a "decline" sticker should be applied to the stork sheet and the stork screen 'declined' indicated, for audit and re-offer purposes. The tests accepted or declined must be clearly identified on the laboratory request form.

### 9 Known HIV and Hepatitis B status

**Quick info:**
Where a prior positive diagnosis of HIV or hepatitis B is documented and known to the healthcare professional this should be recorded and arrangements for prompt clinical evaluation made.

It is essential that the current status of the infection is promptly assessed by an appropriate specialist to evaluate its implications for the care of the woman, the onward management of the pregnancy and care of the baby. Discussion with the woman should cover the same issues as those at appointments to discuss positive screening test results. In the case of hepatitis B [and] the midwife should make arrangements for vaccination of the baby. The requirement for HBIG should be assessed by an appropriate specialist.

**Notes:**
Known HIV and Hep B status must be conveyed to the screening co-ordinator who will arrange follow up as per individual pathways. This must also be clearly identified on the laboratory request form.

### 10 Re-offer the test

**Quick info:**
Screening should be reoffered to all women who declined the initial offer, ideally, by 20 weeks gestation.

This standard refers to routine screening. It should not discourage health professionals offering tests to any woman at continuing risk of infection. Neither should it prevent any woman from requesting a subsequent test at other points in the pregnancy.

**Notes:**
A re-offer letter will be sent to the woman from the ANNSC, with a copy of the "Screening for Infectious Diseases in pregnancy" leaflet, ideally by 20 weeks gestation. The woman should be offered the opportunity to discuss testing with the ANNSC.

### 11 Take samples and request screening tests

**Quick info:**
If the offer of screening, for one or more condition, is accepted the laboratory should receive a fit for purpose antenatal specimen within one working day of the sample being taken. The specimen should be clearly identified as an antenatal screening sample. It is necessary to indicate which tests are being requested and, if relevant, which have been declined. All mandatory fields on the request form should be completed. The requester’s identity should be clear. Local protocols will determine the arrangements for collection, storage and transport of specimens.

Notes:
Blood samples will be taken at booking, marked clearly as an antenatal sample, with date and time. Accurate and full completion of blood request form by CMW, including whether screening is accepted, declined or known to be HIV/hepatitis b positive. Sample will be dispatched to the lab within one working day of the sample being taken.

Failsafe:
All samples taken in the previous month notified to ANNSC and cross referenced with bookings. Any missing results identified and CMW contacted to ensure sample taken. Samples sent to lab by courier service.

If the specimen is taken later in pregnancy following an initial decline, or if the woman has booked late, the specimen must be marked as URGENT.

12 Receipt of screening test results

Quick info:
All confirmed screening test results should be received by maternity services within 10 working days of the screening specimen being taken. Processes should be in place to identify and follow up results that have not been received within 10 working days of the initial test request being made. Arrangements for rapid reporting of positive results, e.g. by telephone, should be made with the laboratory. Recommended laboratory reports for the four conditions can be found in the handbook for laboratories.

Notes:
It is the responsibility of the midwife taking the sample to check results available within 10 days. For women moving into the area that have had screening out of county, a result for each of the four screens should be entered onto the Stork data base by the community midwife at booking if she has seen a copy of the results. If no copy available it should be recommended to the woman to have the test repeated.

Failsafe:
Screening co-ordinator checks for any missing results and e-mails midwife requesting information as to why specimen has not been taken. If a sample cannot be processed due to missing information or poor sample, the lab will notify the ANSC who will ensure midwife is aware and repeat performed.

13 Unacceptable samples and inconclusive test results

Quick info:
Laboratory requests for repeat specimens, for example when the first specimen is unacceptable or the screening test result is inconclusive, should be sent to the laboratory within 10 working days of the request being received by the maternity unit. A process should be in place to ensure that laboratory requests for repeat samples are identified and followed up. This might best be achieved collaboratively e.g. between the maternity unit screening coordinator and the lead for screening within the laboratory.

Failsafe:
With unacceptable or inconclusive results, the lab will contact the screening co-ordinator who will arrange for a follow up sample.

14 Rubella susceptible results (women booking before 1st April 2016)

From the 1st April 2016 women will no longer be offered screening for Rubella Susceptibility.

Quick info:
Women who are susceptible to rubella infection should be informed about the test results at their next antenatal visit.

Notes:
On a monthly basis the ANSC sends a letter and information sheet to any woman who is rubella susceptible, advising post natal MMR vaccination. A letter is also sent to the community midwife and GP.

Failsafe:
MMR vaccination documented in hand held notes and on Stork database
Audit proforma completed when MMR given or declined and sent back to ANSC for completion of database.
Any woman who has miscarried will have a letter sent to her GP by ANSC for completion of MMR vaccination.

15 Managing positive screening results - HIV, Hep B & syphilis
Quick info:
Women with positive screening test results should be contacted and advised about the results, at an appointment made for that purpose, within 10 working days of the result being made available to maternity services. The purpose of the appointment is to discuss the screening test result and to arrange for referral to a relevant specialist service for clinical assessment.

Notes and failsafe:
See individual pathways.

16 Negative results
Quick info:
Negative screening test results should be reported back to women before or at the next antenatal visit, according to local protocol. Advice about risk of acquisition and avoidance of infection should be provided to women receiving negative test results. Information should also be provided on the availability of testing on request should the woman consider herself to be at risk at any point in the pregnancy.

Notes:
Negative results to be recorded in the handheld record at the next antenatal check by the Community Midwife

Failsafe:
Audit of hand held notes completed by Supervisor of Midwives audit on a monthly basis and fed back to staff via team leaders.

17 Confidentiality
Quick info:
Positive screening test results should be made available to the healthcare professionals responsible for the care of the woman and her baby without compromising the woman’s right to confidentiality. Information relevant to the care of the mother and baby should be documented in the woman’s central hospital record. There should be 24 hour access for those professionals who need to be aware of the requirements of the woman and her baby during the antenatal, intrapartum and postnatal period. There should be strict access controls to protect patient confidentiality. Positive results should only be recorded in non-secure sites, for example handheld records, following discussion and with the consent of the woman. Health professionals who are engaged with the woman’s care in contexts other than antenatal care should be informed of positive results. Positive results should be transferred and recorded securely, for example by direct communication to the GP for inclusion in their records. Women should be informed that this action has been undertaken.

18 Rubella Susceptibility - offer of postnatal MMR(for women booking before 1st April 2016)
Quick info:
At the next antenatal visit consent for postnatal MMR vaccination should be sought from all women identified as susceptible to rubella infection. Discussion at the next antenatal visit should include:
• the benefits of vaccination for future pregnancies
• that breast feeding is not contraindicated following vaccination
• that conception should be avoided for at least one month after vaccination.
The offer, acceptance or decline should be recorded in the maternity notes. If the offer is accepted, arrangements should be made to ensure the vaccine is available before discharge from maternity services. If the offer is declined, it should be reoffered again in the postnatal period.

Notes:
A letter and information leaflet to be sent to woman from screening co-ordinator.
GP informed by letter from screening co-ordinator.
Any women who have miscarried will be informed of susceptibility by screening co-ordinator and GP informed in order to offer MMR.
See Rubella Susceptible* pathway
19 Syphilis
Quick info:
Arrangements should be made for urgent referral for assessment by an appropriate specialist (e.g. a GUM physician) in accordance with the BASHH guideline.
The following should be discussed with the woman:
• need for further assessment to provide a diagnostic evaluation, confirm identity and evaluate maternal treatment needs
• the significance of syphilis infection for maternal health, the pregnancy and the baby's health,
• the potential benefits of multi-disciplinary management for the pregnancy, the woman’s health and that of the baby
• practical arrangements for further assessment e.g. date options appointments
Robust mechanisms are required to facilitate rapid assessment of women with screen positive results within a multi-disciplinary environment.
The urgency to complete the assessment is because:
• not all positive screening test results will be confirmed as a syphilis diagnosis or as an infection requiring treatment,
• treatment, when indicated, needs to be instituted as early as possible to avoid adverse outcomes of pregnancy.
Non attendance for assessment should be reviewed by a multidisciplinary team and a management / action plan agreed.

Notes:
Positive results relayed to screening co-ordinator from lab lead by telephone.
Screening co-ordinator generates referral to GUM and obstetric consultant with copy to Neonatologist.
Obstetrician to ensure referral made to neonatologist where letter will be generated to delivery suite Alert File.
Community midwife informed of clinic dates and informs woman face to face, and takes confirmatory sample.

20 HIV
Quick info:
Arrangements should be made for urgent referral to the multi-disciplinary team (MDT) responsible for co-ordinating the woman’s HIV care in accordance with the BHIVA Guidelines.
The following should be discussed with the woman:
• the significance of HIV infection for her own health, the pregnancy and the baby’s health
• need to attend specialist appointment for further tests to confirm identity and evaluate maternal treatment needs
• the potential benefits of multi-disciplinary management for the pregnancy, the woman’s health and that of the baby
• practical arrangements for further assessment e.g. date options for appointments with the multi-disciplinary team
Robust mechanisms are required to ensure that HIV positive women are managed within a multidisciplinary environment.
Non attendance at the specialist appointment should be reviewed by the multidisciplinary team and a management / action plan agreed.

Notes:
Positive results relayed to screening co-ordinator from lab lead by telephone.
Screening co-ordinator generates referral to GUM, HIV specialist nurse, and obstetric consultant with copy to neonatologist.
Obstetrician to ensure referral made to neonatologist where letter will be generated to delivery suite Alert File.
Community midwife informed of clinic dates and informs woman face to face, and takes confirmatory sample.

21 Hepatitis B
Quick info:
Management of the mother
Arrangements should be made for an appointment with an appropriate specialist (e.g. a Hepatologist, gastroenterologist or infectious diseases specialist) within 6 weeks of the screening test result being issued to maternity services. Women booking late, after 24 weeks, for antenatal care should be referred immediately for a clinical evaluation immediately.
The following should be discussed with the woman:
• the significance of hepatitis B infection for her own health, the pregnancy and the baby’s health
• the need for further tests for confirmation of identity and evaluation of maternal management requirements
• the potential benefits of specialist management for the pregnancy, the woman’s health and that of the baby
• practical arrangements for further assessment e.g. date options for appointments with specialist services
Robust mechanisms are required to ensure that hepatitis B positive women are managed within a multidisciplinary environment.
The composition of the multi-disciplinary team may vary locally.
Non attendance at the specialist appointment should be reviewed within a multidisciplinary framework and a management / action plan developed.

Infant vaccination
Maternal consent for the baby to be vaccinated in accordance with the Green Book schedule should be sought and action taken to facilitate this.
The following should be discussed with the woman:
• the benefits to the baby of completion of the infant vaccination schedule
• the importance of specialist assessment in determining the maternal infection status, risk of transmission and the requirement for HBIG. Arrangements should be made to prescribe, order and store the vaccine (+/- HBIG as required) in advance of the estimated delivery date.
The following should be informed of all confirmed positive screening results:
• the specialist responsible for clinical assessment and management of the woman,

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• the health care professional responsible for arranging testing of other older siblings, partner and other household contacts if different from the above specialist,
• the GP, health visitor and/or practice nurse,
• Health Protection Unit (HPU). This is to notify the HPA of the screening result and the details of the clinician responsible for requesting diagnostic laboratory testing,
• the PCT Immunisation Lead should be informed from an early stage

Notes:
Positive results relayed to screening co-ordinator from lab lead by telephone.
Screening co-ordinator generates referral to Hepatology nurse specialist, pharmacy lead and obstetric consultant and copy to Neonatologist.
Obstetrician to ensure referral made to neonatologist where letter will be generated to delivery suit Alert File.
Community midwife informed of clinic dates and informs woman face to face, and takes confirmatory sample

Failsafe:
First dose of vaccine given by Neonatal Unit following referral and letter in alert file.
HPA informed of all cases of Hep B who currently follow on all babies to ensure vaccination schedule is completed.
(Pathway due to change 2012)

Evidence summary for Infectious Diseases in Pregnancy Screening

This pathway is underpinned by the Programme Standards and the Handbook for Laboratories of the NHS Infectious Diseases in Pregnancy Screening (IDPS) Programme. The standards and handbook were developed with reference to work undertaken by multidisciplinary task groups established by the IDPS Programme in early 2008 and a stakeholder consultation exercise which ran between October 2009 and January 2010. The standards were also informed by a national programme mapping exercise and a survey of laboratories. The standards provide maternity services with the information necessary to manage the organisational challenges of the screening process and the service interfaces to support implementation of the relevant guidance for follow on care of mother and baby.
Appendix 6:

DOWN'S, EDWARDS AND PATAU'S SYNDROME SCREENING

- Offer choice of either full screening, T21 only, T13/18 only or dating only
- Screening Declined
- Screening Accepted
- Follow up at delivery

Initial Screening tests

- High risk result – offer diagnostic procedure
- Low risk result
- Follow up at delivery

Amniocentesis / Chorionic Villus Sampling or prenatal non invasive screening (non NHS)

- Procedure completed successfully
- Miscarriage
- Provide support to parents

Diagnosis of chromosomal abnormality

- Discuss options with parents
- Consider referral to obstetrician or paediatrician

PCR normal for Downs, Edwards and Patau's syndrome

- Follow up at delivery

Note in text for Cornwall pathway
Secondary Care
Failsafe processes
Down’s, Edwards and Patau’s Syndrome – screening

Quick info:

Scope:
- Down’s, Edwards and Patau’s Syndrome screening during pregnancy
- The policy is based on the current national screening committee recommendations:
  - care for children and adults with Down’s syndrome
  - management of miscarriage as a result of diagnostic testing, or termination following a positive test result

Definition:
Down’s syndrome occurs when a baby has 47 instead of 46 chromosomes; an extra copy of chromosome 21 is present because of an error in cell division
- some people will have associated health problems, including:
  - heart problems (50%)
  - reduced hearing (50%)
  - visual impairment
- long term health problems include:
  - Alzheimer’s disease
  - leukaemia
- life expectancy of a person with Down’s is 50-55 years
- the risk of having a baby with Down’s syndrome increases with maternal age:
  - 1/1500 at age 20 years
  - 1/900 at age 30 years
  - 1/100 at age 40 years

Edwards syndrome (T18) and Patau’s syndrome (T13) occurs when a baby has 47 instead of 46 chromosomes; an extra copy of chromosome 18 and chromosome 13 is present because of an error in cell division
- Sadly most babies with Edwards or Patau’s syndrome will die before they are born, be stillborn or die shortly after birth. Survival beyond the neonatal period is rare with the exception of the mosaic pattern.
- Abnormalities may include: major brain abnormalities
- Babies affected by T18 can have heart problems, unusual head and facial features, growth problems and be unable to stand or walk. This condition affects about 3 in every 10,000 births
- Babies affected by T13 can have heart problems, a cleft lip and palate, growth problems, poorly formed eyes and ears, problem with their kidneys and be unable to stand or walk. This condition affects about 2 in every 10,000 births

References:
Department of Health. NHS England fetal anomaly screening programmes for Down’s, edwards’ and patau’s syndromes December 2014
NHS Antenatal and Newborn Screening Programmes. Screening tests for you and your baby. UK National Screening Committee; October 2014
Down’s, Edwards’ and Patau’s syndromes, screening tests during your pregnancy

1. Offer Down’s (T21) and/or Edwards(T18) and Patau’s (T13) syndrome screening

Quick info:

• all pregnant women should be offered 1st trimester screening. They can chose to have screening for T12, T18 and T13, or T21 only, or T13/18 only or dating scan only
• explain stages of screening process and inform the woman of the risks, benefits and limitations of the screening test
• ensure that the woman understands that screening cannot indicate whether her baby has a chromosomal abnormality or not, it can only estimate whether she is at low or high risk
• explain that if the screening test result is high risk, then diagnostic testing such as amniocentesis or Chorionic Villus sampling (CVS) carries a risk of miscarriage; either way there is a chance she could lose an affected or non-affected baby. The woman should also be informed about prenatal non invasive testing, that is not NHS funded
• the woman should be given time to consider the information given to her before making a decision as it’s important that she makes a choice which is right for her pregnancy

References:
NHS Antenatal and Newborn Screening Programmes. Screening tests for you and your baby. UK National Screening Committee; October 2014

ANTENATAL AND NEWBORN SCREENING - CLINICAL GUIDELINE
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2. Initial screening tests

Quick info:
- All pregnant women should be offered screening for Down’s Syndrome with a test which provides a detection rate above 85% and screen positive result of 1.9-2.4%, and for Edwards and Patau’s syndrome a detection rate >80% and screen positive of 1.9-2.4%
- These performance measures should be age standardised and based on a cut off of: 1:150 at term (1st trimester)
  1:150 at term (2nd trimester)

Notes:
- All women are offered screening for Downs, Edward’s and Patau’s Syndrome at the booking visit.
- The first trimester combined screening is offered between 11+2 weeks and 14+1 weeks gestation
- Second Trimester quadruple test is offered from 14+2 to 20+0 weeks gestation and if accepted performed by the community midwife

Failsafe:
The decision is recorded on the Stork Sheets which are sent in to Fetal Medicine where women are then either booked for dating scan or Downs, Edward’s or Patau’s Syndrome screening.
If the request is unclear, Screening Administrator checks immediately with CMW responsible for the woman.
The Midwife Sonographer will check that all pre-test information has been given and understood before performing the screen.
Quad test is taken in the Community, CMW rings laboratory to inform them to expect a sample.

3. High risk result - offer diagnostic procedure

Quick info:
- A diagnostic test is offered to the woman if her screening test result falls from: 1 in 2 up to 1 in 150 (1st trimester screening) 1 in 2 up to 1 in 150 (2nd trimester screening)
- The women’s screening test result should be explained to her by an appropriately trained healthcare professional
- The healthcare professional should present the screening test result to the woman in a way in which the woman can understand the numbers being explained e.g. 1 in 150 means 1 chance in 150 chances that the baby has Down’s syndrome. 149 chances out of 150 chances that the baby is unaffected by the condition.
- The healthcare professional should explain how the diagnostic procedure (Amniocentesis or CVS) is undertaken. The risks, benefits and limitations should be discussed and the non NHS funded prenatal non-invasive testing

Notes:
All Increased risk results are telephoned out to woman by screening co-ordinator and appointment made to see her at earliest opportunity to suit the woman.
Counselling may be offered at a location suitable to the woman if she has difficulty in coming into RCHT.
The CMW will be informed of the result by the Fetal Medicine Department in order to give support out of hours.
If screening co-ordinator not available, results rung out to CMW via Fetal Medicine and appointment made for counselling at first available Consultant Clinic for the area in which the woman lives.

Failsafe:
High risk result for first and second trimester screening telephoned to screening co-ordinator followed by a email to the generic screening account at rch-tr.Screening@nhs.net.
Information entered onto screening database for audit and monitoring purposes.

4. Low risk result
Quick info:

- After screening tests, if the risk is less than 1 in 150 (1st trimester) or 1 in 150 (2nd trimester) provide information and ensure that the woman understands that although the test result is reassuring, because it is a screening test, it does not definitely exclude Down’s, Edwards and Patau’s Syndrome.

Notes:

- All low risk results sent to women via letter to be received within 1 week.
- Result to be recorded in hand held record by CMW at next appointment.

Failsafe:

- If results not received or unavailable at 15 week check, CMW will ring FMU for confirmation of result.

5. Follow-up at delivery

Quick info:

- The Newborn Physical Infant Examination (NIPE) should be carried out within 72 hours of birth, and again 6 to 8 weeks later by an appropriately trained healthcare professional.

Notes:

- Outcome of pregnancy recorded on Viewpoint database.

Failsafe:

- All cases of Down’s Syndrome, Edwards’ and Patau’s informed to laboratory and Screening Co-ordinator.

6. Amniocentesis/ Chorionic Villus sampling

Amniocentesis:

- Procedure is suitable from 15+0 weeks of pregnancy
- Is performed under continuous ultrasound guidance
- The procedure involves inserting a fine needle through abdomen into the womb and then into the amniotic sac and aspirating a sample of amniotic fluid (fluid surrounding baby)
- At the cytogenetic laboratory, fetal cells extracted from the amniotic fluid are examined; the number, arrangement and shape of the fetal chromosomes are checked
- About 1 in 100 samples proves to be inadequate
- The risk of miscarriage associated with amniocentesis is 1 fetal loss in 100 procedures

Chorionic Villus sampling (CVS):

- Procedure is usually performed from 11+0 to 14+6 weeks of pregnancy
- Performed in a Fetal Medicine Unit by a specialist trained in fetal medicine
- The procedure involves inserting a fine needle through the abdomen, into the womb and into the placenta and then aspirating a small amount of placental tissue for analysis
- At the cytogenetic laboratory, the fetal cells are extracted for examination; the number, arrangement and shape of the fetal chromosomes are checked
- About 2 in 100 samples proves to be inadequate
- The fetal loss rate associated with a CVS is 1 in 100 procedures

Quick info:

- Discuss with woman how she would like to receive results
- That she may experience discomfort in the lower abdomen after the procedure, which can be relieved with simple analgesics, eg. Paracetamol
- To contact the on call Midwife or FMU if she has a pyrexia (there is a slight risk of infection with invasive procedures), losing either fresh blood or water type loss (not urine) from the vagina, losing any discharge with an offensive odour from the vagina, severe lower abdominal pain, feeling generally unwell or decreased fetal movement (after amniocentesis only where some women have already experienced fetal movements)

Notes:

- Samples sent by ‘Special Delivery’ to Bristol Cytogenetics Lab
- Quantitative Florescent Protein Creatinine Ratio (QFPCR) result expected following 3 working days.
- 1:100 risk of mosaic result from QFPCR, discuss needing further diagnosis at 15 weeks
- CMW informed of procedure with consent of woman

Failsafe:

- Bristol informed that specimen is being sent
- Details of CVS and Amniocentesis, method of contact for result recorded in results file with proforma
• Giving of results recorded onto proforma and onto Viewpoint database

7. Miscarriage

Notes:
• Outcome of pregnancy recorded on Viewpoint database
• CMW informed
• Offer of appt to see Consultant Obstetrician post natal period

Failsafe:
• All miscarriages recorded onto screening database for audit and monitoring purposes

8. Discuss options with parents

Quick info:
• Provide information and support to the woman whose pregnancy is affected by Down’s, Edwards’ or Patau’s Syndrome and sign post her to other health professionals and organisations who have experience in this area e.g. Antenatal Results and Choices (ARC), Down’s, Edwards’ and Patau’s Syndrome Association, Contact a Family (CAF)

Notes:
• All positive results of Down’s, Edwards’ or Patau’s Syndrome conveyed to woman by chosen method.
• Appointment offered with ANNSC to discuss result and options available

9. Consider referral to Obstetrician or Neonatologist

Quick info:
• Depending on the parent’s decision, consider referral to an Obstetrician and Neonatologist

Notes:
• Following counselling if a Termination of Pregnancy (TOP) is chosen, paperwork commenced by ANNSC with support from Fetal Medicine Consultant
• If pregnancy is continuing, referral to Neonatal team made and area Consultant informed.
• All cases discussed at weekly Fetonatal meeting
Fetal Anomaly Screening

Offer fetal anomaly screening

Screening accepted

Initial screening test (dating scan)

Fetal anomaly suspected
Refer to fetal medicine department

Screening normal

Detailed 20 week anomaly scan

Screening accepted

Fetal anomalies suspected
Refer to Fetal Medicine Department

Screening normal
Follow up at delivery

Screening declined
Follow up at delivery

Screening declined
Follow up at delivery

Note in text for Cornwall pathway
Failsafe processes
1. Fetal Anomaly Screening

Quick info:
- The fetal ultrasound anomaly scan is between 18 to 20+6 weeks, to assess the possibility of structural abnormalities in the fetus and to identify:
  - Abnormalities not compatible with life
  - Abnormalities associated with morbidity or disability
  - Fetal conditions with the potential for intra-uterine therapy
  - Fetal conditions that will require postnatal investigation or treatment

Detection rate:
- General prevalence fetal anomaly is approximately 2%
- Congenital abnormalities that may be identified from the scan:
  - Spina Bifida
  - Anencephaly
  - Cleft Lip
  - Edwards’ (T18) and Patau’s Syndrome (T13)
  - Hydrocephalus
  - Major congenital heart problems
  - Diaphragmatic hernia
  - Exomphalos / Gastrochisis
  - Major kidney problems
  - Major limb abnormalities
  - The likelihood of fetal anomaly identification varies and in many cases may present late in pregnancy or after birth

Notes:
- Information will be given prior to booking in the form of “Screening Tests for you and your baby” in appropriate language.
- Dating and anomaly scan discussed at booking and verbal consent gained

Failsafe:
- CMW documents consent in hand held notes
- If woman has moved into area and there is no documented evidence of scan, the test is re-offered and decision recorded

2. Fetal anomaly suspected

Quick info:
- Refer to Fetal Medicine Obstetrician for discussion of implications of scan with parents at the first available appointment or within 72 working hours
- If indicated, refer to a centre with specialists and other relevant practitioners
- The parents can be provided with options:
  - to terminate pregnancy
  - to continue with pregnancy
  - written details about the scan result (and on the abnormality detected if possible) should be provided
  - follow-up and support should be provided

Notes:
- Appointment made with Fetal Medicine consultant within 3 working days to discuss findings and make a plan

Failsafe:
The scan findings are documented in the woman’s hand held maternity record and viewpoint system. Hard copy [thermal] images are taken and put into the hospital notes.

8 Scan normal

Notes:
The woman informed that no fetal anomalies found at the time of the dating scan.

Failsafe:
ANTENATAL AND NEWBORN SCREENING - CLINICAL GUIDELINE
The scan findings are documented in the woman’s maternity record and on the Viewpoint system. Hard copy [thermal] images are taken and put into the hospital notes.

9 Detailed (20 week) ultrasound scan

Quick info:
- all pregnant women should be offered an anomaly scan
- the scan should ideally be carried out between 18 and 20 weeks and 6 days
- the following should be explained to the woman prior to screening:
  - the scope of the screening test
  - the stages of screening process
  - the risks and benefits
  - rates of detection
  - clear, written advice should be provided before the scan
- the scan is used to assess for any obvious structural abnormalities
- the minimum standard for the 18 and 20 weeks and 6 day scan includes assessment of:
  - head
  - spine
  - abdomen
  - kidneys
  - chest
  - limbs and extremities
  - the optimal standard includes assessment of:
    - cardiac outflow tracts
    - lips

References:
UK National Screening Committee. Screening for fetal anomalies in pregnant women. 2007.

Notes:
Anomaly scan appointments generated from main ultrasound following dating scan. Any women booking late will have their anomaly scan booked by the community midwife. Community midwife to check that woman has her appointment at 15 week visit.

12 Scan declined

Failsafe:
The Community midwife documents the decision in her hand held records and offers further appointment if the woman changes her mind.
NIPE offered within 72 hour of delivery.

13 Follow up at delivery

Failsafe:
Newborn Physical and Infant Examination (NIPE) offered within 72 hrs of delivery.
Findings which deviate from the normal are documented in the infant records, PCH Records and stork IT system. Baby referred immediately to the Neonatal team.
The annual ‘10 conditions’ audit will be undertaken and reported in the annual report.
Any abnormalities will be notified to the South West Congenital Anomaly Register (SWCAR) by Fetal Medicine.

14 Fetal anomalies suspected

Quick info:
If fetal abnormalities are detected or suspected:
- refer to obstetrician for discussion of implications of scan with parents within 24 hours
- if indicated, referral to a centre with maternal fetal medicine specialists and other relevant practitioners should occur within 72 hours
- the parents can be provided with options:
• to terminate pregnancy
• to continue with pregnancy
• written details about the scan result (and on the abnormality detected if possible) should be provided
• follow-up and support should be provided

Notes:
The woman is offered the opportunity to see the fetal medicine specialist (unit) within 5 working days for further investigation.
Maternal decision is documented in maternity record.

Failsafe:
Scan findings are documented in the woman’s maternity record and viewpoint system and a second copy is sent to Fetal medicine. Referral can also be made by telephone and hard copy will be printed immediately in fetal medicine.
The community midwife will offer a further appointment if no documented results in hand held records (or received by professional referring from Trust) at next antenatal appointment
Fetal medicine will undertake the annual “10 conditions” audit as required by NHS FASP and will notify any abnormalities to the South West Congenital Anomaly Register (SWCAR).

15 Scan normal

Notes:
The woman informed that no abnormalities have been identified at the time of scan and that ultrasound cannot detect all problems.

Failsafe:
Ultrasound findings are documented in woman’s maternity record and viewpoint system. Digital images of measurements are taken and put with the report.
NIPE is offered within 72 hours of delivery. Evidence of decision will be recorded in maternity and infant record. If consent given for NIPE, evidence of examination will be reported in infant record and PCHR.

16 Follow up at delivery

Notes:
The woman is offered NIPE within 72 hours of delivery. Information will be provided.

Failsafe:
Offer and decision will be documented in maternity, infant record and hospital IT system.
If consent given for NIPE, evidence of examination report in infant record and PCHR.
NEWBORN BLOOD SPOT SCREENING

Links to the Sickle Cell and Thalassaemia screening programme

Check antenatal results and family history

Identify eligible population

Information provided and tests offered

Consent given

Some tests declined

All tests declined

Sample taken and despatched

Xx tests declined recorded on card, GP and HV informed

All tests declined recorded on card and despatched to lab, GP and HV informed

Card received in newborn screening lab

Quality check not OK

Repeat requested 16

Quality check OK

Newborn Screening lab tests sample. Results reported to Child Health Records

Inconclusive result

Not suspected

Carrier

Suspected – lab initiates immediate referral

Result to parents

Results to parents

Diagnosis confirmed and/or intervention/treatment initiated

PKU and MCADD

CHT

SCD

CF

Inherited metabolic diseases

Babies born pre-term or admitted to NICU

Note in text for Cornwall pathway

Secondary Care

Failsafe processes
1. Newborn Blood Spot Screening

**Quick info:**
Newborn blood spot screening (NBS) is a routine test, offered to all parents, for babies between 5 and 8 days of age (ideally at day 5 counting date of birth as day 0). The baby’s heel is pricked using a special lancet device and some drops of blood collected onto a blood spot card. The blood is tested to identify babies at high risk of nine rare conditions:

- Phenylketonuria (PKU) 1:10,000
- Congenital Hypothyroidism (CHT) 1: 4,000
- Sickle Cell Disease (SCD) 1:1,900
- Cystic Fibrosis (CF) 1:2,500
- Medium Chain acyl CoA Dehydrogenase Deficiency (MCADD) 1:10,000-20,000
- Maple syrup urine disease (MSUD) 1:100,000 to 1:150,000
- Isovaleric acidaemia (IVA) 1:100,000 to 1:150,000
- Glutaric aciduria type 1 (GA 1) 1:100,000 to 1:150,000
- Homocystinuria (pyridoxine unresponsive) (HCU) 1:100,000 to 1:150,000

The overall goal is for early detection and referral of babies, found to be high risk, to improve health and prevent severe disability or even death.

2. Links to the antenatal Sickle Cell and Thalassaemia screening programme

**Quick info:**
For parents, the link between antenatal and newborn screening is obvious: the result of an antenatal genetic test will be related to the results of a newborn baby’s genetic test for the same condition, so the linked programme provides a natural failsafe check between the mother and baby result.
The antenatal report given to women should contain all antenatal screening results for communication to primary care and allow linkage to newborn screening results where relevant.
There should be systems in place to inform newborn screening laboratories of all antenatal screening and diagnostic results and there should be a named person in every maternity unit with responsibility to ensure that newborn screening laboratories are informed of carrier women whose pregnancy is ongoing.

**Notes:**
All antenatal screening results are recorded in the woman’s handheld record.
The midwife will note on the newborn screening blood spot card any results significant to the screening process.
The ANNSC will inform the Newborn Screening lab in Bristol of any screening results and of any carrier women whose pregnancy is ongoing.

3. Check antenatal results and family history

**Quick info:**
Antenatal sickle cell and thalassaemia screening results should be recorded on the blood spot card.
Infants with a family history of a metabolic disorder may need special testing in the neonatal period and should be referred to a paediatrician in the antenatal period.

**Notes:**
Family history of metabolic disorders will be checked at the antenatal booking visit and recorded onto the risk page by the community midwife. This information will be written on to the NBBS card at the time of screening.
Infants of mothers with a family history of metabolic disorders will be referred to the Neonatologist by the community midwife or obstetrician if under Consultant Care.

4. Identify eligible population

**Quick info:**
The eligible population includes babies up to one year of age. The population is identified by:

- NN4B issued at birth notification
- NN4B issued at registration with a GP practice for babies born abroad that were not issued with an NHS number at birth.
Notes:
Any babies that move into the managed area of Cornwall will have the blood spot result recorded onto the Child Health Records System.

Failsafe:
All movers in will be identified by CHRD IT system (RIO) and a list sent weekly to lead Health visitor
Parents contacted by HV to arrange a transfer in visit. Requirement for newborn blood spot discussed.

5. Information provided and tests offered

Quick info:
Pregnant women are given the UK NSC information booklet ‘Screening tests for you and your baby’ at their booking appointment and new-born screening is discussed towards the end of pregnancy and at least 24 hours pre-test.
Antenatal sickle cell and Thalassaemia screening should be offered to all women and couples in pregnancy by 8-10 weeks to allow for the option of prenatal diagnosis.
The booklet ‘screening tests for your baby’ can be issued if parents have not kept or did not receive the UK NSC booklet earlier (e.g. families of babies born abroad). This contains new-born screening information only.

Notes:
Information is given at booking and discussed at birth plan visit by community midwife.

Failsafe:
A daily search on the’ new-born blood spot failsafe system (northgate)’ is made either by the ANNSC or Screening administrator and repeat sample requested in accordance with NBBS failsafe pathway.
A weekly search by the screening administrator

6 Consent given

Quick info:
Consent and reference to the provision of written information must be recorded in the maternity/baby health records and Personal Child Health Record (sometimes referred to as the ‘Red book’).

Notes:
Consent recorded in the hand held records and in the Red Book.

7. Some tests declined

Quick info:
Decline and reference to the provision of written information must be recorded in the maternity/baby records and Personal Child Health Record.
Inform parents who to contact if they change their minds.
Communicate parents’ wishes to laboratory by writing which condition(s) have been declined on the blood spot card.
Inform the GP and health visitor, which tests, have been omitted so they are aware, should symptoms arise, that the possibility of an affected child cannot be ruled out.
Send the blood spot card to the newborn screening laboratory. The child health records department is notified of the parents’ decision to decline by the normal communication lines via the laboratory.

8. All tests declined

Quick info:
Decline and reference to the provision of written information must be recorded in the maternity/baby records and Personal Child Health Record.
Inform parents who to contact if they change their minds.
Communicate parents’ wishes to laboratory by writing blood spot screening declined on the blood spot card.
Inform the GP and health visitor, which tests, have been omitted so they are aware, should symptoms arise, that the possibility of an affected child cannot be ruled out.
Send the blood spot card to the new-born screening laboratory. The child health records department is notified of the
parents’ decision to decline by the normal communication lines via the laboratory.

9. Babies born preterm or admitted to NICU

Quick info:
Babies admitted to neonatal intensive care units at less than 5 days of age should have a single circle blood spot sample taken and marked as ‘PRE-TRANSFUSION’

Please check that the pre-transfusion blood spot card has been taken and sent to the laboratory in accordance with local pathway.

Where a baby has already had a blood transfusion, either intrauterine or in the new-born period, before the screening blood sample has been taken, repeat samples are needed at 72 hours (3 days) after the blood transfusion for phenylketonuria, congenital hypothyroidism, cystic fibrosis and Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)

Where a baby has already had a blood transfusion, either intrauterine or in the new-born period, before the screening blood sample has been taken for sickle cell, the sample is sent for DNA analysis. This test identifies babies who might have sickle cell disease or babies who are sickle cell carriers, but not some of the rarer haemoglobin disorders or carriers of other haemoglobin variants identified on the routine test for sickle cell disease.

From 1st April 2012, a baby born before 32 weeks (less than or equal to 31+6 days) should have a repeat blood spot sample taken at age 28 days (birth date being day 0) or on discharge from the NICU, whichever is soonest, for congenital hypothyroidism.

Babies admitted to neonatal intensive care units are likely to have multiple blood samples taken. Blood spot screening should be coordinated with other tests. Venepuncture or venous/arterial sampling from an existing line is an alternative, this is providing the sample is not contaminated with heparin and the line cleared of infusate.

Analgesia in the form of breast feeding, non-nutritive sucking and a dose of sucrose or glucose is recommended for babies who undergo multiple invasive procedures.

Notes:
Day 0 sample is taken for all babies admitted to the neonatal unit and marked "pre-transfusion". If the baby does not go on to have a transfusion, the sample taken on day 5-8 is sent without the day 0 sample. If the baby does go on to have a blood transfusion the day 0 sample is attached to the day 5 sample and both are sent to the screening lab.
Any babies transferred out of the unit prior to day 5 will have the day 0 sample attached to their hospital record and sent with the baby.

10 Sample taken and despatched

Quick info:
The sample should be taken in accordance with Guidelines for Newborn Blood Spot Sampling and despatched on the same day in an identifiable screening envelope and by first class post.
The baby’s NHS number must be recorded on the blood spot card.
Record sickle cell and thalassaemia parental antenatal screening results on the blood spot card.

Notes:
Bar-coded Blood Spot Labels will be printed at the place of birth and sent home within the hand held notes. All bases have the ability to print these labels. If for any reason, bar coded labels are not available, the information will be completed by hand and will include the babies NHS number.
Labels must be checked for correct alignment and completeness of information before applying to blood spot card.
Stamped addressed envelopes for the blood spot cards are available from the ANNSC.
Any relevant genetic disorders in siblings i.e. MCADD will be entered onto the card. This information should also be sent to Consultant Neonatologist Paul Munyard and a copy to alert file on delivery suite.

11. xx tests declined recorded on blood spot card. GP and HV informed
Quick info:
See the information in the 'some tests declined' box, above.

12. All tests declined recorded on the blood spot card and despatch to laboratory. GP and HV informed

Quick info:
See the information in the 'all tests declined' box, above.

13. Blood spot card receipt in newborn screening laboratory

Quick info:
The newborn screening laboratory checks that the sample is satisfactory and that there is sufficient blood to complete screening on all five tests and that all data fields are completed to enable:

• The baby to be identified (NHS number is mandatory and bar-coded labels preferred)
• The laboratory to analyse the blood (including antenatal sickle cell and thalassaemia screening results)
• Identify the responsible PCT to report the results.

Sample receipt in the laboratory is notified to child health records and some maternity units, using the status code 01 to assist the timely identification of untested babies.

Notes:
A daily search on the ‘newborn blood spot failsafe system (Northgate)’ is made either by the ANNSC or Screening administrator and repeat sample requested in accordance with NBBS failsafe pathway. These are recorded on Newborn screening database for audit.

16. Request repeat

Quick info:
A repeat test may be requested because:

301 - Too young for reliable
302 - Too soon after transfusion
303 – Insufficient a) not soaked through, b) Too small spots
304 - Unsuitable sample (blood quality): multi spotted
305 - Unsuitable sample (blood quality): a) compressed/damaged b) stained glassine c) scratched/ridged
d) liquid/water damage e) Contaminated
306 - Unsuitable sample: day 1 and day 5 on same card
308 - Unsuitable sample: NHS number missing/not accurately recorded
309 - Unsuitable sample: Date of sample missing/not accurately recorded
310 - Unsuitable sample: Date of birth not accurately matched
311 - Unsuitable sample: Expired card used
312 - Unsuitable sample: >14 days in transit, too old for analysis
313 - Unsuitable sample: Damaged in transit

A local policy for repeat testing of babies must be in place, reasons for repeat tests must be communicated to the midwife in writing and passed onto parents.

Notes:
Any request for repeat test is sent through from the lab by e-mail to the ANNSC and administrator. The details are added to the Newborn Blood Spot database and a request for the repeat is rung out to the “On Call” midwife for ANTENATAL AND NEWBORN SCREENING - CLINICAL GUIDELINE
17. **Newborn screening laboratory tests sample. Results reported to child health records**

**Quick info:**
Screening test results should be communicated to the child health records departments daily, using the screening status codes.

**Notes:**
Once sample arrives in lab, this will be recorded on the ‘newborn blood spot screening system’ (Northgate).

**Failsafe:**
New births and children under 1 year of age are identified on the CHRD system (RIO). CHRD enters conclusive result onto the CHRD system by 17 days of age. (A small percentage of samples cannot achieve the 17 days standard due to mutation analysis (CF, SCD) and repeat tests (preterm repeat and borderline TSH). CHRDs perform daily checks to identify babies with status codes 01 specimen received in laboratory or 03 repeat/further sample required, for any of the nine conditions aged between 17 days and before their first birthday.

18. **Inconclusive result**

**Quick info:**
Results that are inconclusive require an additional sample to be obtained to complete the screening test. e.g. this may occur in cystic fibrosis and congenital hypothyroidism when the result is borderline.

**Notes:**
Repeat samples required for inconclusive result are requested via the ANNSC. On call midwife for the area will then be contacted to repeat the sample. This must be treated as urgent as screening will not be performed for Cystic Fibrosis after 8 weeks of age. Request recorded on Newborn Blood spot data base for audit.

19. **Not suspected**

**Quick info:**
The term ‘not suspected’ is used because screening test are not 100% certain.

Screening identifies babies who are genetic carriers of sickle cell or other unusual red blood disorders. Carriers of sickle cell are usually healthy and not affected by the condition (see the information in the ‘results to parents’ box, below).

Screening for CF includes testing some babies for the most common gene changes that cause CF. This means screening may identify some babies who are likely to be genetic carriers of cystic fibrosis. These babies may need further testing to find out if they are a healthy carrier, or have CF (see the information in the ‘results to parents’ box, under the ‘carrier’ box to the right).

20. **Carrier**

**Quick info:**

**Carriers of cystic fibrosis**
Screening for cystic fibrosis includes testing some babies for the most common gene changes that cause cystic fibrosis. This means screening may identify some babies who are likely to be genetic carriers of cystic fibrosis. These babies may need further testing to find out if they are a healthy carrier, or have cystic fibrosis (see the information in the ‘results to parents’ box, below).

**Carriers of Sickle cell**
Carriers of Sickle cell are usually healthy and not affected by the condition (see the information in the ‘results to parents’ box under the ‘not suspected’ box).
Carriers of MCADD
Carriers of MCADD are not detected by the screening test but may be detected through diagnostic testing.

21. Suspected - laboratory initiates immediate clinical referral

Quick info:
The laboratory reports phenylketonuria, congenital hypothyroidism, cystic fibrosis and MCADD screen positive results directly to the paediatric specialist team by telephone and in writing; the GP is also informed in writing.

Notes:
Health Visitor and GP informed by Lab by phone call followed by written information with appointment for referral as per disease specific pathways.
Lab informs ANNSC by letter.
Health Visitor will visit parents’ face-to-face to inform them of the diagnosis and give the referral appointment.
Disease specific information is taken to the parents at this point.

Failsafe:
Non attendees’ will be followed up as per the DNA policy, in a timely fashion. Confirmation of attendance at first appointment with clinical team will be reported back to the referring laboratory on all babies with screen positive results.

22. Results to parents

Quick info:
The child health records department notifies results to health visitors for reporting to parents. Best practice is for the child health records department to inform parents of normal results by letter. The screening results are recorded in the personal child health record.

Results to parents/ carriers of sickle cell
Robust processes should be in place to report newborn screening carrier results to parents. The format for reporting should be agreed locally but could be a face-to-face meeting or letter as appropriate.
For sickle cell screening carrier results, these should routinely be linked to maternal (and paternal) results where these are available.

Notes:
Any carrier results are sent to the GP and Health Visitor who will inform the parents at a face-to-face visit. The genetics counselling service will be informed by the screening laboratory and an appointment will be made to discuss the implications of the result.

Failsafe:
The child health records departments’ records results on the child health information system and notifies results to health visitors for reporting to parents.
The CCG ensures that parents have received results and they are recorded in the PCHR. Responsibilities for ensuring parents receive results lies with the health visitor. The HV will check the PCHR at the first visit post nata tally to ensure that there is a record of the test being performed. The HV will then ensure that the results are given to the parents by 6 weeks of age.

23. Results to parents

Quick info: Results to parents/ carriers of cystic fibrosis
All women and their partners with an infant who has a cystic fibrosis carrier result should be informed of the result, receive relevant information and material about the result, and be offered access to an appropriately trained healthcare professional to discuss the result. GPs should also routinely receive information about the baby’s carrier result.
A trained health care professional will make a visit to the family to inform them that their baby is thought to be a carrier of cystic fibrosis.
The cystic fibrosis carrier leaflet should be given.

Notes:
Any carrier results are sent to the GP and Health Visitor who will inform the parents at a face to face visit. The
24. **Diagnosis confirmed and / or intervention / treatment initiated**

**Quick info:**
The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome.

25. **PKU**

**Quick info:**
The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome.

**Notes:**
Pathway for referral from Bristol: First contact Neonatal consultant with special interest in PKU
If unavailable on-call paediatrician will be contacted
Specialist support from Regional Paediatric Metabolic Team, Bristol Children's Hospital

26. **CHT**

**Quick info:**
The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome.

**Notes:**
Pathway for referral from Bristol: First contact Neonatal consultant with special interest in CHT
If unavailable NICU Consultant on call to be contacted.

27. **SCD**

**Quick info:**
Confirmatory tests should be processed by laboratories who have experience in handling neonatal blood samples. Sickle cell centres and local hospitals should have robust follow up arrangements to identify and follow up any child who does not attend their hospital appointments and should be able to track children who have moved out of the area in order to make appropriate handover arrangements.

Infants should be prescribed oral penicillin by 3 months of age, routine Prevenar vaccinations and additional pneumococcal prophylaxis as outlined in the standards by 15 months of age.

**Notes:**
Pathway for referral from Bristol: First contact Neonatal consultant with special interest in CHT
If unavailable on call NICU paediatrician
Specialist support from Consultant Paediatric Haematologist, Bristol Children's Hospital.

28. **CF**

**Quick info:**
The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome.

**Notes:**
Pathway for referral from Bristol: First contact Neonatal consultant with special interest in CF
If unavailable back up available from CF Centre in Exeter

29. **MCADD and Inherited Metabolic Diseases**

**Quick info:**
The paediatric specialist team notifies the newborn screening laboratory of the diagnostic outcome.

**Notes:**
Pathway for referral from Bristol: First contact Neonatal consultant with special interest in MCADD
If unavailable on call paediatrician
Specialist support from Regional Paediatric Metabolic Team, Bristol Children's Hospital