Antenatal and Newborn Screening

Summary

- There are three national antenatal screening programmes (infectious diseases in pregnancy, sickle cell and thalassaemia, and fetal anomaly) and three national newborn screening programmes (newborn and infant physical examination, newborn hearing screening, and newborn bloodspot screening). These programmes are offered to all women and their babies in Hampshire.
- Each screening programme works to national quality standards. Screening programmes in Hampshire generally perform to the level required, although improvements can be made within some programmes.

Recommendations

- All providers of antenatal and newborn screening services need to use cohort data in their performance reports.
- Every maternity service provider should include outcome data for all high risk screens.
- All providers need to reduce their avoidable repeat rates for newborn bloodspot screening to the 0.5% achievable target.
- There should be plans to move to fully electronic processes and connections between computer systems and databases.
- Links between provider pathways should be maintained and strengthened to ensure a seamless and safe patient journey.

1. Introduction

Health screening 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, diagnostic tests and appropriate treatment to reduce their risk and/or any complications arising from the disease or condition.

The UK National Screening Committee (UK NSC) advises Ministers and the NHS in the UK about all aspects of screening and supports implementation of screening programmes[1]. There are 8 non-cancer screening programmes and 3 cancer screening programmes in England. Of the 8 non-cancer screening programmes, 6 are related to the antenatal and newborn periods.

1.1 Antenatal screening

There are three national antenatal screening programmes which are offered to all pregnant women. Midwives provide both written (Screening tests for you and your baby[2]) and verbal information at the booking visit to all pregnant women to inform them about the opportunities to protect them and their future offspring by screening. The programmes are:

- **Infectious Diseases in Pregnancy** Screening Programme (IDiPs) which offers pregnant women a blood test to identify infections which can then be treated to protect the health of the woman, her baby and sometimes her family. The blood is tested for Syphilis, Hepatitis B, HIV and Rubella.
- **Sickle Cell and Thalassaemia** Screening Programme (SCT) which offers screening for sickle cell disease during pregnancy for all pregnant women. The programme supports people to make informed choices during pregnancy; identifies babies with sickle cell disease promptly so they can get the care they need; provide high quality and accessible care throughout England and to promote understanding and awareness of Sickle Cell and Thalassaemia.
- **Fetal Anomaly** Screening Programme (FAS) which offers all women a blood test for Down's syndrome and an ultrasound scan between 18 – 20 weeks and 6 days to check for physical fetal abnormalities. This information is used to help the woman make decisions about her pregnancy and decide if she wants further diagnostic testing.

1.2 Newborn Screening

There are three national newborn screening programmes which are offered to all newborn babies in Hampshire supported by written and verbal information from the relevant healthcare professionals for the mothers and families.

- **Newborn and Infant Physical Examination** (NIPE): this consists of two examinations; one within 72hrs of birth and the other at 6-8 weeks of age. This screen offers parents the opportunity of a head to toe physical examination for their baby to check for problems or abnormalities. The examination is carried out

[1] http://www.screening.nhs.uk/searchwebsite.php?searchstring=tag%3AEngland&programme=Screening Portal&page=1

[2] http://www.screening.nhs.uk/annbpublications

within 72 hours of birth and then again at 6 to 8 weeks of age, as some conditions can develop or become apparent later. Each screen includes:

- A general all over physical check, as well as specific examination of the baby's eyes: About two hundred children a year are born in the UK with opacity of the lens of one or both eyes - a cataract.
- Heart: Congenital heart anomalies affect about 8 in 1000 (approximately 1%) new born babies.
- Hips: 1 to 2 in 1,000 babies born may have a hip that is dislocated at birth.
- Testes, in boys: around one in 20 male babies is born with an undescended testicle which is mote common in premature babies. The incidence at the age of one year is around 1%.

- **Newborn Hearing** Screening (NHSP): This screening programme aims to identify all children born with moderate to profound permanent bilateral deafness within 4-5 weeks of birth. This helps to ensure that safe high quality age-appropriate assessment and support for deaf children and their families is instigated as soon as possible. One to two babies in every 1,000 are born with a hearing loss in one or both ears and most of these babies are born into families with no history of hearing loss.
- **Newborn Bloodspot** screening (NBS): This involves taking blood from the baby's heel and then testing it for the presence of Phenylketonuria (PKU); Congenital Hypothyroidism (CHT); Sickle Cell Disease (SCD); Cystic Fibrosis (CF); and Medium-chain acyl-CoA dehydrogenase deficiency (MCADD).
    a. PKU is an autosomal recessive genetic condition that affects approximately 1 in 10,000 babies in the UK.
    b. CHT affects 1 in 4,000 babies in the UK.
    c. SCD affects 1 in 2,000 babies in the UK.
    d. CF is an autosomal recessive genetic condition which affects 1 in 2,500 babies in the UK.
    e. MCADD is an autosomal recessive genetic condition which affects around 1 in 10,000-20,000 babies in the UK.

1.3 Providers

The antenatal and newborn screening programmes are commissioned from a range of healthcare providers across Hampshire. These include the maternity units in acute hospitals, community healthcare providers, Public Health England accredited laboratories and regional specialist laboratories. Data for these programmes come directly from the service providers but also from Southern Health NHS Foundation Trust which is commissioned to provide the child health records database for all Hampshire babies. The data for these programmes are mostly collected and presented by maternity provider. Table 1 contains a list of providers and their acronyms.

Table 1: maternity providers in Hampshire and abbreviations

Acronym	Maternity Provider
UHS **SUHT**	University Hospital Southampton NHS FT formerly Southampton University Hospital NHS Trust
PHT	Portsmouth Hospital NHS Trust
HHFT **WEHCT** **BNHFT**	Hampshire Hospital NHS FT formerly Winchester and Eastleigh Healthcare NHS Trust and Basingstoke and North Hampshire Hospitals NHS FT
FPH	Frimley Park Hospital NHS FT

2. Level of need in the population

In 2011 there were 15,238 births in Hampshire (up from 14,970 in 2010). Trend analyses suggest a steady rise in the number of births in Hampshire, from 13,320 in 2000, to 15,238 in 2011 (an increase of 13%). The latest aggregated birth projections (2010 to 2033) for Hampshire show a slower, but on-going increase in delivery numbers until 2015 where they appear to plateau, followed by a possible decline.

In 2011/12 over 21,000 women and over 18,000 babies were offered screening as part of the national programme by Hampshire's main providers. Table 2 lists these numbers by provider; however only UHS was able to provide information solely for Hampshire residents.

Table 2: Offer of antenatal and newborn screening Hampshire 2011/12

Maternity Provider	No. of bookings	No. of births
FPH	5756	4162
BNHFT	3482	3472
WEHCT	3101	4326
PHT	6507	*3372
UHS	*2795	*2677
TOTAL	21,641	18,009

**Hampshire residents only*

3. Projected service use and outcome in 3-5 years and 5-10 years

Interim 2011-based Subnational Population Projections from the ONS published in 2012, indicate an on-going increase in delivery numbers. Therefore need for these services will be affected accordingly.

As Hampshire becomes more diverse, the potential for the prevalence of certain diseases will inevitably change. Currently Hampshire is defined as a low prevalence area for Sickle Cell and Thalassemia disease. Consequently routine red blood cell indices are used to screen for Thalassaemia whilst the Family Origin Questionnaire is used to identify women at risk of Sickle Cell and other haemoglobin variants. If a full blood count and family origin questionnaire suggests that the woman is a carrier for Thalassaemia, the laboratory will also screen for sickle cell disorders. When a significant risk is identified, the woman and her partner are then both offered a blood test. In high prevalence areas - where the disorders are more common - a blood test

is always done to determine if the woman is a carrier of Sickle Cell, Thalassaemia or another haemoglobin disorder.

The prevalence for each area is determined by the National Screening Committee. If and when Hampshire moves from a low to a high prevalence area there would be associated laboratory capacity issues for the commissioner to consider.

4. Current services in relation to need

Antenatal screening, 72 hour NIPE and newborn blood spot screens are provided by UHS, PHT HHFT and FPH maternity departments. NHSP is provided by NHS Solent, SHFT Health Visiting team and PHT whilst GPs are commissioned to provide the 6-8 week NIPE.

Coverage[3] and uptake of antenatal and newborn screening is high across all the programmes apart from the 72hr NIPE. This may relate to a data recording and transfer issue between paediatricians and child health records department and is being addressed.

A number of changes to the programmes since 2011/2012 have stretched the providers, including the following:

- National Key Performance Indicators[4] were introduced for the antenatal and newborn screening programmes. This has increased data collection and reporting requirements when providers may not have recognised the need for commensurate clerical or analyst support.
- Hampshire was volunteered as a pilot site for the development of a national Quality Assurance programme. This involved three maternity departments of acute hospitals and the associated providers.
- A National Failsafe audit had to be completed by all providers. This required the collating of policies, pathways and audits whilst the face to face review took a day to complete.
- Two hospital Trusts merged (WEHCT & BNHFT) requiring a streamlining of teams, roles, policies and procedures.
- FPH saw the merge of three pathology services which resulted in additional pressure on the antenatal screening co-ordinator to ensure that robust pathways continued to be followed for the IDiP and SCT screening programmes.
- Expansion of the FPH maternity service into the Berkshire area.

[3] Screening coverage refers to the proportion of persons eligible to be screened within a population who have been invited for screening during a specified period. The full antenatal screening data for 2011/12 can be accessed at: http://www.screening.nhs.uk/SEreports

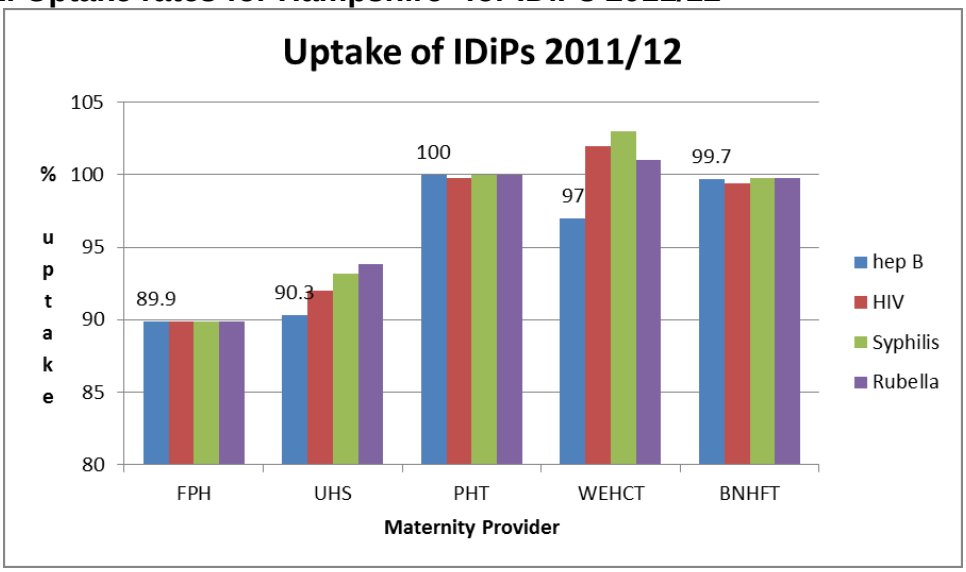
[4] http://www.screening.nhs.uk/kpi

4.1 Antenatal

4.1.1 Infectious Disease in Pregnancy Screen data

The IDiPs requires maternity service providers to achieve a coverage of at least 90.0% for hepatitis B; HIV; syphilis; and rubella. Figure 1 shows that the uptake of screening across all providers in Hampshire was above the 'acceptable' target level of 90% apart from FPH which achieved 89.9%. This seems to be a result of cross-border issues when women booking for delivery at FPH actually live outside the catchment area covered by the community part of the maternity service. Therefore the woman's community care is provided by another team who, when any blood samples are taken, send them to their own hospital laboratory for analysis. This means the results have not available in electronic form for the hospital where the woman has booked for delivery and consequently omitted from Key Performance Indicator (KPI) returns. This has been resolved. It is also worth noting from figure 1 that WEHCT did not provide cohort data as part of the quarterly KPI returns.

Figure 1: Uptake rates for Hampshire* for IDiPs 2011/12



**Includes women from Southampton, Portsmouth and Surrey*

4.1.2 Rubella

The data show that of the 25,600 women screened in Hampshire, 1,658 women were rubella non-immune - 6.5%. Rubella is serious for pregnant women and their fetus. Fetal abnormalities depend on the week of pregnancy in which the woman is unwell and include mental handicap, cataract, deafness, cardiac abnormalities, growth retardation and inflammatory lesions of brain, liver, lungs and bone marrow.

There has been a national increase in the number of cases of rubella, mostly in males at university. This is because young men were not offered rubella vaccination in the late 1980s. There has also been an increase in the number of reports of rubella infections in pregnant women. There will continue to be a small number of infections in pregnant women, through contact with young men in this country or acquired abroad.[5] Therefore it is important that women planning a pregnancy ensure

[5] http://www.hpa.org.uk/Topics/InfectiousDiseases/InfectionsAZ/Rubella/GeneralInformation/rubGeneralInfo/

there are fully vaccinated against this disease. Once pregnant, if the IDiPs identifies the woman as rubella non-immune, the MMR vaccine (which offers protection against rubella) cannot be offered until the postnatal period.

The following actions should be considered:

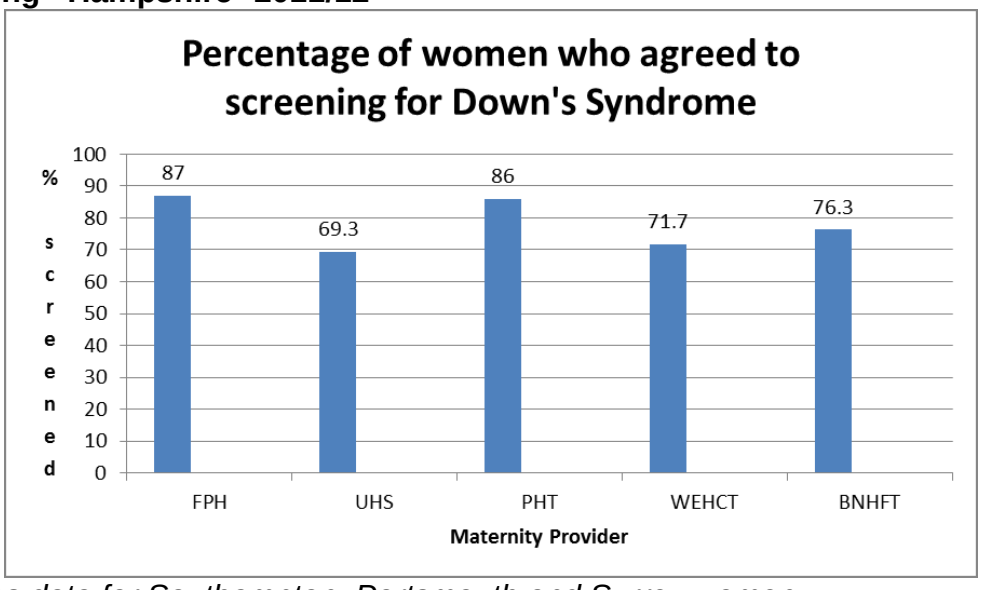
- FPH uptake rates for IDiPs to be monitored quarterly to ensure that the new system is effective.
- HHFT to provide cohort data for both sites.
- All maternity providers to offer MMR vaccine to eligible women as soon as possible after delivery.

4.1.3 Downs' Syndrome Screening

The FAS programme includes screening for Down's Syndrome. All pregnant women are offered screening for Down's syndrome with an early ultrasound scan to assess gestational age, nuchal translucency (NT) measurement and biochemistry testing between 10+0 and 14+1 week's gestation. The screening achieves a detection rate greater than 90% of affected pregnancies and a screen positive rate of less than 2%. All maternity providers also offer second trimester screening for Down's syndrome using the quadruple test between 14+2 and 20+0 weeks gestation where booking for maternity care has been too late to undertake first trimester screening.

Figure 2 shows the proportion of women who consented to screening for Down's syndrome. Although coverage is high (the number of women offered screening), the uptake of screening is lower. This reinforces the premise that screening is voluntary and some women may choose to decline screening.

Figure 2: Percentage of women who consented to Down's Syndrome screening –Hampshire* 2011/12



**Includes data for Southampton, Portsmouth and Surrey women*

Table 3 shows the high level outcome data for Down's Syndrome screening. UHS appears to be most likely to offer 2nd trimester screening. This may relate to the higher risk pregnancies cared for at this specialist provider. Second trimester screening is not as good at detecting Down's syndrome as first trimester tests.

Table 3: Outcome data for Down's Syndrome screening - Hampshire* 2011/12

		No. of Tests					
	No. of Bookings	**1st trimester**	**2nd trimester**	**Higher Risk**	**Prenatal Diagnosis**	**Total no. of cases**	**Unscreened cases**
WEHCT	3101	2105	121	68	46	9	1
BNHFT	3482	2265	326	69	28	10	0
UHS	6205	3475	830	120	59	7	1
PHT	6507	4944	558	158	135	18	1
FPH	5756	4838	167	151	109	14	0

**Includes data for Southampton, Portsmouth and Surrey women*

The following actions may need to be considered:

- Monitor the number of second trimester screens at UHS.
- Ensure every maternity provider collects and shares outcome data for all high risk screens.

4.1.4 Sickle Cell and Thalassaemia

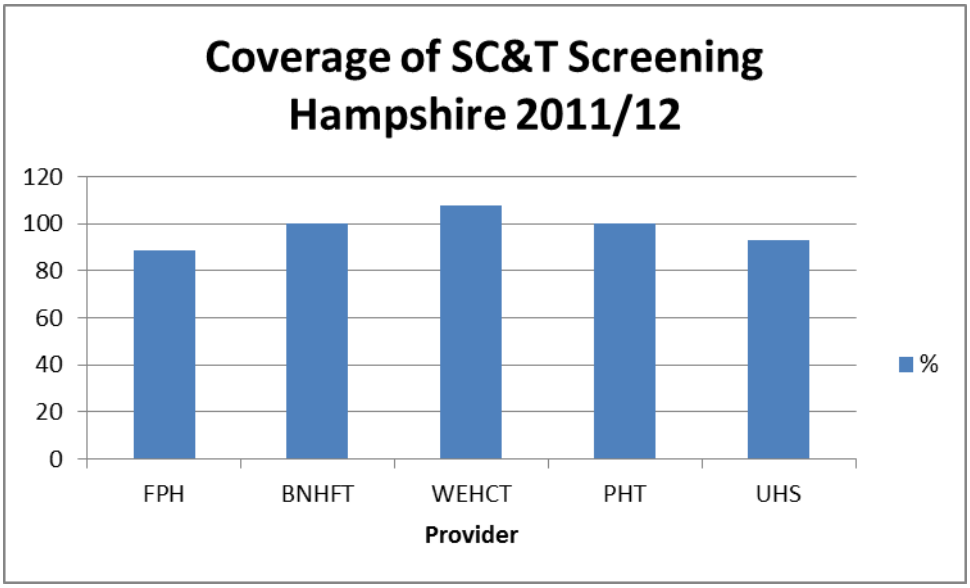
Hampshire is defined as a low prevalence area for Sickle Cell disease and the target for coverage is 95% acceptable and 99% achievable. Over 24,500 women were screened, 423 were identified as carriers and a total of 25 'at risk' were offered prenatal diagnosis (figure 3).

4.2 Newborn

4.2.1 Newborn and Infant Physical Examination

Table 4 shows the coverage for the newborn physical examination which is carried out before the baby is 72hrs of age. The national target is set at 95% 'acceptable' and 99.5% 'achievable'. The recorded uptake for Hampshire is well below 95%. This has been identified as a reporting error and work is on-going to address it.

Figure 3: Coverage of Sickle Cell and Thalassaemia Screening in Hampshire* 2011/12



**Includes data for Southampton, Portsmouth and Surrey women*

Although Hampshire has reported these data to the National Screening Committee it is not mandatory. A pilot is currently taking place to assess the effectiveness of a new electronic system known as SMaRT (Screening Management and Reporting Tool). This will capture the data at the point of screening which will then be automatically uploaded to a national database. It is anticipated that the tool will be ready for roll out nationally during 2013/14.

Table 4: coverage of the 72hr NIPE - Hampshire 2011/12

	Q1	Q2	Q3	Q4
Percentage of eligible babies with NULL (no) development checks recorded	7.0%	5.1%	6.7%	6.7%
Total number given NIPE *	59.4%	63.3%	60.7%	59.3%
Number given NIPE within 72 hours (**KPI NP1**)	**53.0%**	**58.2%**	**55.8%**	**55.5%**
Number given NIPE outside 72 hours	6.4%	5.1%	4.9%	3.8%

**Duplicate NIPE records on RiO Child Health Information System*

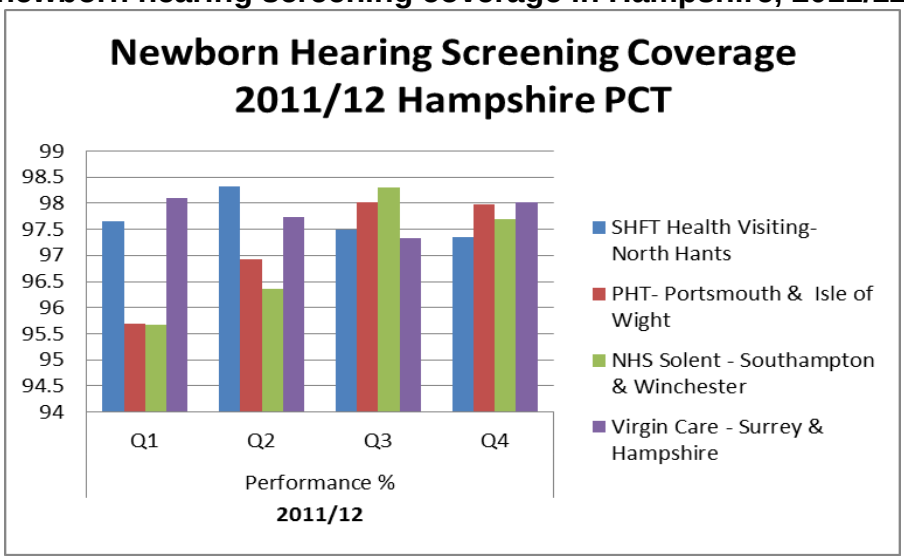
The following action may need to be considered:

- Encourage the National Screening Committee to ensure that the roll out of the SMaRT tool is accompanied by adequate implementation resources.

4.2.2 Newborn Hearing Screening

The national target for the coverage of the NHSP is 95% for ‘acceptable’ and 99.5% for ‘achievable’. Figure 4 shows that the ‘acceptable’ target was achieved across Hampshire in 2011/12 by all four providers.

Figure 4: newborn hearing screening coverage in Hampshire, 2011/12



4.2.3 Newborn Bloodspot screening

Poor quality blood samples for the NBS result in avoidable repeat tests which cause delay in identification and treatment of screen positive babies. It also leads to greater anxiety for parents and wastes healthcare resources. The NSC has set the avoidable repeat rate for NBS screening at ≤2.0% as the 'acceptable' target and ≤ 0.5% as the 'achievable' target.

Table 5 shows that only one provider across Hampshire achieved the 'acceptable' rate and none the 'achievable' rate. Analysis of the data confirms that the majority of avoidable repeat tests were due to 'insufficient samples'.

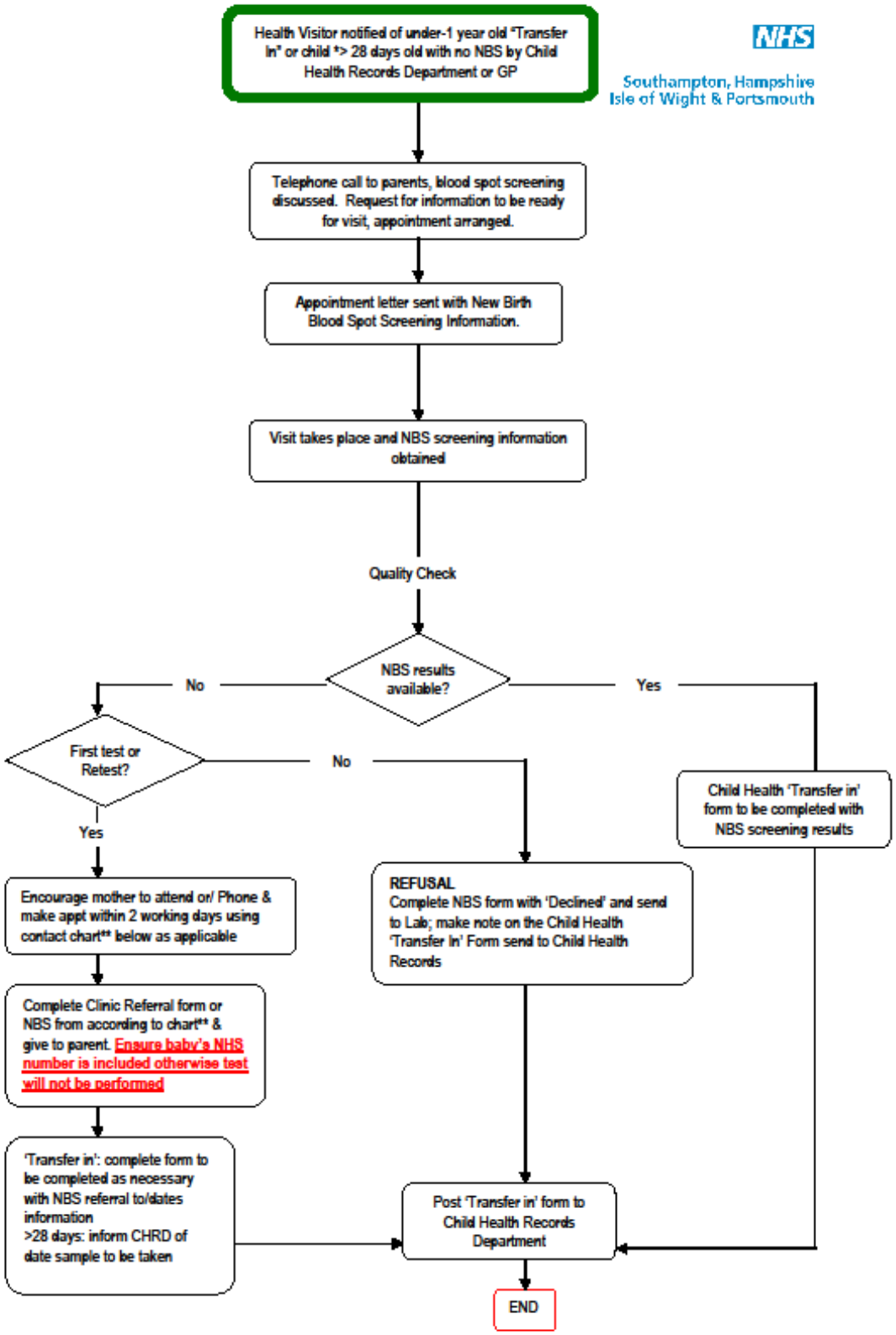
Table 5: avoidable repeat rates for newborn bloodspot screening in Hampshire 2011/12

TRUST	No. of tests	Total no. repeats	Avoidable repeat rate %
SUHT	6586	136	2.28%
WEHCT	2800	101	2.31%
BNHFT	3363	62	1.88%
FPH	4173	70	1.25%
PHT	6080	236	3.42%

Failsafe procedures are in place across Hampshire to help ensure that all babies receive a timely NBS screen. Timely screens are important to ensure that conditions are detected and treatment commenced as soon as possible. Some screens such as for cystic fibrosis (CF) have to be carried out within a set timeframe to be of use.

In response to several untoward incidents a newborn screening pathway has been implemented locally to ensure the failsafe mechanisms. This pathway has defined the roles and responsibilities of the health visitor, midwife, specialist laboratory and child health records department (figure 5).

Figure 5: Newborn Bloodspot Screening Pathway in Hampshire



A national failsafe to be introduced will highlight any samples that get lost in transit before the 14 day failsafe check is automatically carried out by Child Health Records Departments (CHRD). Currently all results from the laboratory are sent to CHRD where the data is manually uploaded by day 14. A letter is then generated for each parent confirming their baby's screen was negative. Any positive results are handled by the Director of Newborn Screening (Wessex) or by the Specialist BMS3 at Epsom and St Helier University Hospitals NHS Trust.

In 2011/12 a total of 12,669 Hampshire babies were screened and the following conditions identified (table 6):

Table 6: babies diagnosed with conditions from newborn screening tests, Hampshire 2011/12

CONDITION*	NO. CASES
PKU	0
MCADD	1
CHT	4
CF	10*

**Includes data from Portsmouth and Southampton cities*

The following actions should be considered:

- All providers must work to reduce their avoidable repeat rates to the 0.5% achievable target.
- Move towards electronic processes and connections between systems and databases to reduce human error and ensure failsafe measures are as robust as possible.

5. Evidence of what works

- Training for all personnel involved in any screening programme at any stage is vital. For example training was provided for CHRD staff explaining the importance of their role in relation to the NBS. This training raised awareness of the vital part the team play in the screening programme and the impact missed screens can have on the short and long term health of the baby.
- Seamless pathways which merge the roles of a variety of providers to ensure gaps in care are negated.
- Professional Networks or Fora. These provide opportunities to share problems and solutions; ideas and work enabling adaptation not reinvention.

6. Recommendations

- All providers of antenatal and newborn screening services need to use cohort data in their performance reports.
- Every maternity service provider should include outcome data for all high risk screens.
- All providers need to reduce their avoidable repeat rates for newborn bloodspot screening to the 0.5% achievable target.

- There should be plans to move to fully electronic processes and connections between computer systems and databases.
- Links between provider pathways should be maintained and strengthened to ensure a seamless and safe patient journey.