Newborn Bloodspot Screening Wales
Sgrinio Smotyn Gwaed Newydd-anedig Cymru

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

# Newborn Bloodspot Screening Wales Annual Statistical Report 2016-17

November 2017

*Screening Division of Public Health Wales*

# About us

Public Health Wales exists to protect and improve health and wellbeing and reduce health inequalities for people in Wales.

We are part of the NHS and report to the Minister for Health and Social Services in the Welsh Government.

Our vision is for a healthier, happier and fairer Wales. We work locally, nationally and, with partners, across communities in the following areas:

**Health protection** – providing information and advice and taking action to protect people from communicable disease and environmental hazards

**Primary, community and integrated care** – strengthening its public health impact through policy, commissioning, planning and service delivery

**Microbiology** – providing a network of microbiology services which support the diagnosis and management of infectious diseases

**Safeguarding** - providing expertise and strategic advice to help safeguard children and vulnerable adults

**Screening** – providing screening programmes which assist the early detection, prevention and treatment of disease

**Health intelligence** – providing public health data analysis, evidence finding and knowledge management

**NHS quality improvement and patient safety** – providing the NHS with information, advice and support to improve patient outcomes

**Policy, research and international development** – influencing policy, supporting research and contributing to international health development

**Health improvement** – working across agencies and providing population services to improve health and reduce health inequalities

## Further information


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**Facebook:** www.facebook.com/PublicHealthWales

The report is only available electronically from the screening programme and will be available on the website:
www.newbornbloodspotscreening.wales.nhs.uk

This report is a detailed summary of information on work undertaken by Newborn Bloodspot Screening Wales for the financial year from April 2016 to the end of March 2017. Results are reported by Health Board where screening has been carried out. Further details are available on request.

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**Quality Assurance Statement**

Screening data records are constantly changing. The databases used by Public Health Wales Screening Division are updated on a daily basis when records are added, changed or removed (archived). This might relate to when a person has been identified as needing screening; has had screening results that need to be recorded, or has a change of status and no longer needs screening respectively. Data is received from a large number of different sources with varying levels of accuracy and completeness. The Screening Division checks data for accuracy by comparing datasets for example GP practice data – and corrects the coding data where possible. It should be noted that there are sometimes delays in data collection for example a person might not immediately register with their GP. These delays will therefore affect the completeness of the data depending on individual circumstances. In addition, the reader should be aware that data is constantly updated and there might be slight readjustments in the numbers cited in this document year on year because of data refreshing.

# Contents



# Tables and Graphs

# 1 Introduction

This is the first annual statistical report published by Newborn Bloodspot Screening Wales (NBSW). This report covers data for babies born between 1st April 2016 and 31st March 2017.

Newborn bloodspot screening is when a small sample of blood is taken from the baby's heel on day five of life (counting day of birth as day 0). This blood sample is screened for rare but serious diseases that respond to early intervention to reduce mortality and/or morbidity. The screening test is part of routine postnatal care.

The aim of the Newborn Bloodspot Screening programme in Wales is to offer all eligible babies, at day five of life, quality assured screening for rare but serious diseases that would benefit from early intervention and reduce mortality and/or morbidity from the disease.

In Wales all eligible babies are offered screening for the conditions below which are recommended by the UK National Screening Committee:

- Congenital hypothyroidism (CHT)
- Cystic fibrosis (CF)
- Inherited metabolic disorders (IMDs):
    - Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)
    - Phenylketonuria (PKU)
    - Maple syrup urine disease (MSUD)
    - Isovaleric acidaemia (IVA)
    - Glutaric aciduria type 1 (GA1)
    - Homocystinuria (HCU)
- Sickle cell disorders (SCD)

## 1.1 Key messages for parents

Information for parents and the general public has been produced and is summarised in the NBSW key messages leaflet. The following messages are included:

- Newborn bloodspot screening identifies babies who may have rare but serious conditions
- If your baby is found to have any of the conditions they will receive early specialist care and treatment
- Early treatment can improve your baby's health and prevent severe disability or even death

- Screening is not 100% accurate. If the screening test suggests a problem, your baby will need further tests to confirm that they have the condition
- Newborn bloodspot screening is recommended
- The 'Information for Parents' leaflet, which is available from your midwife, explains the conditions screened for and how the sample is taken

## 1.2 Programme delivery

The Screening Division of Public Health Wales is responsible for the planning, preparation and delivery of the Newborn Bloodspot Screening Wales (NBSW) programme. NBSW is one of three programmes within Maternal and Child (MAC) Screening, which has an overall Programme Lead. There are two NBSW programme co-ordinators with administration support across the MAC programmes. The other two programmes are Antenatal Screening Wales (ASW) and Newborn Hearing Screening Wales (NBHSW).

The offer of newborn bloodspot screening to eligible babies and the collection of bloodspot samples is undertaken by health professionals within the seven health boards in Wales.

The Wales Newborn Screening Laboratory in Cardiff is responsible for testing the screening samples taken in Wales and for the referral of babies suspected of having conditions. Babies are referred to a network of clinicians and designated medical leads in the health boards. The programme has external Quality Assurance Advisors which include some of the medical leads.

The Newborn Bloodspot Screening Wales System (NBSWS) has been developed to support the management of a safe and sustainable programme across Wales. This system collects and collates information across the programme to monitor the quality of newborn bloodspot screening and provides quality assurance and management reports based on the policies and standards.

NBSWS also identifies babies for whom the programme expects to receive either a bloodspot card or decline for the test(s), and initiates failsafe procedures for possible 'missed' babies. This failsafe system identifies babies in Wales who do not have a newborn bloodspot screening sample in the Newborn Screening Laboratory by day 14 of life. Every baby identified

by the failsafe is followed up by the administration failsafe teams. The three regional teams across Wales are staffed by newborn screening managers and administrative staff who work across both the NBSW and Newborn Hearing Screening Wales (NBSHW) programmes.

In each health board there is a Governance Lead for Antenatal and Newborn Screening. This role, funded by Screening Division, Public Health Wales, is to act as liaison between the health board and NBSW, and to lead the provision of newborn bloodspot screening in the health board to ensure the provision of an effective and efficient service.

## 1.3 Screening pathway

Babies who are eligible for screening are identified in each health board from midwife birth notifications. Eligible babies up to one year of age who move in to Wales are identified following registration on to the Welsh Child Health System.

The offer of screening and collection of bloodspot samples is carried out by health professionals within the health boards in accordance with the NBSW guidance, standards and policies. The majority of samples are taken in the baby's home by the midwife. Neonatal or paediatric unit staff offer the screening and take samples for those babies who are inpatient in those areas at day five of life. Health visitors take responsibility for offering and arranging sample collection for eligible babies who have moved into Wales.

Newborn bloodspot screening samples are sent by prepaid envelopes (first class) to the Wales Newborn Screening Laboratory in Cardiff for testing. The laboratory accepts samples according to the UK sample quality guidelines that were implemented in April 2015. Babies suspected of having one of the conditions screened for are referred, according to the relevant clinical referral guidelines, to the appropriate specialist clinician for diagnostic tests and treatment. The newborn screening laboratory currently sends a hard copy of the result report for each baby to the local Child Health Department. The results are entered onto the Child Health system and the report is then forwarded to the baby's health visitor so that the results can be discussed with the parents.

More information is available at:
www.newbornbloodspotscreening.wales.nhs

# 2 Headline Statistics

### April 2016 to March 2017

- The number of eligible births across Wales was 33,627
- The number of these babies tested was 33,505 (99.6%)

## Screening

### Completeness of offer and coverage (eligible newborns)

- Completeness of offer - 97.3% of babies had a bloodspot card (for screening or decline) received in the laboratory by day 14 of life
- Coverage - 94% of babies had conclusive bloodspot screening results by day 17 of life

### Timeliness of sample collection

- Timely collection of sample (day 5-8 of life) – 98.0%
- Timely collection of sample (day 5 of life) – 75.3%

To monitor performance more closely the rate of sample collection at day 5 is also being measured.

### Avoidable repeat rate

- Avoidable repeat rate - 5.5%

Improving performance in collecting good quality samples remains a high priority for the programme to avoid delays in the referral of babies.

### NHS number on bloodspot card

- 99.1% bloodspot cards received in the laboratory had a valid NHS number for the baby recorded.

### Timely receipt of card in laboratory

- 95.6% of bloodspot cards were received within 4 working days of sample collection

# Outcomes

The number of screen positive babies detected in the year was as follows: phenylketonuria (9), congenital hypothyroidism (15), medium-chain acyl-CoA dehydrogenase deficiency (4), cystic fibrosis (8), sickle cell disorders (3).

# 3 Data

The data tables in this section outline the performance of the programme against the standards that have been set.

**Table 1:** The number of eligible births in Wales in the period April 2016 to March 2017, and the number of these babies tested.

| | Aneurin Bevan | Abertawe Bro Morgannwg | Betsi Cadwaladr | Cardiff & Vale | Cwm Taf | Hywel Dda | Powys | Wales |
|---|---|---|---|---|---|---|---|---|
| Births | 6467 | 5560 | 7076 | 5859 | 3317 | 3687 | 1212 | 33627 |
| Tested | 6451 | 5552 | 7050 | 5834 | 3315 | 3675 | 1209 | 33505 |
| % | 99.8 | 99.9 | 99.6 | 99.6 | 99.9 | 99.7 | 99.8 | 99.6 |

The Wales total includes some babies who do not map to a Health Board.

## 3.1 Standards

This table outlines the standards set by the screening programme to monitor performance.

**Table 2:** Programme performance standards

| NBSW Standards – Screening Programme | | | | |
|---|---|---|---|---|
| | **Objective** | **Criteria** | **Minimum Standard** | **Actual Value** |
| 1A | Completeness of Offer (Newborns) | The percentage of eligible newborn babies who have a notification of receipt of the bloodspot card in the laboratory by day 14 of life | 95% | 97.3% |
| 1B | Completeness of Offer (All) | Eligible babies (up to one year of age) who have a notification of receipt of the bloodspot card in the laboratory within 18 days of registration | 95% | 98.4% |
| 1C | Coverage (Newborns) | Eligible newborn babies who have a conclusive bloodspot screening result by day 17 of life | 95% | 94.0% |

| | | | | |
|---|---|---|---|---|
| 1D | Coverage (All) | Eligible babies (up to one year of age) who have a conclusive bloodspot screening result within 21 days of registration | 95% | 95.6% |
| 3A | Timely Collection of Sample (Day 5-8 of Life) | The first bloodspot sample should be taken between day 5 and day 8 of life (counting day of birth as day 0) | 95% | 98.0% |
| 3F | Timely Collection of Sample (Day 5 of Life) | The first bloodspot sample should be taken on day 5 of life (counting day of birth as day 0) | 90% | 75.3% |
| 3B | Timely Collection of Avoidable Repeat Samples | Repeat testing for insufficient/poor quality samples or incomplete/incorrect card information should be conducted within 3 calendar days of the request | 95% | 60.9% |
| 3C | Timely CHT Second Sample Collection for Pre-Term Babies | Pre-term babies with a second bloodspot card received in the laboratory which was taken at day 28 of life or on day of discharge | 95% | 2016-17 performance not reportable |
| 3D | Timely Second Sample Collection for Borderline TSH | Babies with a borderline TSH result who have a second bloodspot card for TSH received in the laboratory which was collected between 7 and 10 days after the initial borderline sample | 95% | 100% |
| 4A | Avoidable Repeat Rate | Repeat cards that are required because of poor quality bloodspots or incomplete/incorrect information recorded | <=2% | 5.5% |
| 4B | Poor Quality Repeat Rate | Repeat cards that are required because of poor quality bloodspots | <=2% | 4.2% |

| | | | | |
|---|---|---|---|---|
| 4C | NHS Number on Bloodspot Card | Bloodspot cards received in the laboratory that have a valid NHS number for the baby recorded | 100% | 99.1% |
| 5 | Timely Receipt of Card in Laboratory | Bloodspot cards received within 4 working days | 99% | 95.6% |
| 6A | Timely Processing of IMD and CHT Positive Samples | Clinical referral for IMD/CHT screen positive results initiated within 3 working days of sample receipt | 100% | 100% |
| 6B | Timely Processing of CF Positive Samples | Clinical referral for CF screen positive results initiated within 25 days of sample receipt | 95% | 100% |
| 6C | Timely Processing of SCD Positive Samples | Clinical referral for SCD screen positive results initiated within 42 days of sample receipt | 95% | 100% |
| 7A | Timely Clinical Care Receipt of IMD Positive Babies | First clinical appointment attendance for IMD screen positive results by day 17 of life | 100% | 100% |
| 7B | Timely Clinical Care Receipt of CHT Positive Babies | First clinical appointment attendance for CHT screen positive results by day 17 of life or initial borderline results followed by a positive by day 24 | 100% | 100% |
| 7C | Timely Clinical Care Receipt of CF Positive Babies | First clinical appointment attendance for CF screen positive results by day 28 of life | 95% | 75.0% |
| 7D | Timely Clinical Care Receipt of SCD Positive Babies | First clinical appointment attendance for SCD screen positive results by day 56 of life | 90% | 100% |

## 3.2 Completeness of Offer and Coverage

***Standard 1A**: 95% of newborn babies are offered screening*
***Standard 1B**: 95% of all babies are offered screening*
***Standard 1C**: 95% of newborn babies complete screening*
***Standard 1D**: 95% of all babies complete screening*

**Table 3:** Babies offered and completing newborn bloodspot screening

| Health Board | % Offered (Newborn) | % Offered (All) | % Coverage (Newborn) | % Coverage (All) |
|---|---|---|---|---|
| Abertawe Bro Morgannwg University Health Board | 97.5 | 98.2 | 94.5 | 96.1 |
| Aneurin Bevan University Health Board | 97.2 | 98.6 | 93.9 | 95.4 |
| Betsi Cadwaladr University Health Board | 97.3 | 98.7 | 92.9 | 95.1 |
| Cardiff and Vale University Health Board | 96.6 | 97.9 | 94.9 | 96.0 |
| Cwm Taf University Health Board | 98.3 | 99.3 | 95.0 | 96.6 |
| Hywel Dda University Health Board | 98.1 | 98.8 | 94.0 | 95.9 |
| Powys Teaching Health Board | 97.3 | 98.2 | 95.5 | 95.7 |
| **All Wales** | **97.3** | **98.4** | **94.0** | **95.6** |

The standards for offer of screening have been met by all health boards. Work to improve timeliness of sample collection and dispatch may have contributed to this.

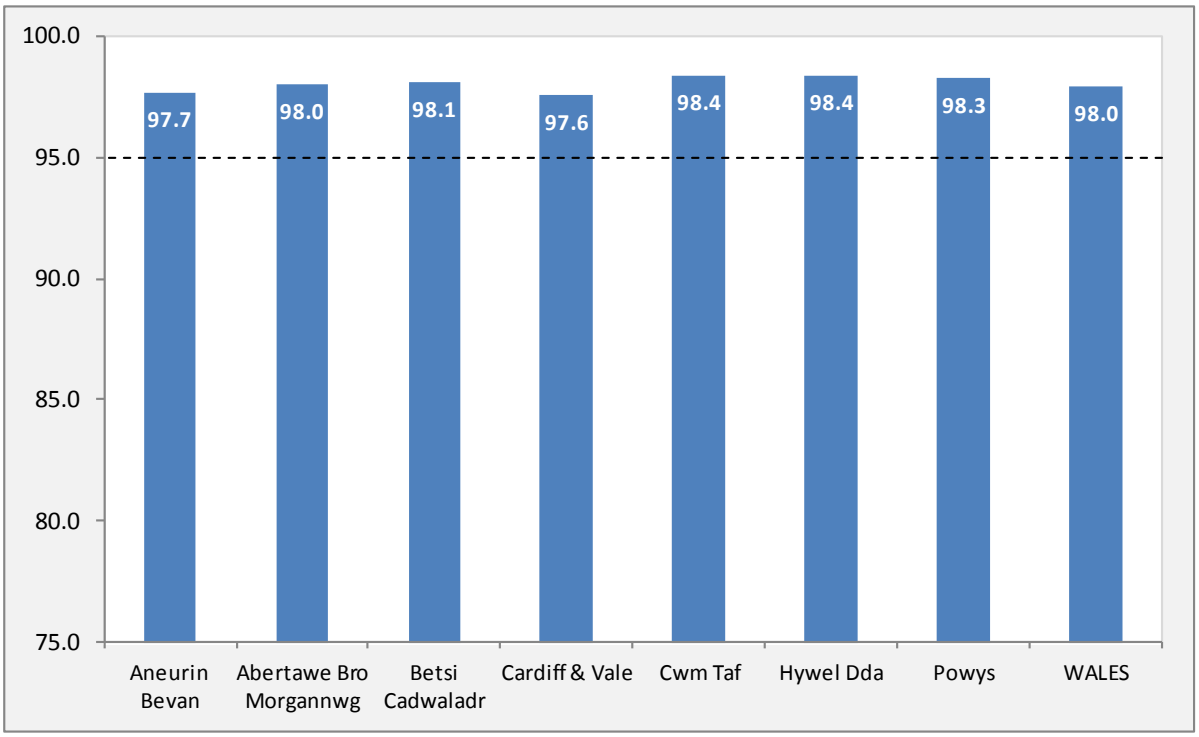
Across Wales, 94% of newborn babies had screening completed in the specified timeframe. The relatively high avoidable repeat rate and timeliness issues in collecting repeat samples have had an impact on performance in coverage, and addressing these problems is a high priority for the programme.

## 3.3 Timeliness of testing

***Standard 3A**: 95% of samples are taken between Day 5 – 8 of Life*
***Standard 3F**: 90% of samples are taken on Day 5 of Life*
***Standard 3B**: 95% of avoidable repeat samples are taken within 3 calendar days of request*
***Standard 3C**: 95% of CHT repeat samples for pre-terms babies are taken at day 28 of life or date of discharge*
***Standard 3D**: 95% Timely Second Sample Collection for Borderline TSH*

### Standard 3A

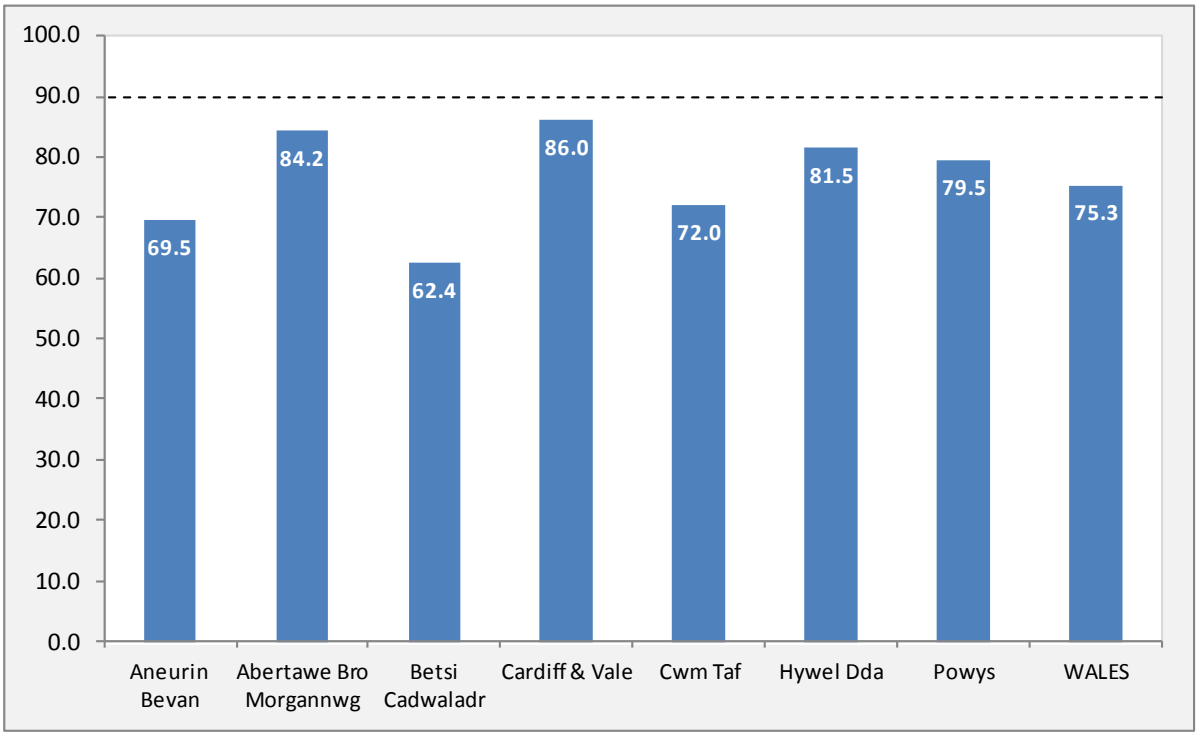
**Graph 1**: Timely Collection of Samples (Day 5 – 8 of Life)




In Wales, 98% of samples were taken between day 5 and day 8 of life. The programme is working with the health boards to improve timeliness of sample collection.

## Standard 3F

**Graph 2**: Timely Collection of Samples (Day 5 of Life)



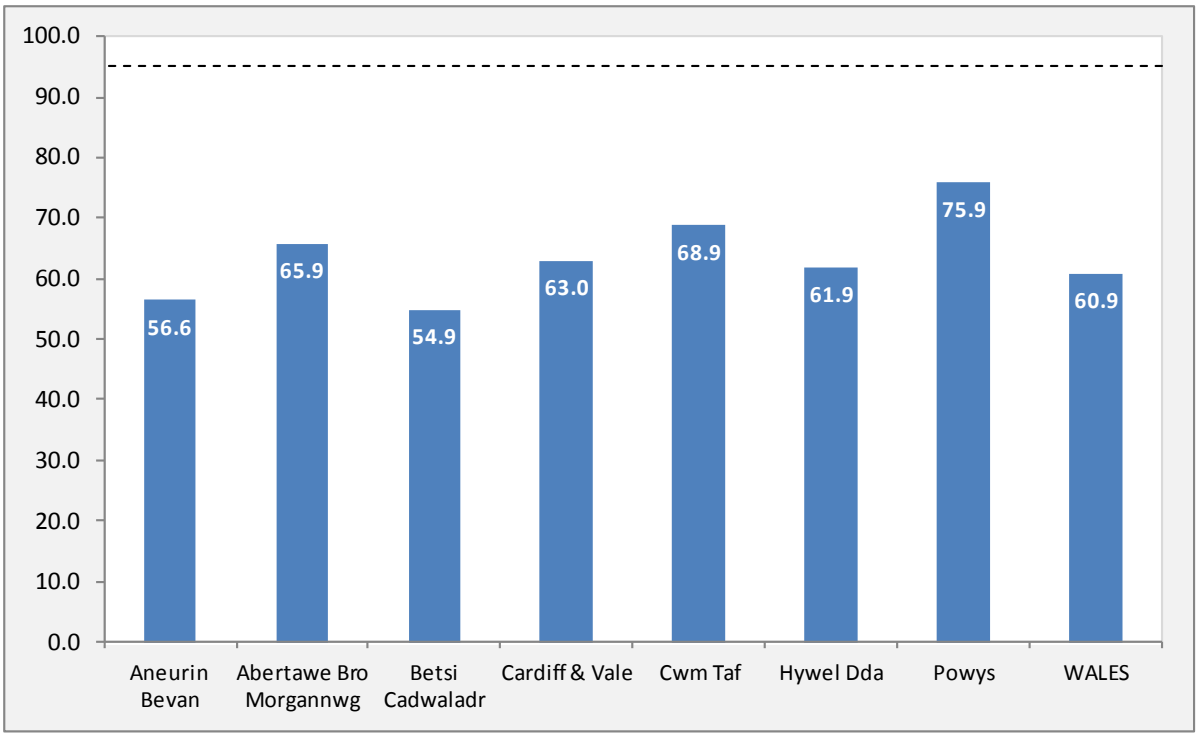

To monitor performance in timely collection of samples more closely, data is collected for sample collection at day 5 of life. Improving performance in this is a high priority for the programme to enable timely referral of screen positive babies to clinical care.

Across Wales, 75.3% of samples were taken at day 5 of life.

This information is periodically fed back to the health board governance leads and Heads of Midwifery. Action taken in the health boards to improve timeliness of sample collection has included the education of sample takers of the importance of taking samples on day 5, and changes in working practices to enable samples to be taken on weekends and bank holidays whenever possible. Work is continuing to improve performance in sample collection timeliness.

## Standard 3B

**Graph 3**: Timely Collection of Avoidable Repeat Samples




Across Wales, 60.9% avoidable repeat samples were taken within 3 calendar days of the request.

Requests for repeat samples were made by fax to the maternity or neonatal unit in the health board. This method was problematic and associated with delays in actioning these requests. To address this a new process has now been implemented.

From 27th June 2017 all requests are made by email to designated generic email addresses in the maternity units and neonatal units. The emails contain an image of the card, details of the sample problem and advice for the sample taker to improve performance. The health board governance lead is copied into the email to enable monitoring of sample taker performance within the health board and timely identification of training needs. It is anticipated that this will improve the avoidable repeat rate and performance in the timely collection of repeat samples over the next year.

## Standard 3C

Timely CHT Second Sample Collection for Pre-Term Babies

Pre-term babies should have a second sample taken for CHT testing on day 28 of life or earlier if they are to be discharged home.

**Table 4:** The actual day of testing for the total number of pre-term babies in the year.

| Day of life second CHT sample taken | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| <28* | 28 | 29 | 30 | 31 | 32 | 33 | 34 | 35 | >35 | Total |
| 56 | 91 | 36 | 22 | 9 | 7 | 3 | 8 | 8 | 19 | 259 |

* Performance by Health Board and all Wales has not been given for this standard as the IT systems were not able to capture this data during 2016-17. An IT systems change will allow accurate analysis of the 2017-18 births in the next report.

The programme is working with the neonatal units across Wales to improve performance in this standard as there are a high number of samples taken before day 28 for babies who have not been discharged.

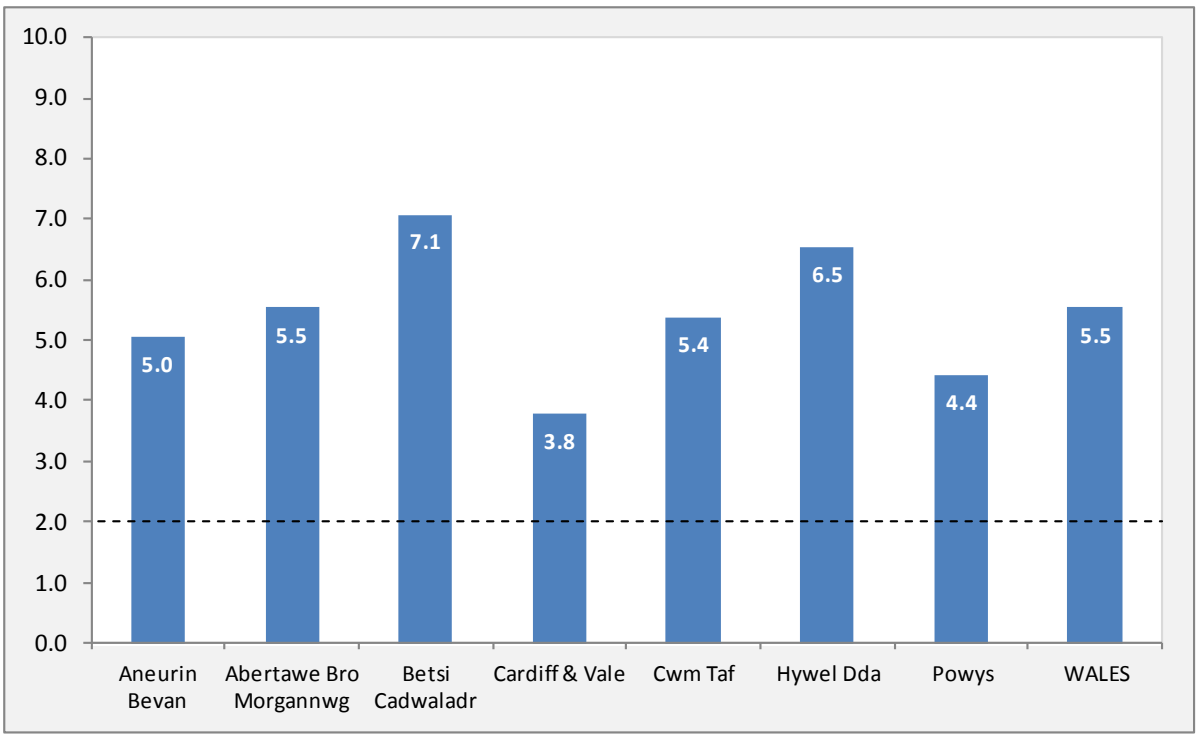
The BadgerNet Neonatal IT system used in the neonatal units across Wales has day of birth programmed as day 1. Samples taken on day 28 according to this system are therefore taken too early and need to be repeated. Health professionals have been alerted to this and the importance of counting day of birth as day 0 when working out when to take the sample.

## 3.4 Poor Quality Repeat Samples Required

***Standard 4A:*** *Avoidable Repeat Rates - <=2% repeat cards required because of poor quality bloodspots or incomplete/incorrect information recorded*
***Standard 4B:*** *Poor Quality Repeat rate - <=2% repeat cards required because of poor quality bloodspots*
***Standard 4C:*** *NHS Number on Bloodspot Card - 100% of Bloodspot cards received in the laboratory have a valid NHS number for the baby recorded*

**Standard 4A**

**Graph 4**: Avoidable Repeat Rate




The avoidable repeat rate in Wales is 5.5%. Achieving the standard of 2% is a high priority to avoid delays in the referral of babies and to avoid the other costs associated with repeating samples. The laboratory accepts samples according to the UK sample quality guidelines that were implemented in April 2015. These UK standards for bloodspot quality are based on work undertaken in the Wales Newborn Screening Laboratory to determine whether bloodspot quality has an effect on newborn bloodspot screening results.

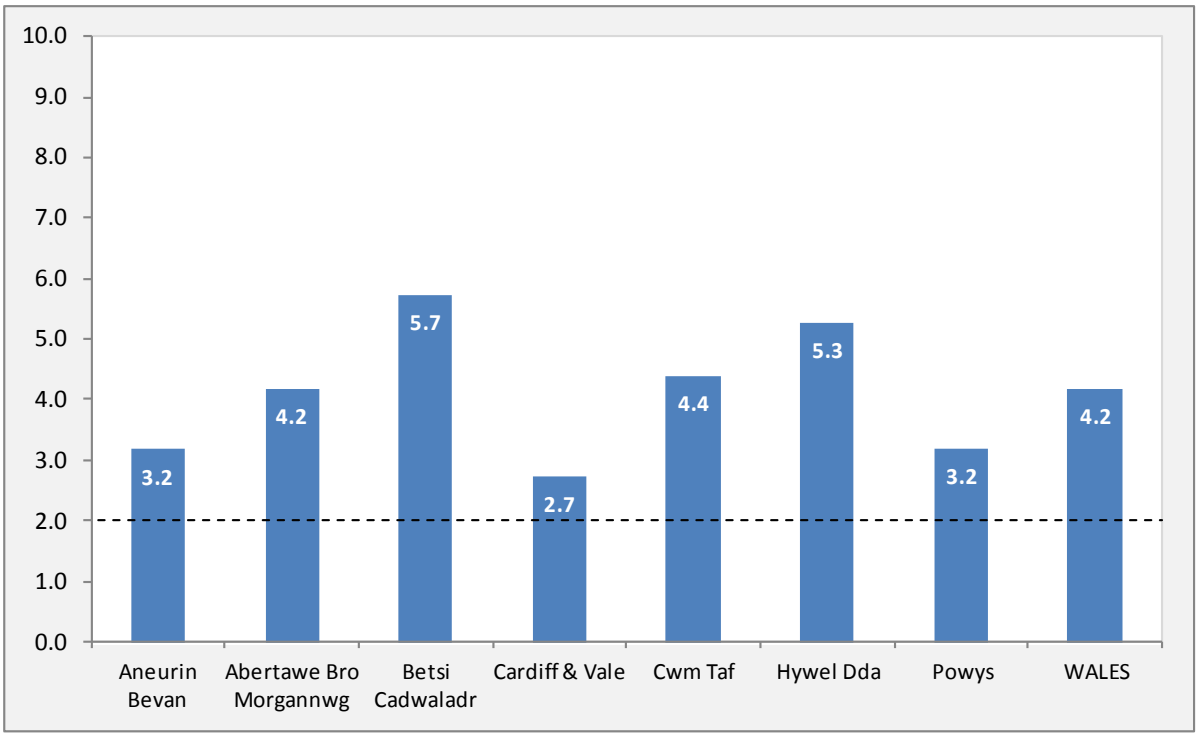
On a monthly basis the governance leads and Heads of Midwifery are sent reports detailing sample quality performance in their health board so that

appropriate action can be taken. Interventions in the health boards to improve sample quality have included the review of lancets used, the development of sample taker performance management processes and the review of education/training for sample takers.

Over the coming year NBSW will be providing training sessions for sample takers in the health boards which focus on improving sample quality. Training resources are also being developed for the different staff groups including health visitors and neonatal unit staff, to provide additional information relevant to their role in newborn bloodspot screening.

## Standard 4B

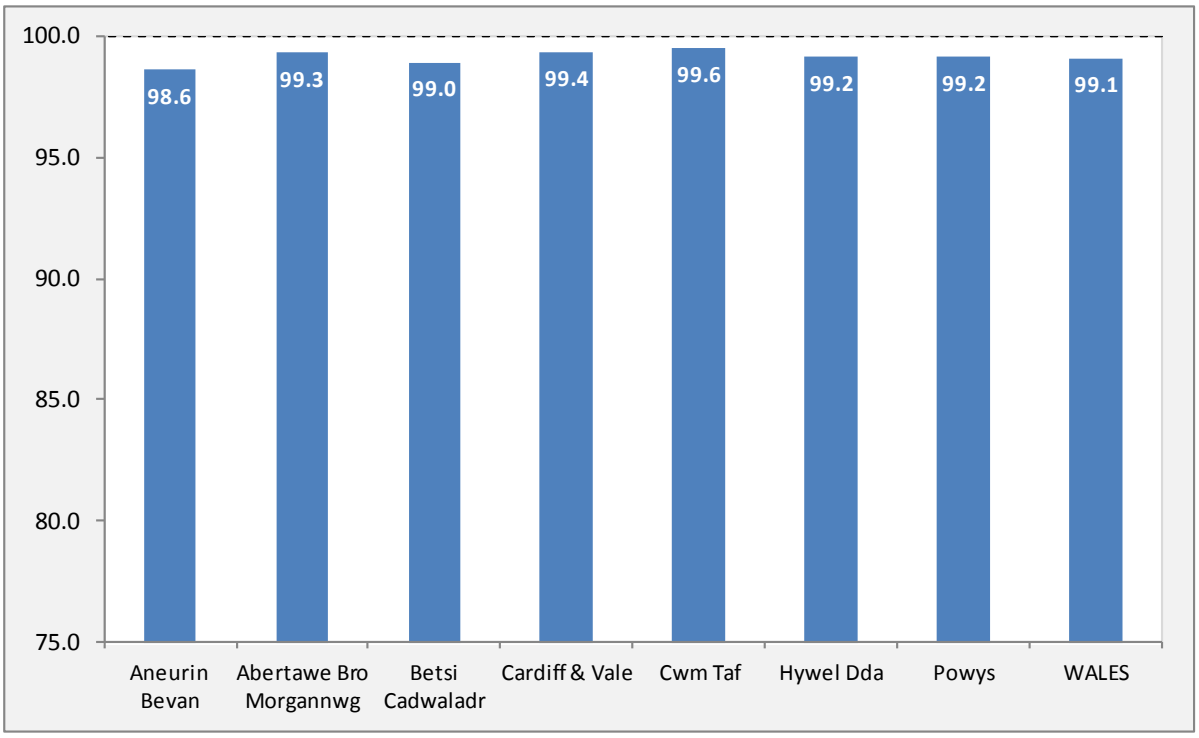
**Graph 5:** Poor Quality Repeat Rate




Across Wales, 4.2% samples required repeating due to poor quality bloodspots. As with the avoidable repeat rate, there is some variation in performance across Wales.

## Standard 4C

**Graph 6:** NHS Number on Bloodspot Card




The valid NHS number for the baby was recorded on 99.1% bloodspot cards received in the laboratory. Illegibility of NHS numbers, inaccuracy or mixing up the numbers with another baby are the main reasons for non compliance.

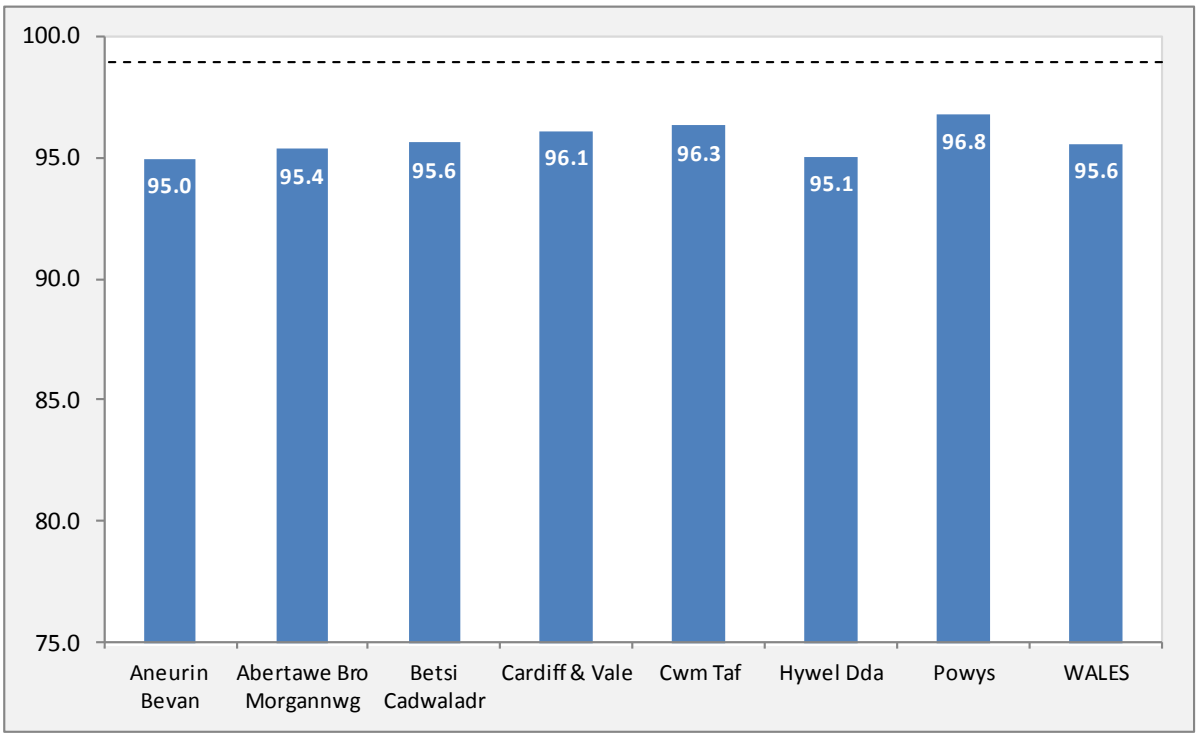
Sample takers who can not find, or would like to check the correct NHS number for a baby, are advised to contact the NBSWS failsafe administration teams who will be able to provide this if it has been generated. The use of bar coded NHS number labels for bloodspot screening is being scoped and it is planned barcoded labels will be introduced in Wales in 2018.

## 3.5 Timely Receipt of Card in Laboratory

***Standard 5:*** *Timely Receipt of Card in Laboratory - 99% of bloodspot cards received within 4 working days*

**Standard 5**

**Graph 7:** Timely Receipt of Card in Laboratory




Across Wales, 95.6% samples were received in the laboratory within 4 working days. The need to improve timeliness of receipt of samples in the laboratory has been highlighted to the sample takers. Measures to improve performance include ensuring the sample is posted in a Royal Mail post box and the avoidance of internal mail systems which have been associated with long delays in the despatch of samples.

The NBSW prepaid envelopes were reviewed and a revised version with NBSW logo in colour with 'Urgent delivery – post in a Royal Mail Box today' printed in bold red ink on the back of the envelope were introduced.

Holiday periods, particularly Christmas, have had a noticeable impact on the timeliness of receipt of samples in the laboratory. The programme liaises with Royal Mail to ensure any issues are resolved as soon as possible to minimise delays in samples reaching the laboratory.

## 3.6 Laboratory Processing and Referral

The current standard for timely processing of samples was met for all conditions. For the inherited metabolic disorders a total of 9 phenylketonuria (PKU) cases and 4 medium-chain acyl-CoA dehydrogenase deficiency (MCADD) cases were identified. Confirmation on receipt into clinical care was available for 13 out of the 13 cases, (100%). The average age of timely receipt into clinical care was day 12 of life (range 8-17 days). Of the 4 MCADD cases, one was a baby from England and was referred to the appropriate clinical team in England on day 11 of life. No cases of maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (HCU) were identified during 2016/17.

All 14 congenital hypothyroidism (CHT) cases were referred into clinical care in a timely manner. Confirmation on receipt into clinical care was available for only 9 out of the 14 cases (64%). Of the 14 CHT cases, 7 were screen positive on the initial screening sample and the average age at the first clinic appointment was day 12 of life (range 10-14 days). The remaining 7 cases were identified following an initial borderline result and the average age at the first clinic appointment was day 22 of life (range 21-23 days).

A total of 8 cystic fibrosis (CF) cases were identified, with the mean age of referral into clinical care being day 22 of life (range 18-30 days). Confirmation on receipt into clinical care was available for only 4 out of the 8 cases (50%). The average age at the first clinic appointment was day 24 of life (range 20-30 days). The case received into clinical care on day 30 of life was due to the sample being collected late on day 11 of life and the sample then received late into the laboratory on day 20 of life.

All three sickle cell disorder (SCD) cases were referred into clinical care by day 56 of life.

The collection of timeliness of appointment and diagnostic outcome data is a concern. The laboratory is reliant on the clinician, that received the screen positive referral, reporting the age at first appointment and the diagnostic results back to the screening laboratory.

# 4 Definitions

**Eligible babies (newborn)**

- A baby who is resident in Wales at day 5-8 of life
- A baby who is resident in Wales at day 5-8 of life, but is registered with an English GP
- A baby whose usual place of residence is outside Wales if they are under routine midwife care in Wales at day 5-8 of life

Babies who have been recorded as having died before the age of 5 days are not eligible.

**Eligible babies (all)**

- All babies up to one year of age who are resident in Wales
- A baby whose place of residence is outside Wales if they are under routine midwifery care in Wales at the time the newborn bloodspot test is due

Babies who have been recorded as having died before the age of 5 days are not eligible.

**Screen positive result**

Screening results are not 100% conclusive. Instead they provide presumptive results which are then confirmed using diagnostic tests. For example a screen positive result for congenital hypothyroidism (CHT) means that it is highly likely that the child has CHT, but that this must be confirmed by further tests. A screen positive result will be reported as 'suspected'.

**Screen negative result**

Screening results are not 100% conclusive. Instead they provide presumptive results which are then confirmed using diagnostic tests. For example a screen negative result for cystic fibrosis (CF) means that it is highly likely that the child does NOT have CF. This screen negative result is NOT usually confirmed using further tests, but it is assumed the child is not affected. A screen negative result will be reported as 'not suspected'.

**Conclusive result**

A conclusive result is any of the following; not suspected, suspected, not suspected other disorder or carrier. This includes any results that were tested by DNA for sickle cell disorders. For babies greater than 8 weeks of age, not tested for CF is also a conclusive result.

### Parent/guardian surveys

Parent/guardian surveys will be carried out to gather views of parents/guardians on their experience of newborn bloodspot screening. These surveys will also be used to monitor the performance of NBSW in the *informed consent* and *information provision* standards. The survey will include the views of those who accept screening and also of those who decline screening.

## The Conditions

### Congenital hypothyroidism (CHT)

Congenital hypothyroidism (CHT) is a condition where the baby's thyroid gland fails to develop or work properly and fails to make the thyroid hormone called thyroxine. Thyroxine is needed for normal growth and development.

### Cystic fibrosis (CF)

Cystic fibrosis (CF) is one of the UK's most common inherited life-limiting diseases. CF is a disease in which abnormal movement of salt and water into and out of cells causes a build up of thick, sticky mucous. This occurs particularly in the lungs and digestive system.

### Inherited metabolic disorders (IMDs):

- **Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)**
  MCADD is a rare inherited condition in which there is a deficiency in the enzyme medium-chain acyl-CoA dehydrogenase which is needed for the breakdown of certain stored fats (medium-chain fatty acids). Fatty acids are an important energy reserve during periods of poor calorie intake, prolonged periods between meals or during infections and sickness. In these situations people with MCADD have high levels of partially broken down fatty acids and low blood glucose concentrations which can result in a metabolic crisis.

- **Phenylketonuria (PKU)**
  Phenylketonuria (PKU) is a rare inherited condition that prevents the breakdown of a building block of protein, the amino acid phenylalanine. This then causes a build up of phenylalanine in the blood. The build up of phenylalanine is neurotoxic and harmful to the brain. Without treatmentPKU can cause severe, irreversible mental disability.

- **Maple syrup urine disease (MSUD)**
  Maple syrup urine disease (MSUD) is a rare inherited disorder that prevents the breakdown of some of the building blocks of protein, the amino acids leucine, isoleucine and valine in the blood. Without treatment MSUD can lead to serious problems including coma and permanent brain damage. The condition is named maple syrup urine disease because high levels of these amino acids can cause an unusual sweet smell in the urine and sweat.

- **Isovaleric acidaemia (IVA)**
  Isovaleric acidaemia (IVA) is a rare inherited disorder that prevents the breakdown of a building block of protein, the amino acid leucine. This then causes a harmful build up of a substance called isovaleric acid in the blood. Without treatment IVA can lead to serious problems including coma and permanent brain damage.

- **Glutaric aciduria type 1 (GA1)**
  Glutaric aciduria type 1 (GA1) is a rare inherited disorder that prevents the breakdown of certain building blocks of protein, in particular the amino acids lysine and tryptophan, and can cause a harmful build up of a substance called glutaric acid in the blood. Without treatment GA1 can lead to serious problems including coma and permanent brain damage.

- **Homocystinuria (HCU)**
  Homocystinuria (HCU) is a rare inherited disorder that prevents the breakdown of a building block of protein, the amino acid homocysteine. This then causes a harmful build up of homocysteine in the blood. Without early treatment this can lead to long term health problems including learning difficulties, eye problems, osteoporosis and blood clots or strokes.

- **Sickle cell disorders (SCD)**
  Sickle cell disorders (SCD) is a term that describes a group of conditions in which haemoglobin in red blood cells is abnormal in structure. This causes red blood cells to take up a shape like a crescent moon or farmer's sickle when de-oxygenated. Sickled red blood cells are not as flexible as normal red blood cells and can cause blockages within small blood vessels. Sickled red blood cells do not last as long as normal red blood cells and therefore their rate of production is increased.