

6. CHILDREN, YOUNG PEOPLE AND THEIR FAMILIES

6.5 Antenatal and Newborn Screening

Screening is a process of identifying apparently healthy people who are at increased risk of a disease or condition, to offer information, further tests, and where appropriate, interventions to reduce risk or treat the condition. Antenatal and newborn screening aims to identify a range of conditions during pregnancy and the newborn period which are amenable to different types of interventions. These range from providing parents with information on which to base reproductive choices or to prepare themselves to care for a baby who may have congenital abnormalities, to identifying the need for specific preventative interventions such as immunisation, or for treatment which will prevent the baby developing a serious disabling condition.

There are a number of antenatal and newborn screening programmes¹ offered to women and their babies in the United Kingdom with some variation across the four countries. The details of the programmes with timelines are shown in appendix one. The UK National Screening Committee (UK NSC)² assesses evidence and makes recommendations³ to the four UK governments about population screening programmes. NHS England commissions all antenatal and newborn screening based on these recommendations.

6.5.1 The importance of antenatal and newborn screening

Midwives and other healthcare professionals provide information and offer testing as part of antenatal and postnatal care. Taking up full antenatal care including screening increases the chances of women having a problem-free pregnancy and a healthy baby.

6.5.1.1 Sickle Cell and Thalassaemia (SCT)⁴

The antenatal SCT screening programme aims to identify genetic carriers for sickle cell, thalassaemia and other haemoglobin disorders. If two people who are carriers have a baby together, there is an increased risk that their baby could inherit a haemoglobin disorder. About one in 2,000 babies born in the UK have sickle cell disease, a serious inherited blood disease. The screening test means that parents can make reproductive decisions early in pregnancy, and babies born with sickle cell disease can receive early treatment to help them live healthier lives. All pregnant women should be offered screening by the start of their 10th week of gestation using red blood cell indices, as well as a screen for haemoglobin variants using a combination of a questionnaire and, if appropriate, a blood test.

6.5.1.2 Infectious diseases in pregnancy⁵

All pregnant women are offered screening for Hepatitis B virus (HBV), HIV and syphilis early in their pregnancy. The aim of identifying HBV, HIV or syphilis is to prevent harm to the baby of the current pregnancy, to treat any possible infection or

reduce the risk of transmission of infection from the mother. For example, the baby of HBV-infected pregnant women should receive hepatitis B immune globulin and hepatitis B vaccine within 12 hours of birth⁶.

6.5.1.3 Foetal anomaly screening (FAS)⁷

FAS aims to identify foetal anomalies and Down's, Edwards' and Patau's syndromes, which are due to chromosomal abnormalities. The prevalence of Down's syndrome in England and Wales in 2010 was 26 per 10,000 total births⁸. This approximates to 10 to 15 babies in Buckinghamshire annually. The prevalence of congenital anomalies notified to the British and Irish Network of Congenital Anomaly Researchers (BINOCAR) registers in 2010 was 224 per 10,000 total births⁹, which is equivalent to approximately 100 to 125 babies in Buckinghamshire annually. The BINOCAR register estimates 16% of perinatal deaths (stillbirths and early neonatal deaths) had a congenital anomaly. This is likely to be an underestimate of the true incidence of congenital anomalies as some pregnancies with severe congenital anomalies are terminated before 24 weeks, whereas without intervention these pregnancies would have been likely to end in a perinatal death.

All pregnant women should be offered the combined test (which has superseded the double and triple tests) to screen for Down's syndrome usually before their 14th week of gestation. The combined test consists of a blood test for β -human chorionic gonadotrophin (β -hCG) and pregnancy associated plasma protein A (PAPPA), ultrasound assessment of nuchal translucency, and the quadruple test for women who come to antenatal services after their 14th week of pregnancy up to the 20th week of gestation.

All pregnant women in England should also be offered a minimum of two ultrasound scans during pregnancy. The early pregnancy scan, after eight weeks and before 13 weeks six days gestation, is to assess gestational age, check for viability and assess the number of fetuses. The second scan, between 18 weeks zero days and 20 weeks six days, is to screen for or diagnose foetal structural abnormalities.

6.5.1.4 Physical examination of newborns and infants¹⁰

Mothers of all infants are offered two screening physical examinations of their babies, under the Newborn and Infant Physical Examination Screening Programme (NIPE), commonly referred to as "baby checks". The first is conducted within the first 72 hours of birth, and then another at six to eight weeks of age. This is an overall physical check plus four different screening examinations:

- **Eyes:** looking for cataracts and other conditions. About two or three in 10,000 babies are born with problems with their eyes that require treatment.
- **Heart:** heart murmurs are common in babies, but about one in 200 babies has a heart problem that needs treatment.
- **Hips:** Developmental dysplasia of the hip (DDH) is due to hip joints that are not formed properly, and early treatment can prevent later disability. About one or two in 1,000 babies have DDH that needs treating.

- **Testicles:** In about one in 100 baby boys, the testicles descend only partially or not at all. This needs treating to prevent possible problems later in life, such as reduced fertility.

6.5.1.5 Newborn Blood Spot Programme¹¹

This test is offered on day five after birth, and aim to identify newborn babies at high risk of cystic fibrosis, congenital hypothyroidism, sickle cell disease, and a range of inherited metabolic conditions, to improve health and reduce disability or death.

- **Cystic fibrosis:** One in 2,500 babies born in the UK has cystic fibrosis. Early treatment can help them live longer, healthier lives.
- **Congenital hypothyroidism:** One in 3,000 babies born in the UK has congenital hypothyroidism. Early treatment with thyroxine prevents serious developmental problems and disability.
- **Sickle cell disease:** newborn babies are screened in addition to the antenatal parental screening (6.5.1.1. above).
- **Inherited metabolic diseases:** Six conditions are currently screened for which are rare, but may be life threatening or cause severe developmental problems and if detected early can be treated with a carefully managed diet and, in some cases, with medicines.

6.5.1.6 Newborn Hearing screening¹²

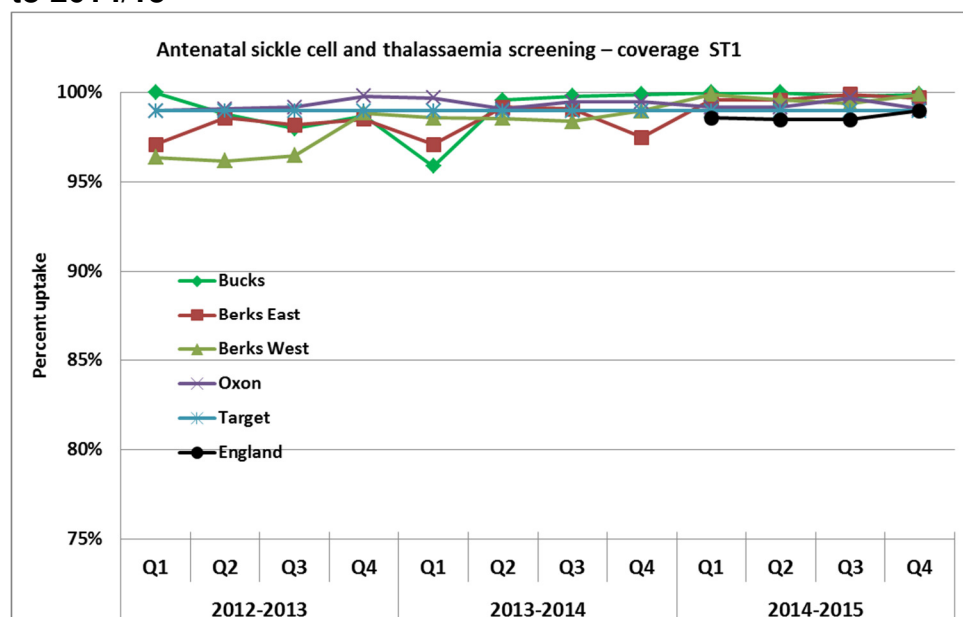
The incidence of significant permanent congenital hearing impairment is about one in 1,000 live births in most developed countries although this may be three to four times higher in certain communities or parts of the UK. Early diagnosis of hearing loss is important as it has been shown to improve outcomes for children's language, social and emotional development and education¹³. Newborn hearing screening detects hearing impairment of at least 40dB in the better ear. All parents are offered a hearing screen for their baby within four to five weeks of birth; in Buckinghamshire this is usually carried out before discharge from hospital. All positive babies are referred for full audiological assessment and should be seen for within four weeks of screen completion.

6.5.2 Information about antenatal screening programmes in Buckinghamshire

6.5.2.1 Antenatal sickle cell and thalassaemia screening

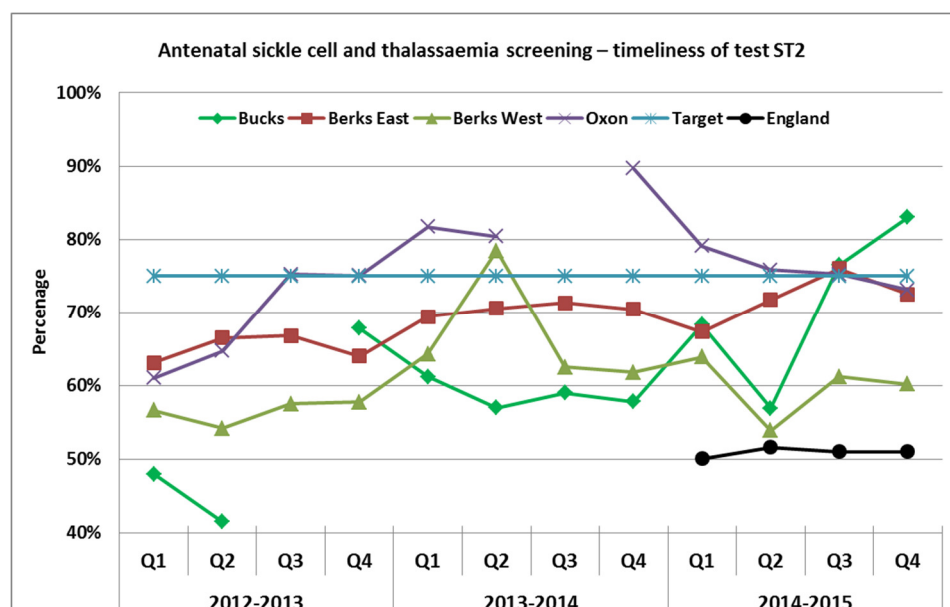
Figure 1 shows that uptake of antenatal sickle cell and thalassaemia screening in Buckinghamshire remains high, above the 99% target set by the Department of Health's Screening Standards. However, there have been wide fluctuations both in the timeliness of the test (figure 2) and completion of the Family Origin Questionnaire (FOQ) which can indicate increased risk of these conditions (figure 3). Although both these have shown improvements in the last few quarters of 2014/15, completion of the FOQ remains below the national target and national average. Timeliness of the test is subject to fluctuation as some of the delay can be because of late pregnancy booking.

Figure 1 Antenatal sickle cell and thalassaemia screening coverage (%) in Bucks compared to neighbouring areas and national averages (ST1), 2012/13 to 2014/15



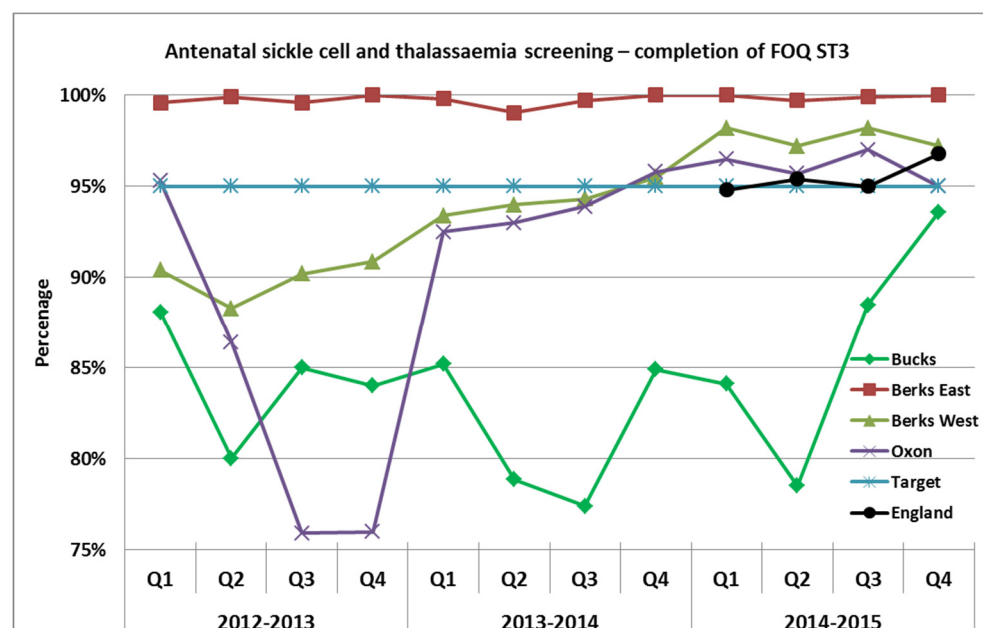
Source: NHS Screening Programmes

Figure 2 Antenatal sickle cell and thalassaemia screening, timeliness of test (%) in Bucks compared to neighbouring areas and national averages (ST2), 2012/13 to 2014/15



Source: NHS Screening Programmes

Figure 3 Antenatal sickle cell and thalassaemia screening, completion of FOQ (%) in Bucks compared to neighbouring areas and national averages (ST3), 2012/13 to 2014/15



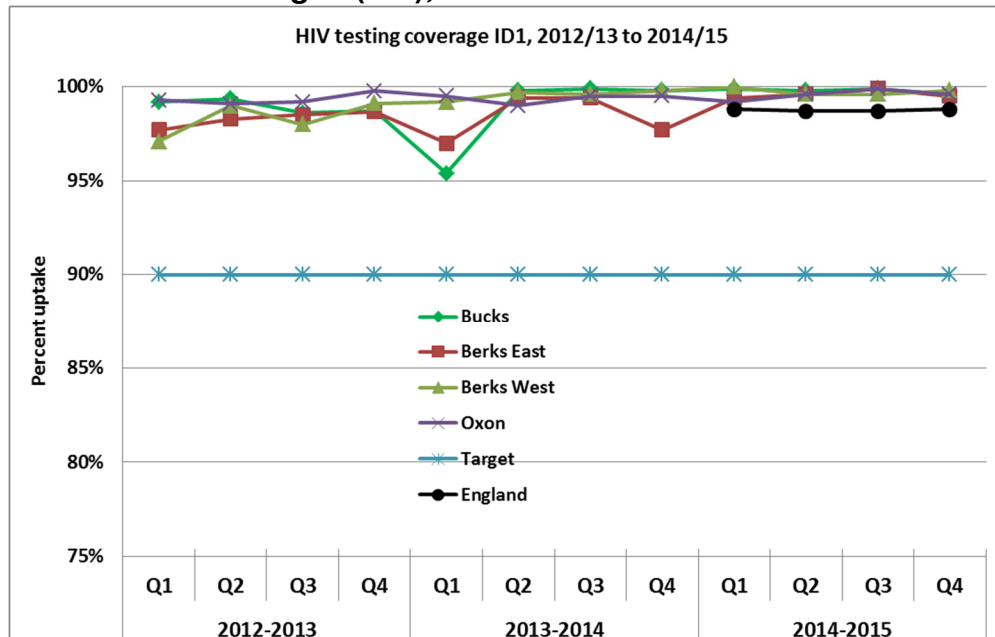
FOQ: Family Origin Questionnaire

Source: NHS Screening Programmes

6.5.2.2 Infectious diseases in pregnancy

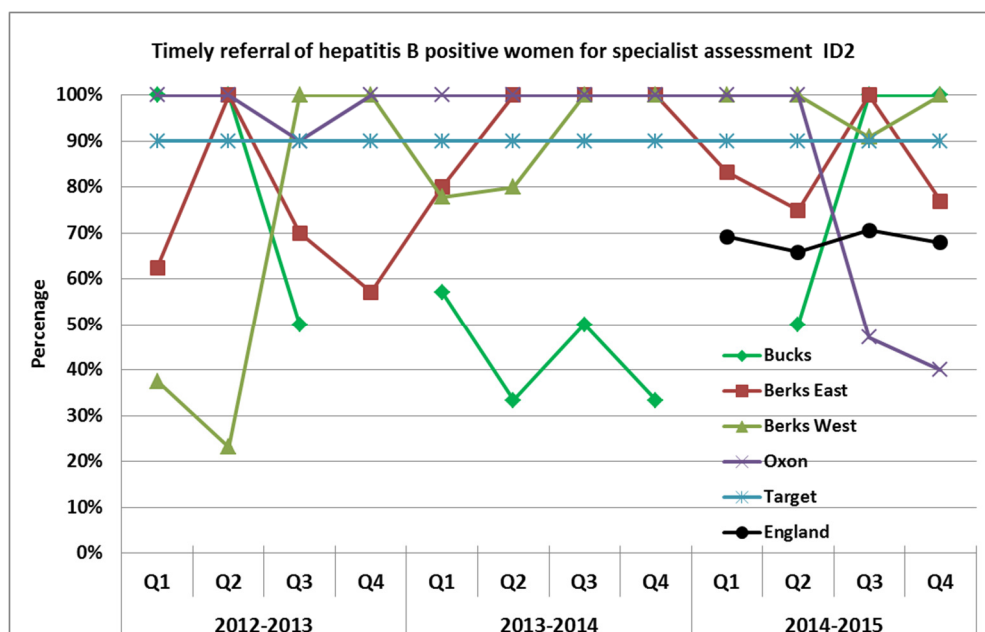
The Department of Health has set a target of 90% for the uptake of screening all four infections¹⁴, and the UK National Screening Committee has also set Key Performance Indicators (KPIs) for HIV coverage and timely referral of hepatitis B positive women for specialist care¹⁵. The coverage of antenatal screening for hepatitis B, HIV, syphilis, and susceptibility to rubella infection in Buckinghamshire remains well above 90% (see figure 4, HIV testing coverage). The number of screened women who tested positive for HIV, syphilis and hepatitis B is relatively small. However, there have been some issues in Buckinghamshire with timely referral of hepatitis B positive women for specialist assessment (figure 5). These have now been resolved as the hospital offers a dedicated slot to ensure pregnant women referred for hepatology services are seen in a timely manner.

Figure 4 HIV testing coverage (%) in Bucks compared to neighbouring areas and national averages (ID1), 2012/13 to 2014/15



Source: NHS Screening Programmes

Figure 5 Timely referral of hepatitis B positive women for specialist assessment (%) in Bucks compared to neighbouring areas and national averages (ID1), 2012/13 to 2014/15

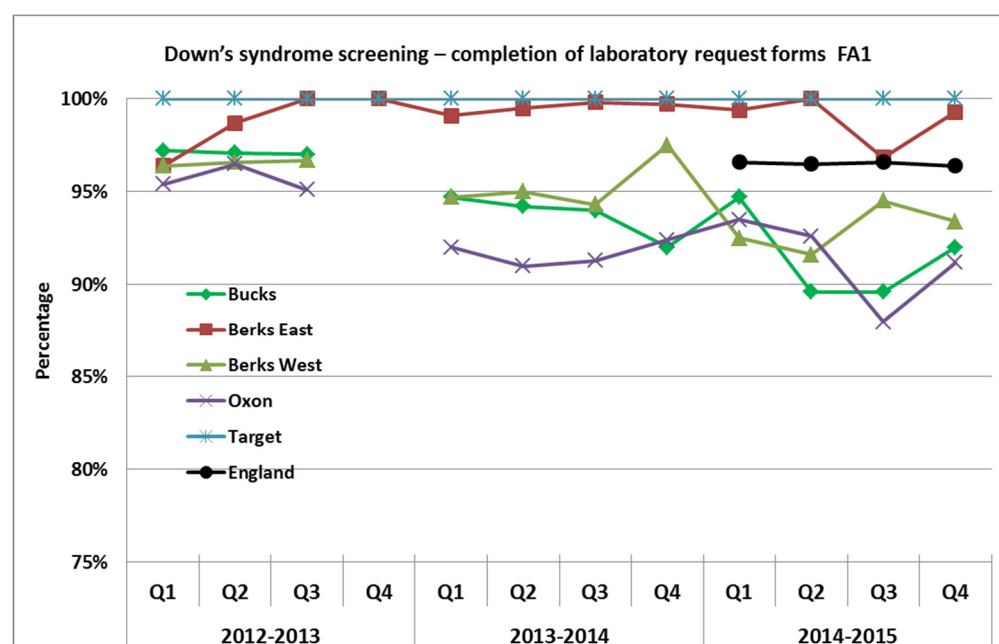


Source: NHS Screening Programmes

6.5.2.3 Foetal anomaly screening

Overall, the coverage of foetal anomaly screening is good in Buckinghamshire and is similar to neighbouring areas and England (relevant data from NHSE should be available shortly). However, the completion rate of laboratory request forms for Down's syndrome screening in Buckinghamshire is well below the national target, a number of neighbouring local authorities and national averages (figure 6).

Figure 6 Down's syndrome screening – completion of laboratory request forms (%) in Bucks compared to neighbouring areas and national averages (FA1), 2012/13 to 2014/15



Source: NHS Screening Programmes

6.5.3 Information about newborn screening programmes in Buckinghamshire

6.5.3.1 Physical examination of newborns and infants

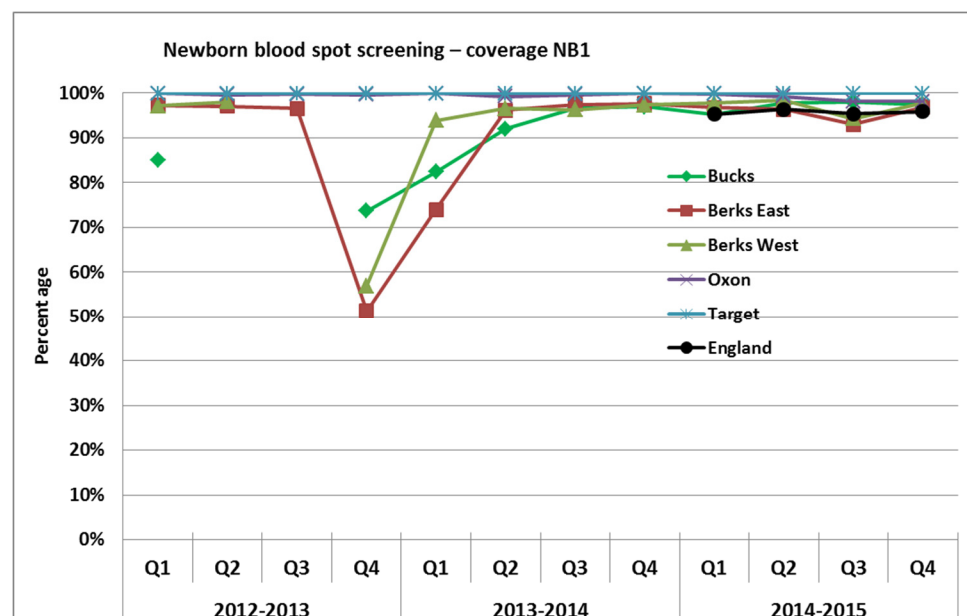
This programme has only recently been systematically implemented so local data are not yet available, but should be available soon as the programme is rolled out across Buckinghamshire.

6.5.3.2 Newborn blood spot screening

Recorded coverage of this test in Buckinghamshire was 75% in Q4 of 2012/13 due to a number of factors including data quality. However, the coverage has improved since (97.5% in Q4 2014/15) and remains higher than the national average, but below the 99.9% target set by the Department of Health (figure 7). The avoidable repeat test (NB2) in Buckinghamshire has recently increased and is higher (worse) than the national target; national average and some neighbouring authorities (figure 8). This coincided with the implementation of more stringent laboratory criteria across Thames Valley since January 2015. It is expected that this practice will

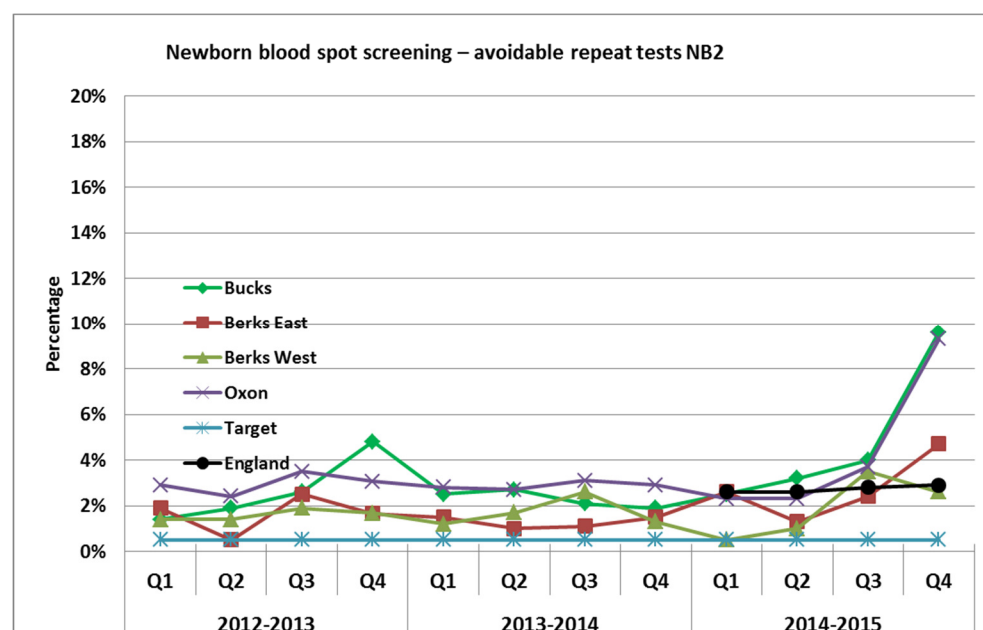
become embedded and avoidable repeats should reduce over time. Timeliness of result availability in Buckinghamshire has been 100% in all four quarters of 2014/15 and is better than the national target (98%).

Figure 7 Newborn blood spot screening – coverage (%) in Bucks compared to neighbouring areas and national averages (NB1), 2012/13 to 2014/15



Source: NHS Screening Programmes

Figure 8 Newborn blood spot screening – avoidable repeat tests (%) in Bucks compared to neighbouring areas and national averages (NB2), 2012/13 to 2014/15

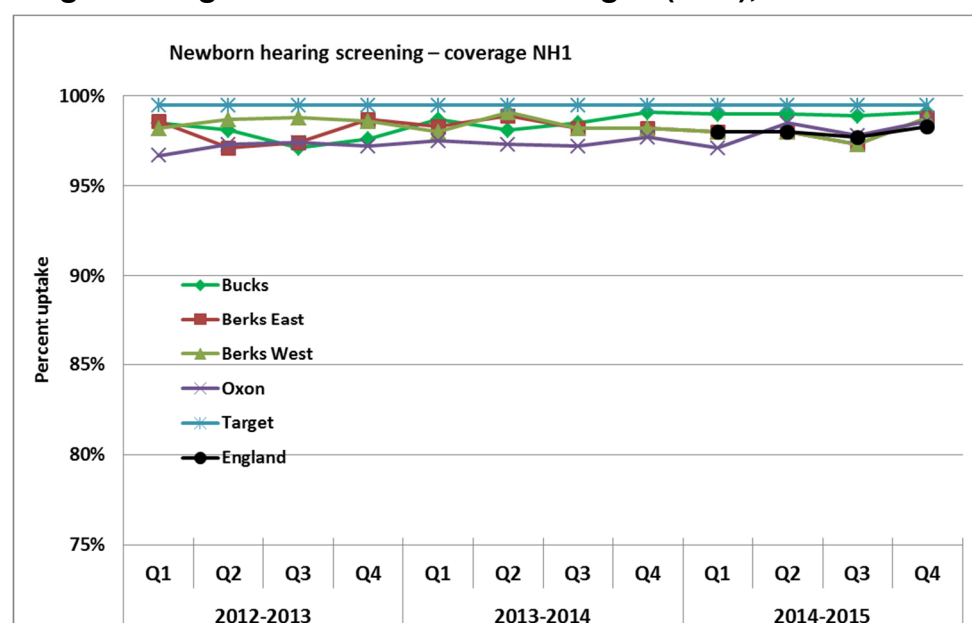


Source: NHS Screening Programmes

6.5.3.3 Newborn hearing screening

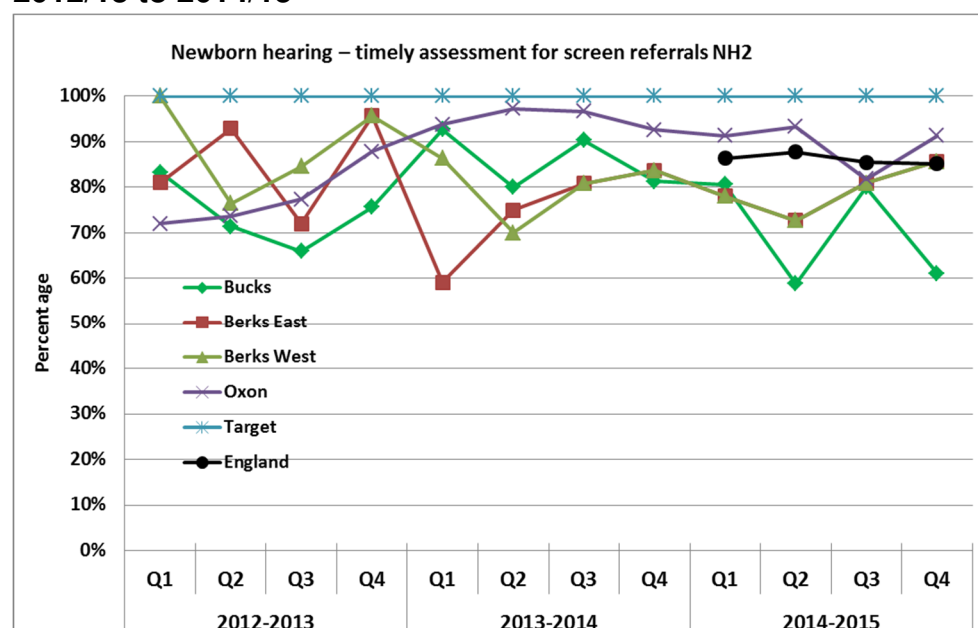
The coverage of newborn hearing screening in Buckinghamshire has improved, and remains higher (99.1% in Q4 2014/15) than the national average (98.3%) and a number of neighbouring areas, but just below the 99.5% target set by the Department of Health (figure 9). Coverage in Buckinghamshire was higher than most other areas in the South East in 2013/14, and significantly higher than the national average. The timely assessment of screen referrals in Buckinghamshire was between 60-80% over the last year, lower than the national target (100%), national average (85.2%) and a number of neighbouring authorities (figure 10).

Figure 9 Newborn hearing screening – coverage (%) in Bucks compared to neighbouring areas and national averages (NH1), 2012/13 to 2014/15



Source: NHS Screening Programmes

Figure 10 Newborn hearing screening – timely assessment of screen referrals (%) in Bucks compared to neighbouring areas and national averages (NH1), 2012/13 to 2014/15



Source: NHS Screening Programmes

6.5.4 Demand

The commissioning and delivery arrangements for antenatal and newborn screening programmes are complex. NHS England holds commissioning responsibility but funding arrangements remain with clinical commissioning groups (CCGs). In addition, acute, community trust and primary care providers are responsible for various elements of the pathway leading to complexities, which need to be managed carefully to ensure safe delivery. Partnership working across a number of organisations and professional groups is vital to the delivery of whole screening pathways and all appropriate partners must work together to make these entire programmes safe and effective locally.

6.5.5 Horizon scanning

Each English Screening Programme has a defined set of standards that providers have to meet to ensure that local programmes are safe and effective. Quality assurance (QA) is the process of checking that these standards are met and encouraging continuous improvement across screening and referral pathways, in order to ensure that pregnant women and their babies have access to a high quality service wherever they reside. This is essential in order to minimise harm and maximise benefits. Buckinghamshire County Council Health Protection Committee should have greater influence in overseeing and making sure these programmes are safe and effective locally.

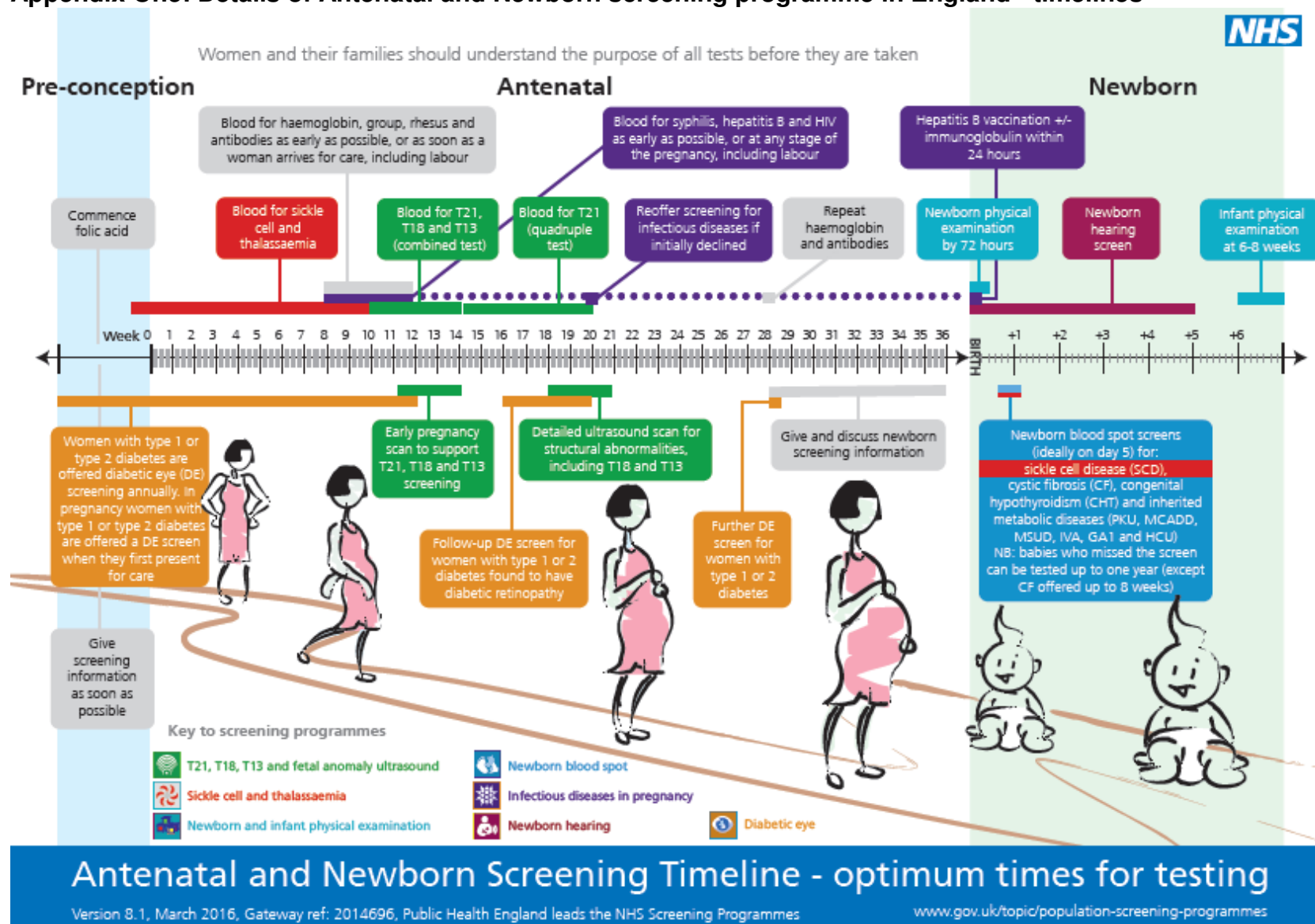
6.5.6 Conclusions

A number of antenatal and newborn screening programmes are offered to women and their babies in Buckinghamshire in line with the UK National Screening Committee recommendations. Robust implementation and monitoring are required to ensure the programmes are delivered as effectively as possible to improve health and reduce the risks of ill-health for mothers and their babies.

Overall, all antenatal and newborn screening programmes in Buckinghamshire are performing well. However, certain elements of some screening programmes appear to need further investigation and action to improve delivery. These include an increase in completion of the Family Origin Questionnaire and more consistency in the timeliness of testing in antenatal sickle cell and thalassaemia screening; an increase in the correct completion of laboratory request forms for Down's syndrome screening; a reduction in the proportion of avoidable repeat tests in newborn blood spot screening, and more timely assessment of screen referrals in newborn hearing screening.

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Appendix One: Details of Antenatal and Newborn screening programme in England - timelines



References

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- ⁸ http://www.binocar.org/content/Annual%20report%202010%20FINAL%2031_07_12%20v2.pdf (accessed on 17/02/2016)
- ⁹ Congenital Anomaly Statistics, 2010; England and Wales. British Isles Network of Congenital Anomaly Registers, July 2012
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- ¹² NHS newborn hearing screening programme (NHSP). <https://www.gov.uk/topic/population-screening-programmes/newborn-hearing> (accessed on 14/12/2015)
- ¹³ Yoshinaga-Itano C, Coulter D, Thomson V. Developmental outcomes of children with hearing loss born in Colorado hospitals with and without universal newborn hearing screening programs. *Semin Neonatol* 2001;6:521-9.
- ¹⁴ UK National Screening Committee (2010). IDPS Programme: programme standards, <http://infectiousdiseases.screening.nhs.uk/standards> (accessed on 14/12/2015)
- ¹⁵ IDPS Programme: key performance indicators, <http://infectiousdiseases.screening.nhs.uk/reporting> (accessed on 14/12/2015)