Sgrinio Smotyn Gwaed Newydd-anedig Cymru

Newborn Bloodspot Screening Wales

GIG CYMRU NHS WALES

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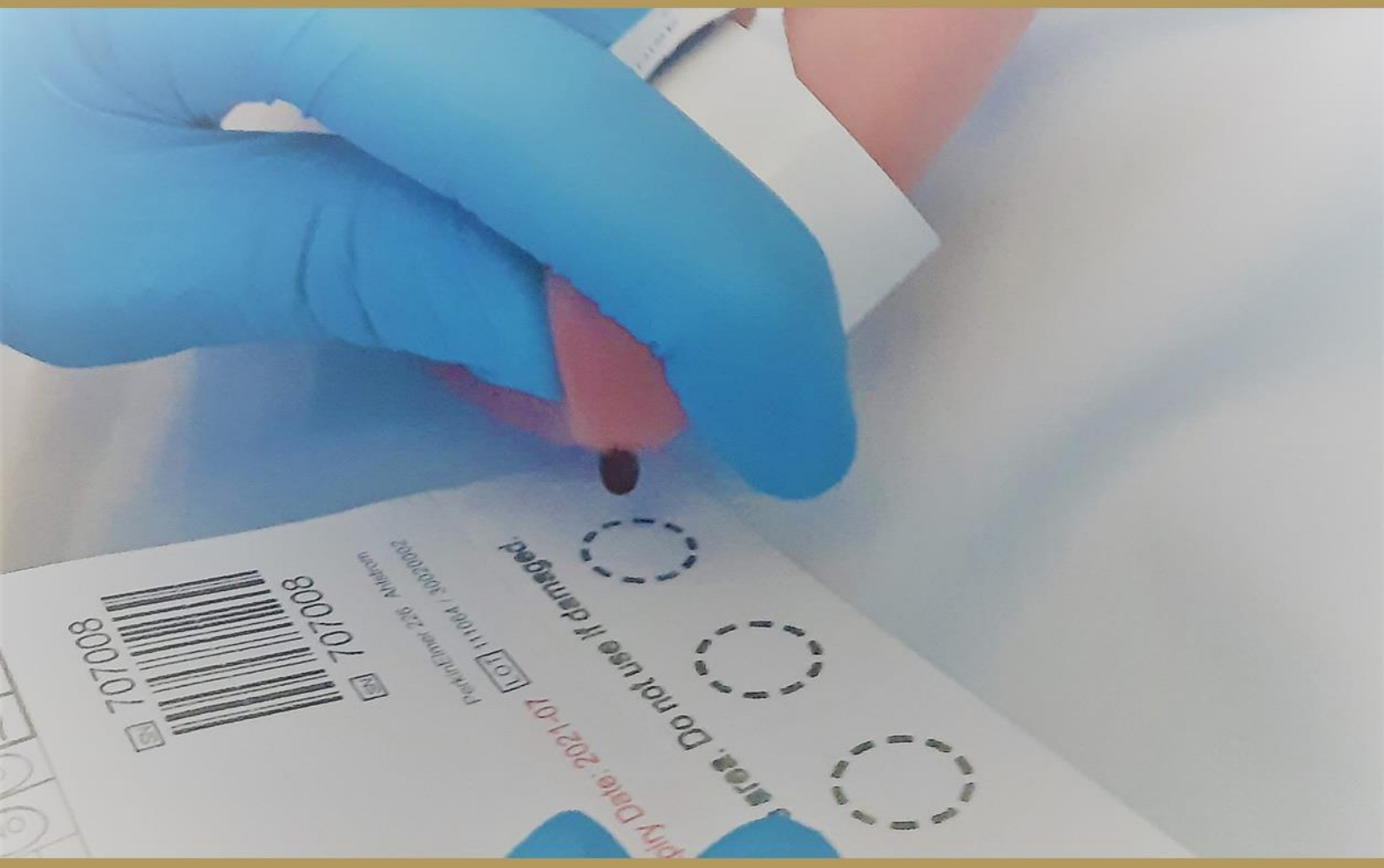
Public Health Wales

Newborn Bloodspot Screening Wales Annual Statistical Report 2020-21



Screening Division of Public Health Wales Publication

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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Twitter: @PublichealthW
Facebook: facebook.com/PublicHealthWales

The report is only available electronically from the screening programme and will be available on the website:
phw.nhs.wales/newborn-bloodspot-screening

This report is a detailed summary of information on work undertaken by Newborn Bloodspot Screening Wales for the financial year from April 2020 to the end of March 2021. Results are reported by health board where screening has been carried out. Further details are available on request.

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Rydym yn croesawu gohebiaeth a galwadau ffôn yn Gymraeg. Byddwn yn ateb gohebiaeth yn Gymraeg heb oedi / We welcome correspondence and phone calls in Welsh. We will respond to correspondence in Welsh without delay.

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. When dealing with data from small geographical areas we occasionally suppress numbers lower than five when the data is potentially sensitive.

This document is also available in Welsh.

Contents

Tables and graphs

1 Introduction

This report covers data for babies born between 1 April 2020 and 31 March 2021. 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 zero). 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 conditions that would benefit from early intervention to reduce mortality and/or morbidity.

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

- 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)
- Congenital hypothyroidism (CHT)
- Cystic fibrosis (CF)
- Sickle cell disorders (SCD)

Normal service provision was significantly affected throughout 2020/21 by the COVID-19 pandemic. In March 2020, the UK entered the first lockdown of the COVID-19 pandemic, which continued until June 2020. Restrictions remained in place throughout 2020/21 in Wales, including local lockdowns during September and October 2020, followed by a national firebreak lockdown until early November. A further national lockdown was in place from December 2020 to March 2021. Newborn bloodspot screening continued throughout this time and we are very grateful to colleagues in health boards, working under challenging circumstances, who prioritised taking good quality, timely samples.

To support safe service delivery for families and staff during the COVID-19 pandemic, NBSW programme staff worked closely with health board colleagues to adjust service protocols without affecting the quality of the screening results. As a contingency measure, from March 2020, the NBSW laboratory accepted samples taken at day four of life (counting day of birth

as day zero). This allowed the sample capture appointment to be combined with other planned postnatal visits, reducing the number of face to face appointments required by midwives and minimising potential exposure to COVID-19 by families and health board staff.

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. During 2020/21, NBSW was one of three programmes within Maternal and Child (MAC) Screening, which had an overall Programme Lead. There were two NBSW programme co-ordinators with administration support across the MAC programmes. The other two programmes were 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 the Screening Division of 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 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 collect samples for those babies who are inpatients in those units 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 Royal Mail) to the Wales Newborn Screening Laboratory in Cardiff for testing. The laboratory accepts samples according to the UK bloodspot quality guidelines for screening laboratories. 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. This is within 24 hours of the screening result.

The programme sends bloodspot screening results to parents by letter within six weeks of the sample being taken.

For babies who have a suspected result for any of the conditions, the results letter is sent via the baby's health visitor. The programme contacts the health visitor to inform them of the result after the baby has been received into clinical care. The baby's health visitor is then sent the results letter and information to enable an informed discussion of the results with the parents. The results for each baby are sent to the local Child Health Department electronically and are entered onto the Child Health System by health board staff.

More information is available at:
phw.nhs.wales/newborn-bloodspot-screening

2 Headline statistics

April 2020 to March 2021

- The number of eligible births across Wales was 28,612
- The number of babies tested was 28,445 (99.4%)

Screening

Completeness of offer and coverage by day 17 of life (eligible newborns)

- Completeness of offer – 96.9% of babies had a bloodspot card (for screening or decline) received in the laboratory by day 14 of life
- Coverage – 96.0% of babies had conclusive bloodspot screening results by day 17 of life

Timeliness of sample collection

- Timely collection of sample (day five-eight of life) – 79.0%
- Timely collection of sample (day five of life) – 63.2%

Performance against these standards was directly impacted by the COVID-19 contingency measure of allowing samples collected on day four of life to be accepted for testing.

- Timely collection of sample (day four-eight of life) – 98.0%
- Timely collection of sample (day four or five of life) – 82.3%

Avoidable repeat rate

- Avoidable repeat rate – 1.9%

Improving and maintaining performance in collecting good quality samples remains a high priority for the programme to avoid delays in the referral of babies with suspected conditions. This standard was recognised as being particularly important during 2020/21, due to the additional risks associated with providing avoidable face to face appointments during a pandemic.

NHS number on bloodspot card

- 99.8% bloodspot cards received in the laboratory had a valid NHS number for the baby recorded

Timely receipt of card in laboratory

- 94.3% of bloodspot cards were received within four working days of sample collection

Outcomes

The number of screen positive babies detected in the year was as follows: medium-chain acyl-CoA dehydrogenase deficiency (2), glutaric aciduria type 1 (1), congenital hypothyroidism (25), cystic fibrosis (15) and sickle cell disorders (1).

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 by health board area in the period April 2020 to March 2021 and the number of these babies tested.

	Aneurin Bevan	Swansea Bay	Betsi Cadwaladr	Cardiff & Vale	Cwm Taf	Hywel Dda	Powys	Wales
Births	5,658	3,254	6,063	4,774	4,263	3,125	1,056	28,612
Tested	5,634	3,222	6,033	4,744	4,245	3,105	1,050	28,445
%	99.6	99.0	99.5	99.4	99.6	99.4	99.4	99.4

The Wales total includes some babies who do not map to a health board.

There were 167 babies that were not tested in this period. Parents declined screening in 49 newborns and 47 babies that moved into Wales from outside the UK. Sadly, there were 46 deaths after day 5. A suspended status was recorded for 25 babies. These were movements into Wales from outside the UK where, at the time of reporting, the programme had been unable to contact the parents or the health visitor to obtain a definitive answer regarding consent for screening.

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**	**Variance from 2019-20**
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	99%	96.9%	no change

	Objective	Criteria	Minimum Standard	Actual Value	Variance from 2019-20
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	99%	98.9%	no change
1C	Coverage (Newborns)	Eligible newborn babies who have a conclusive bloodspot screening result by day 17 of life	95%	96.0%	+0.9
1D	Coverage (All)	Eligible babies (up to one year of age) who have a conclusive bloodspot screening result within 21 days of registration	95%	97.2%	+0.5
3A	Timely Collection of Sample (Day Five-Eight of Life)	The first bloodspot sample should be taken between day five and day eight of life (counting day of birth as day zero)	95%	79.0%	-18.5
3B	Timely Collection of Avoidable Repeat Samples	Repeat testing for insufficient/poor quality samples or incomplete/incorrect card information should be conducted within three calendar days of the request	95%	72.6%	-1.8
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%	50.2%	+1.1
3D	Timely Second Sample Collection for Borderline TSH (thyroid stimulating hormone)	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%	58.3%	-13.5
3J	Timely Collection of Sample (Day Four-Eight of Life)	COVID Contingency monitoring. The first bloodspot sample should be taken between day four and day eight of life (counting day of birth as day zero)	95%	98.0%	n/a Contingency monitoring standard

	Objective	Criteria	Minimum Standard	Actual Value	Variance from 2019-20
4A	Avoidable Repeat Rate	Repeat cards that are required because of poor quality bloodspots or incomplete/incorrect information recorded	<=2%	1.9%	-2.9 (improved performance)
4B	Poor Quality Repeat Rate	Repeat cards that are required because of poor quality bloodspots	<=2%	1.3%	-2.4 (improved performance)
4C	NHS Number on Bloodspot Card	Bloodspot cards received in the laboratory that have a valid NHS number for the baby recorded	100%	99.8%	+0.5
5	Timely Receipt of Card in Laboratory	Bloodspot cards received within four working days	99%	94.3%	-0.9
6A	Timely Processing of IMD and CHT Positive Samples	Clinical referral for IMD/CHT screen positive results initiated within three working days of sample receipt	100%	100%	no change
6B	Timely Processing of CF Positive Samples	Clinical referral for CF screen positive results initiated within 25 days of sample receipt	95%	100%	no change
6C	Timely Processing of SCD Positive Samples	Clinical referral for SCD screen positive results initiated within 42 days of sample receipt	95%	100%	no change
7A	Timely Clinical Care Receipt of IMD Positive Babies	First clinical appointment attendance for IMD screen positive results by day 14 of life	100%	66.6%	-33.4
7B	Timely Clinical Care Receipt of CHT Positive Babies	First clinical appointment attendance for CHT screen positive results by day 14 of life or initial borderline results followed by a positive by day 21	100%	76.0%	-4
7C	Timely Clinical Care Receipt of CF Positive Babies	First clinical appointment attendance for CF screen positive results by day 28 of life	95%	95.70%	+26.7

	Objective	Criteria	Minimum Standard	Actual Value	Variance from 2019-20
7D	Timely Clinical Care Receipt of SCD Positive Babies	First clinical appointment attendance for SCD screen positive results by day 90 of life	90%	100%	no change

3.2 Completeness of offer and coverage

***Standard 1A**: 99% of newborn babies are offered screening - notification of receipt of the bloodspot card in the laboratory by day 14 of life*
***Standard 1B**: 99% of all babies are offered screening - notification of receipt of the bloodspot card in the laboratory within 18 days of registration*
***Standard 1C**: 95% of newborn babies complete screening - a conclusive bloodspot screening result by day 17 of life (coverage)*
***Standard 1D**: 95% of all babies complete screening - a conclusive bloodspot screening result within 21 days of registration (coverage)*

Table 3: Babies offered and completing newborn bloodspot screening

Health Board	% Offered (Newborn)	% Offered (All)	% Coverage (Newborn)	% Coverage (All)
Aneurin Bevan UHB	96.8	98.9	96.1	97.3
Swansea Bay UHB	96.7	98.6	95.5	96.8
Betsi Cadwaladr UHB	96.6	98.9	95.9	97.1
Cardiff and Vale UHB	96.2	98.6	95.2	97.1
Cwm Taf UHB	98.0	99.2	96.7	97.5
Hywel Dda UHB	97.6	98.9	96.6	97.5
Powys Teaching Health Board	97.7	98.8	97.3	97.7
All Wales	96.9	98.9	96.0	97.2

The All Wales figures show that the standards for offer of screening have not been met, with performance very similar to last year. Work to improve timeliness of sample collection and dispatch is continuing. It is noted that extended postal delays were experienced at certain points during 2020/21 due to the pandemic, which impacted on sample arrival times within the laboratory. Additionally, in response to Royal Mail staffing pressures, daily postal deliveries to the laboratory were reduced from two to one for the entire year.

Both measures for coverage have improved this year and achievement of these standards has been maintained. Across Wales, 96.0% of newborn babies had screening completed in the specified timeframe, with improved performance for all health boards compared with the previous year. Continued improvements in sample quality have reduced the avoidable repeat rate and increased coverage performance.

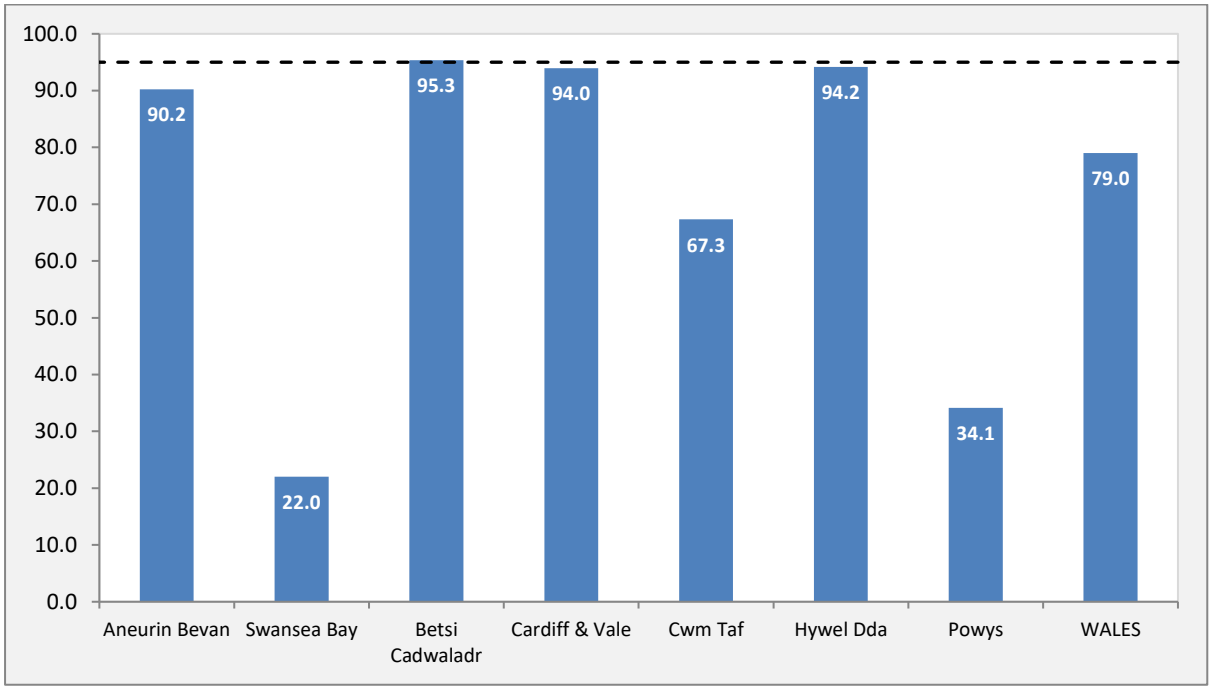
Achievement of the coverage standards is dependent on timely, high-quality sample capture and dispatch, effective sample transport arrangements and prompt laboratory testing. COVID-19 resilience plans were adopted by the laboratory in March 2020 and in place throughout 2020/21, with split shift working by staff to mitigate the potential for COVID-19 infections to disrupt delivery.

3.3 Timeliness of testing

Standard 3A*: 95% of samples are taken between day five-eight of life*
Standard 3B*: 95% of avoidable repeat samples are taken within three 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 collected between seven and ten days after initial borderline sample*
Standard 3J*: 95% of samples are taken between day four-eight of life (COVID-19 contingency measure)*

Standard 3A

Graph 1: Timely collection of samples (day five–eight of life)

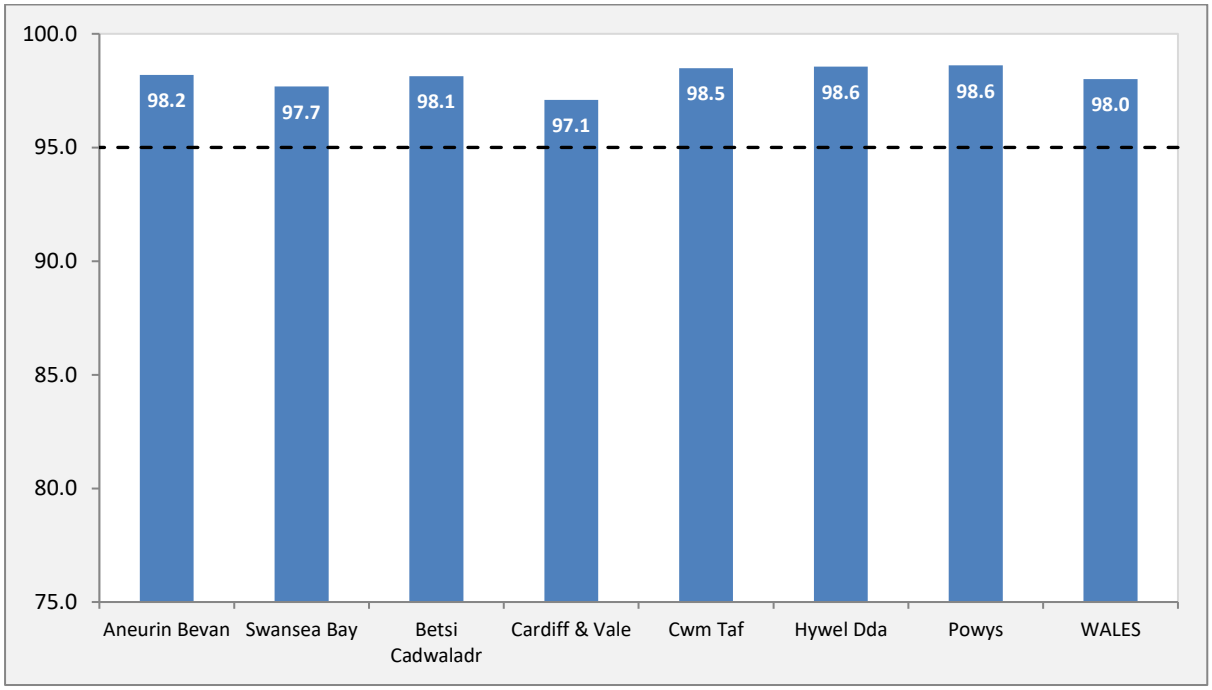


This standard has only been met in one health board area. Across Wales, 79.0% of samples were taken between day five and day eight of life, which is an 18.5% drop in performance against this standard compared to the previous year.

Performance against this standard has been directly affected by the contingency measure adopted throughout all of 2020/21. Whilst sample collection was strongly recommended for days five to eight of life, health boards were advised that collection on day four could be undertaken to avoid multiple face-to-face appointments where day four postnatal visits were planned. On an all-Wales basis, 98.0% of samples were collected between day four and eight of life, meaning that 19% of samples were collected on day four. The contingency standard 3J was introduced to monitor the position of samples taken between days four to eight, as below.

Standard 3J

Graph 2: Timely collection of samples (day four–eight of life)



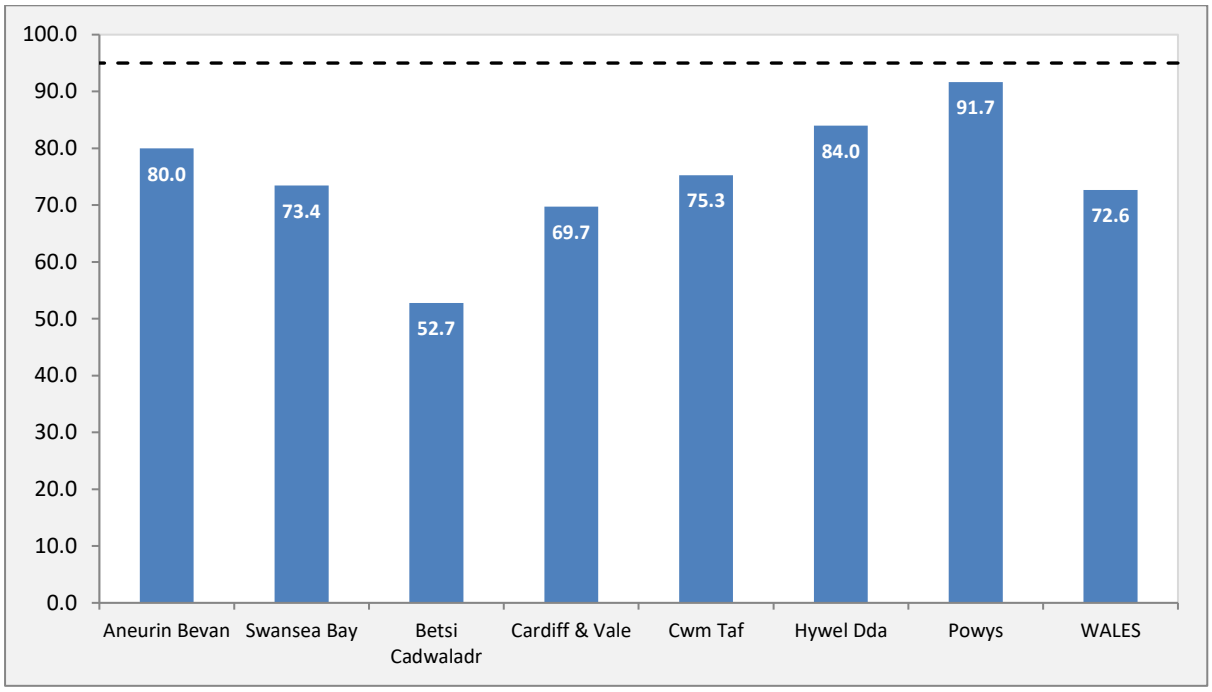
A key area of focus for the programme for a number of years has been improving timeliness of sample collection, with an emphasis on taking the sample on day five to enable earlier identification and referral of screen positive babies to clinical care. Internal monitoring processes routinely review the proportion of samples captured at day five of life to support focussed engagement with the health boards. The active use of the day four contingency measure has resulted in a greater proportion of samples captured on or before day five of life being processed, supporting early referral.

Across Wales, 63.4% of samples were taken at day five of life which is a drop of 9.8% compared with the previous year. However, 82.3% of samples were taken on days four and five as a result of the contingency measure, mitigating the clinical impact.

Performance data for sample collection timeliness is fed back quarterly to the health board governance leads and Heads of Midwifery. The programme continues to work with the health boards to further improve timeliness of sample collection.

Standard 3B

Graph 3: Timely collection of avoidable repeat samples



Across Wales, 72.6% of avoidable repeat samples were taken within three calendar days of the request. The standard has not been met, and performance dropped by 1.8% compared with 2019/20. There was some variation in performance across the health boards.

All requests for repeat samples are emailed to designated generic email addresses in the maternity and neonatal units. Regular reviews of the process take place to support improvement. The programme continues to work closely with the Newborn Screening Laboratory to identify any factors that may impact on the timely collection of repeat samples.

Work also continues with the health boards to improve the timeliness of collecting the repeat samples. This has included sample taker education and the review of processes to ensure that they are robust for accessing the repeat request emails daily and repeating the samples without delay. A contributing factor to the under performance of this standard was the flexible use of staff in sites and services that they would not normally work within and staff shortages caused by the COVID pandemic.

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*
29	89	46	20	8	11	3	6	4	17	233

* Across Wales 2 (0.9%) CHT second samples were taken too early, meaning before day 28 but not discharged. This compares to 12 samples (5.3%) in 2019-20. CHT second samples that are taken too early are not accepted for testing and require a repeat sample to be taken.

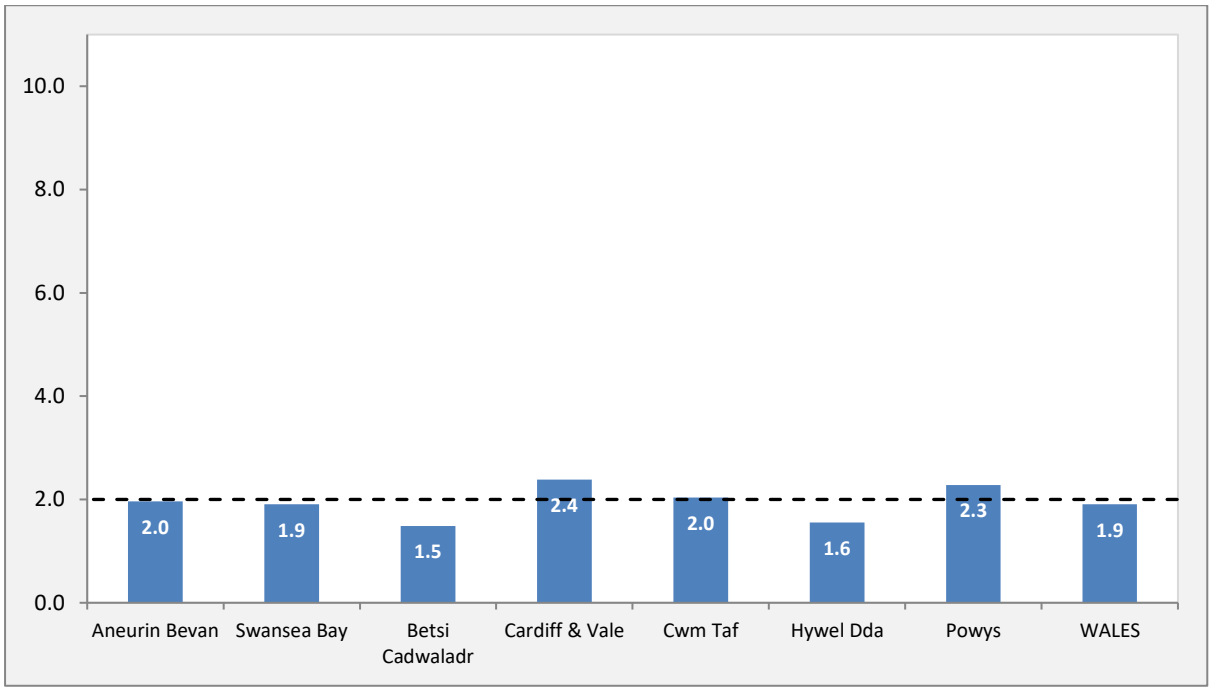
The programme continues to work closely with the neonatal units across Wales to improve performance so that CHT second samples are taken at the correct time.

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 1.9%, a considerable improvement and decrease of 2.9 percentage points compared with the previous year. An improvement has been seen in every health board. Sustaining this sample quality improvement and continuing to achieve the standard of ≤2% is a high priority for the programme to avoid delays in the referral of babies and to avoid the other costs associated with repeating samples.

The health boards and the programme have continued to work in collaboration to further improve and sustain this standard. Despite the COVID-19 pandemic, colleagues in the health boards worked tirelessly to ensure that good quality samples were captured first time.

During this reporting period, the programme developed two E-learning packages, 'Introduction to Newborn Bloodspot Screening' and 'Taking a Good Quality Newborn Bloodspot Screening Sample'. These packages were available for sample takers to complete from March 2021 to support them in taking timely good quality samples.

The programme reviewed the two training films 'Newborn bloodspot screening – the laboratory processes' and 'Newborn bloodspot screening in neonatal units'. Feedback had been received from sample takers regarding the length of the films and the difficulty in having time to view them when

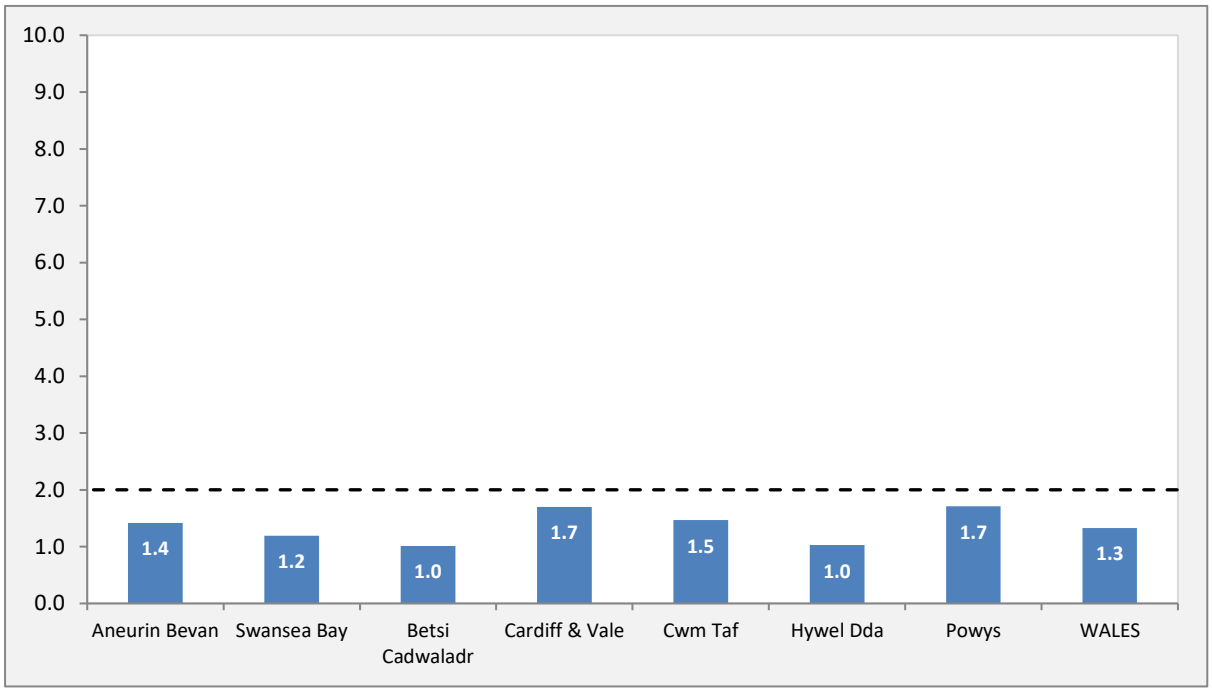
busy. In view of this the films were condensed with the viewing time reduced to five minutes and nine minutes respectively.

The programme continued to offer training to student midwives and this was undertaken virtually due to the pandemic. This training was well received. The virtual training sessions were also extended to newly qualified midwives, to sample takers in the paediatric and neonatal units and in the health visiting service. These training sessions play an important part in ensuring that sample takers have a good understanding of the newborn screening pathway and how to take a good quality sample before they commence taking samples.

Each month the governance leads and heads of midwifery are sent a monthly sample quality performance report for their health board to enable monitoring and appropriate action to be taken. The governance leads are also copied into the emails sent from the laboratory requesting repeat samples so are alerted to quality issues requiring more immediate attention.

Standard 4B

Graph 5: Poor quality repeat rate

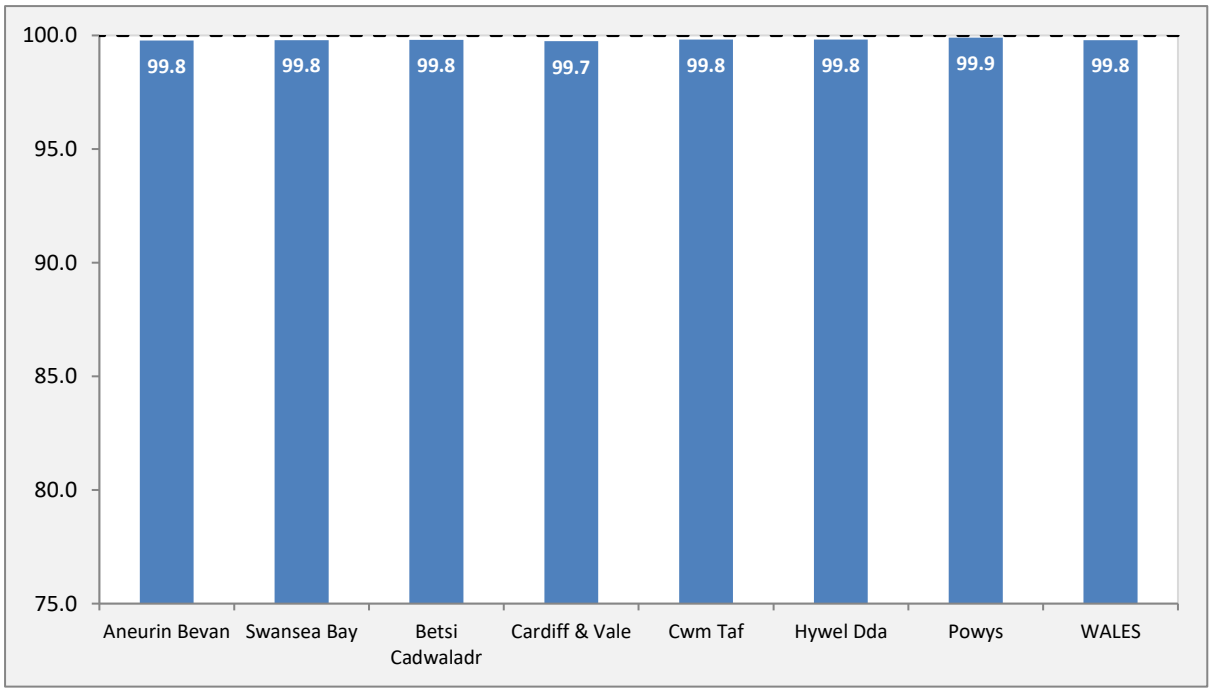


Across Wales, 1.3% of samples required repeating due to poor quality bloodspots, which is a performance improvement of 2.4 percentage points

compared with the previous year. For the first time, in 2020/21, all health boards achieved this standard. Sustaining this improvement and maintaining the standard of ≤2% is a high priority for the programme.

Standard 4C

Graph 6: NHS number on bloodspot card

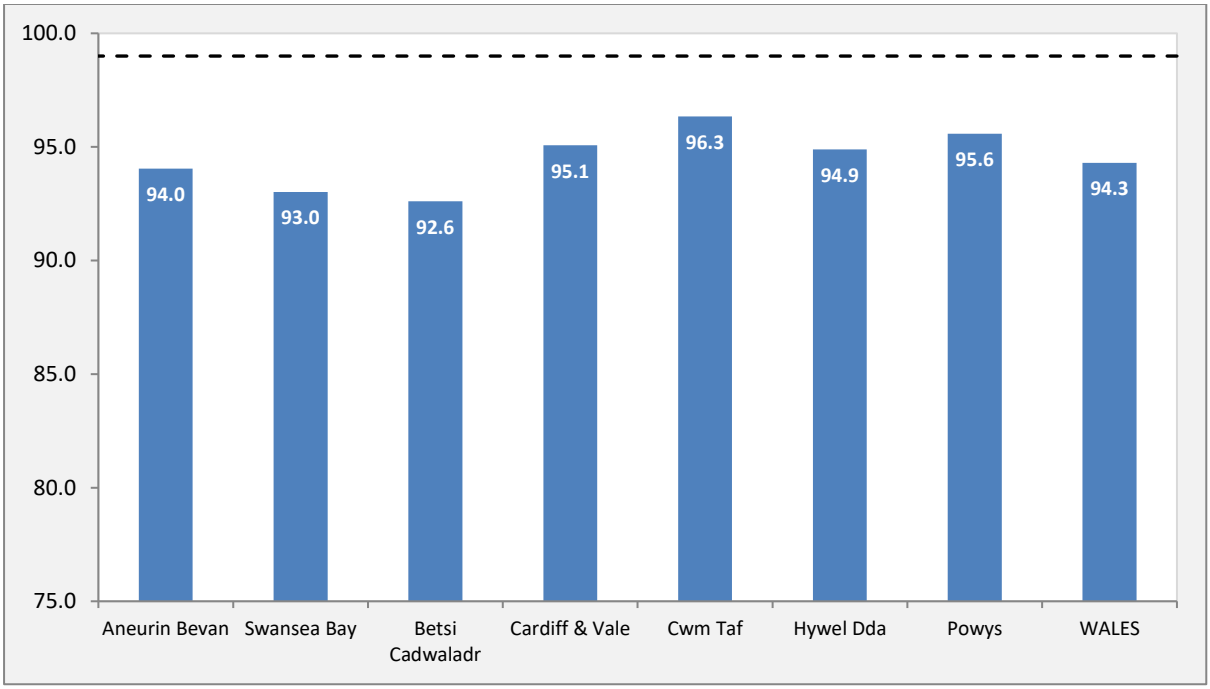


A valid NHS number for the baby was recorded on 99.8% of bloodspot cards received in the laboratory, a 0.5 point improvement on the previous year. Illegibility of NHS numbers, inaccuracy or mixing up the numbers with another baby are the main reasons for non-compliance. Work continues with the health boards to improve performance still in this standard.

3.5 Timely receipt of card in laboratory

Standard 5: *timely receipt of card in laboratory - 99% of bloodspot cards received within four working days*

Graph 7: Timely receipt of card in laboratory



Across Wales, 94.3% of samples were received in the laboratory within four working days. This is a decrease of 0.9% compared with the previous year. Improving performance in this standard remains a high priority to enable timely referral of screen positive babies into clinical care.

To monitor performance more closely, additional data is collated for receipt of samples received within three working days of the sample being taken. This performance data is fed back quarterly to the health board governance leads and heads of midwifery.

Work continues with the health boards to reiterate the importance of posting samples into a Royal Mail post box on the day they are taken and avoiding the use of internal mail systems, which have been associated with delays in the receipt of samples.

Close engagement with Royal Mail has continued to minimise delays in samples reaching the laboratory. Postal services were subject to delays caused by the COVID-19 pandemic during 2020/21, including reduced delivery frequency to the laboratory throughout the year. Delays in sample

receipt are routinely experienced during the Christmas period, with these being exacerbated by pandemic-related delays during December 2020.

3.6 Laboratory processing and referral

The current standard for timely processing of samples in the laboratory was met for all conditions. For the inherited metabolic disorders; a total of 3 infants were referred for follow up. Of these, 1 infant was confirmed to have medium-chain acyl-CoA dehydrogenase deficiency (MCADD), the other 2 infants were false positive cases (one false positive for MCADD due to prematurity and systemic illness and one false positive for Glutaric Aciduria; due to renal failure). Confirmation of receipt into clinical care was available for all 3 cases (100%). The MCADD case was received into care on day 9 of life, the false positive MCADD by day 23 of life (baby on NICU and day 5 sample not collected), and the false positive GA1 case on day 7 of life.

Twenty-five cases with increased TSH concentrations were identified and referred for follow up. Of these, 15 were identified as having a raised TSH ≥20mU/L on the initial sample and 10 cases were identified with a borderline raised TSH (≥8 but <20mU/L) on the initial sample with a positive repeat sample. All cases identified as having a raised TSH were referred into clinical care in a timely manner. Confirmation of the first clinic appointment was available for all 25 cases (100%). Of the 15 CHT cases with a raised TSH ≥20mU/L on the initial screening sample, 11 (73.3%) were received into care by day 14 of life. The average age at the first clinic appointment was day 12 of life (range 8-23 days). Of the 10 cases identified following an initial borderline test result, 8 (80%) cases were received into care by day 21 of life (the average age at the first clinic appointment was day 17 of life, (range 10-24 days)). Feedback as to whether or not patients were commenced on thyroxine treatment was poor with 11/25 (44%) of cases being reported. A total of 60 second samples were collected for borderline TSH results. The second sample collection for borderline TSH results should be collected 7 to 10 days after the initial sample was taken. The number of samples collected at the appropriate time was 35 (58.3%) (standard = 95%), with 16 (26.7%) of samples being collected on or after day 11 (range 11-24 days). A few samples, n=9 (15%) were collected before day 7.

For CF screening, a total of 15 cases were referred for follow-up. Of the 15 cases referred; 6 had 2 CF pathogenic variants, 7 had 1 pathogenic variant and 2 cases had an IRT concentration ≥170ng/ml but with no pathogenic variants detected. A total of 7 cases (6 cases with 2 variants and 1 case with

1 variant) were confirmed as having CF, following a positive sweat test. Confirmation of receipt into clinical care was available for 14 out of the 15 cases (93.3%). Of these 14 cases, 12 (85.7%) cases were seen at the first clinic appointment by day 28 of life (standard = 95%). The average age at the first clinic appointment was 24 days (range 19-33).

1 case of sickle cell disorder (SCD) was detected and this case was referred into clinical care in a timely manner. This case was also identified via the Wales antenatal screening programme.

4 Definitions

Eligible babies (newborn)

- A baby who is resident in Wales at day five-eight of life
- A baby who is resident in Wales at day five-eight 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 five-eight of life

Babies who have been recorded as having died before the age of five 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. A screen positive result is a result which shows that the child is likely to have the condition for which they are screened. Sometimes people will say that the child is affected. Positive screening results 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 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. A screen negative result is a result which suggests that the child does not have the condition for which they are being screened. Sometimes people will say that the result is 'normal'. 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. Without thyroxine, babies do not grow properly and can develop permanent, serious physical problems and learning disabilities.

Babies with CHT can be treated early with thyroxine tablets and this will allow them to develop normally.

CHT has been screened for in Wales since 1981.

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. Babies with CF may not gain weight well, have frequent chest infections and a limited life span.

If babies with CF are treated early with a high-energy diet, medicines and physiotherapy, they may live longer, healthier lives.

CF has been screened for in Wales since December 1996.

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). This makes it difficult for the body to break down fatty acids and produce energy, and can cause sudden death in infants. 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. Most of the time children are well, but an infection or relatively long period without food upsets their metabolism causing coma and sometimes death.

  Treatment involves ensuring that children do not go for long periods without food and special management if they do get an infection. Periods of not eating can safely get longer as the child grows.

  MCADD has been screened for in Wales since June 2012.

- **Phenylketonuria (PKU)**

  Phenylketonuria (PKU) is a rare inherited condition that prevents the breakdown of a building block of protein, the amino acid phenylalanine. For people with PKU, eating normal amounts of protein can cause a harmful build-up of phenylalanine in the blood. The build-up of phenylalanine is neurotoxic and harmful to the brain. Without treatment PKU can cause severe, irreversible mental disability.

  If identified early, the child can be put on a restricted-protein diet with supplements and the brain can develop normally.

  PKU has been screened for in Wales since 1970.

- **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. For people with MSUD, eating normal amounts of protein can cause a harmful build-up of these amino acids in the blood. Many babies with MSUD

become unwell when they are a few days old. Without treatment, this leads to a coma and permanent brain damage. In older children a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. As in babies, this can lead to a coma unless treated correctly.

MSUD can be treated with a protein-restricted diet. A different regime is required when the child is ill, and they may need to be hospitalised. 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.

MSUD has been screened for in Wales since January 2015.

- **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. Children with IVA can become severely unwell. Without treatment, this can lead to a coma and permanent brain damage. Some babies with IVA have problems within a few days of birth; other children become unwell at a few months or years of age, maybe during a minor illness, such as a chest infection or a tummy upset.

IVA can be treated with a protein-restricted diet and carnitine and glycine. A different regimen is required when the child is ill, and they may need to be hospitalised.

IVA has been screened for in Wales since January 2015.

- **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. For people with GA1, eating normal amounts of protein can cause harmful substances to build up in the blood and urine. In children with GA1, a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. Without treatment, the child can go into a coma. Though most children come out of the coma, they usually have brain damage that affects their ability to control their muscles and

movements. This means that they may be unable to sit, walk, talk or swallow.

GA1 can be treated with a protein-restricted diet and carnitine. A different regimen is required when the child is ill, and they may need to be hospitalised.

GA1 has been screened for in Wales since January 2015.

- **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 and eye problems, osteoporosis and blood clots or strokes.

  HCU can be treated with a protein-restricted diet and extra supplements and medicines.

  HCU has been screened for in Wales since January 2015.

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. Babies who have these conditions will need specialist care throughout their lives. People with SCD can have attacks of severe pain, get serious, life threatening infections and are usually anaemic (their bodies have difficulty carrying oxygen).

Babies with SCD can receive early treatment, including immunisations and antibiotics, which, along with support from their parents, will help reduce the chance of serious illness and allow the child to live a healthier life.

SCD has been screened for in Wales since 2013.

5 Production team

The production team for this report are all employed within Public Health Wales and are listed below.

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