Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

GIG CYMRU NHS WALES
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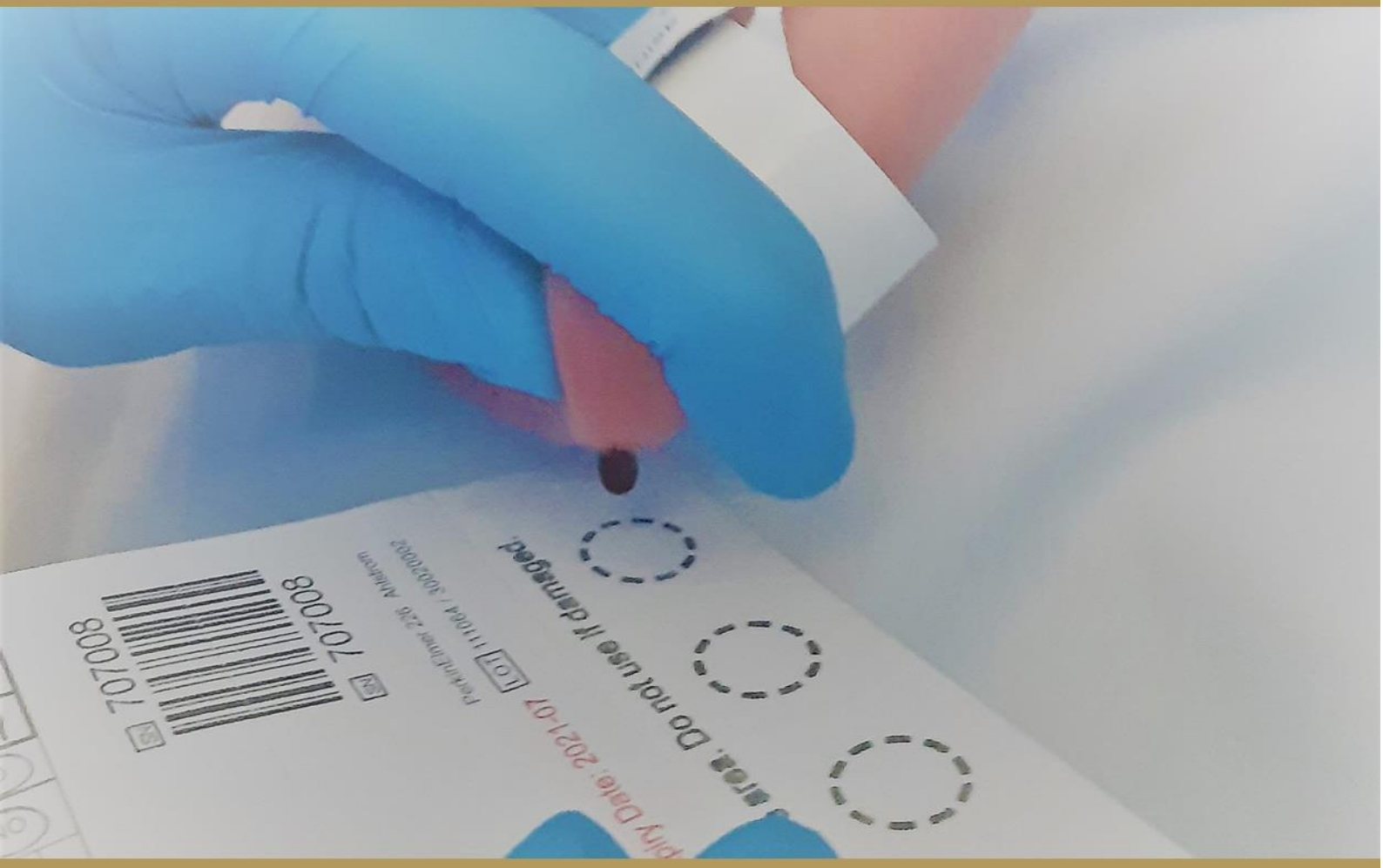
Public Health Wales

Newborn Bloodspot Screening Wales Annual Statistical Report 2021-22



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

This report is a detailed summary of information on work undertaken by Newborn Bloodspot Screening Wales for the financial year from April 2021 to the end of March 2022. 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 supress 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 2021 and 31 March 2022. 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 (NBSW) 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)

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 by the NHS;
- 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 NBSW programme. Until December 2021, NBSW was one of three programmes within Maternal and Child (MAC) Screening, which shared an overall Programme Lead. MAC Screening incorporated NBSW, Antenatal Screening Wales (ASW) and Newborn Hearing Screening Wales (NBHSW). There are two NBSW programme co-ordinators with administration support across the MAC programmes. From December 2021, the MAC function split, with one Head of Programme leading ASW, and another responsible for both NBSW and 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) is a computer records system 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 NBHSW 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 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 units at day five of life. Health visitors take responsibility for offering and arranging sample collection for older 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.

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

1.4 Revised service standards

The programme undertook a comprehensive review of service standards during 2021-22. Changes were made to indicator definitions to support clarity and to standard requirements for closer alignment with other UK nations or to adjust the minimum standard threshold to improve quality. Additionally, four standards were removed, either because other standard changes meant that they were no longer required, or because they were used for monitoring purposes only and no longer beneficial.

The changes came into effect in November 2021, with NBSW performance against both the old and new standards detailed in this report. Where the definition or standard threshold has changed, this is indicated to support comparison with previous programme performance.

2 Headline statistics

April 2021 to March 2022

- The number of eligible births across Wales was 29,716
- The number of babies tested was 29,587 (99.6%)

Screening

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

- Completeness of offer – 97.2% of babies had a bloodspot card (for screening or decline) received in the laboratory by day 14 of life
- Coverage – 95.9% 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) – 74.1%
- Timely collection of sample (day five of life) – 58.0%

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) – 97.8%
- Timely collection of sample (day four or five of life) – 81.7%

Avoidable repeat rate

- Avoidable repeat rate – 2.4%

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

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

- 94.9% 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: phenylketonuria (1), medium-chain acyl-CoA dehydrogenase deficiency (4), isovaleric acidaemia (2), congenital hypothyroidism (17), cystic fibrosis (17) 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 in the period April 2021 to March 2022 and the number of these babies tested.

	Aneurin Bevan	Swansea Bay	Betsi Cadwaladr	Cardiff & Vale	Cwm Taf	Hywel Dda	Powys	Wales
Births	5,973	3,432	6,194	5,081	4,380	3,123	1,111	29,716
Tested	5,948	3,405	6,174	5,064	4,369	3,102	1,105	29,587
%	99.6	99.2	99.7	99.7	99.7	99.3	99.5	99.6

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

There were 129 babies that were not tested in this period. Parents declined screening in 35 newborns and 33 babies that moved into Wales from outside the UK. Sadly there were 26 deaths after day 5. A suspended status was recorded for 35 babies. These were movements into Wales from outside the UK for whom, 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 2020-21**
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%	97.2%	+0.3

	Objective	Criteria	Minimum Standard	Actual Value	Variance from 2020-21
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%	98.7%	-0.2
1C	Coverage (Newborns)	Eligible newborn babies who have a conclusive bloodspot screening result by day 17 of life	95%	95.9%	-0.1
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%	no change
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%	74.1%	-4.9
	Timely Collection of Sample (Day Five-Six of Life)	The first bloodspot sample should be taken between day five and day six of life (counting day of birth as day zero)	95%	71.4%	N/A (new standard)
3J	Timely Collection of Sample (Day Four-Six of Life)	The first bloodspot sample should be taken between day four and day six of life (counting day of birth as day zero)	95%	97.8%	-0.2 (contingency monitoring standard)
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%	72.4%	-0.2
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%	59.7%	+9.5

	Objective	Criteria	Minimum Standard	Actual Value	Variance from 2020-21
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%	71.4%	-0.4
4A	Avoidable Repeat Rate	Repeat cards that are required because of poor quality bloodspots or incomplete/incorrect information recorded	<=2%	2.4%	+0.5 (reduced performance)
4B	Poor Quality Repeat Rate	Repeat cards that are required because of poor quality bloodspots	<=2%	1.7%	+0.4 (reduced performance)
4C	NHS Number on Bloodspot Card	Bloodspot cards received in the laboratory that have a valid NHS number for the baby recorded	100% (original) 99% (new standard)	99.8%	no change
5	Timely Receipt of Card in Laboratory	Bloodspot cards received within four working days	99%	94.9%	+0.6
	Timely Receipt of Card in Laboratory	Bloodspot cards received within three working days	95%	85.1%	N/A (new standard)
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%	87.5%	+20.9

	Objective	Criteria	Minimum Standard	Actual Value	Variance from 2020-21
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%	94.1%	+18.1
7C	Timely Clinical Care Receipt of CF Positive Babies	First clinical appointment attendance for CF screen positive results by day 28 of life	95%	76.5%	-19.2
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	97.3	98.8	96.4	97.6
Swansea Bay UHB	96.8	98.1	95.4	96.8
Betsi Cadwaladr UHB	96.8	98.9	95.4	96.8
Cardiff and Vale UHB	96.3	98.4	95.2	97.0
Cwm Taf Morgannwg UHB	98.2	99.1	96.5	97.6
Hywel Dda UHB	97.9	98.9	96.7	97.5
Powys Teaching Health Board	97.8	98.9	96.0	97.2
All Wales	97.2	98.7	95.9	97.2

The All Wales figures show that the standards for offer of screening have not been met, with performance similar to the previous year. Work to improve timeliness of sample collection and dispatch continues.

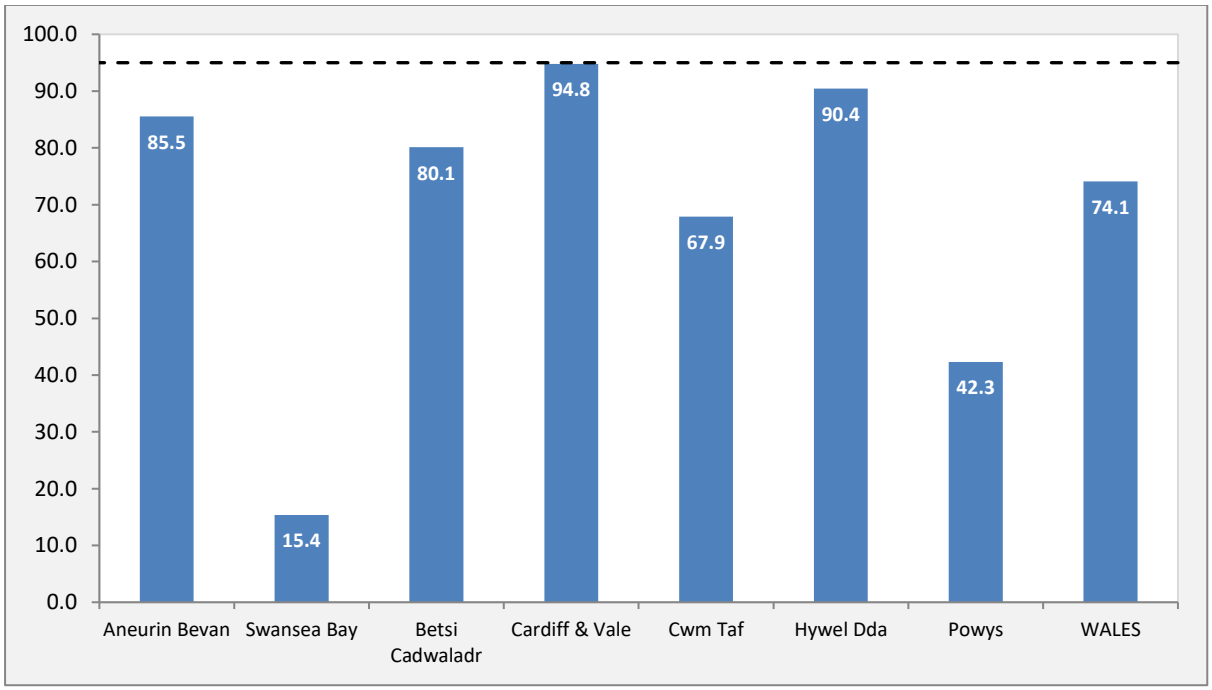
Achievement of coverage measures have been maintained. Coverage performance is affected by the timeliness and quality of sample capture and dispatch, effectiveness of sample transport arrangements and speed of laboratory testing. Throughout a number of points in 2021-22, service delivery was impacted by Covid-19 safety measures and disruption caused by staffing shortages which affected performance.

3.3 Timeliness of testing

***Standard 3A**: 95% of samples are taken between day five-eight of life (standard amended from November 2021)*
***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 3A

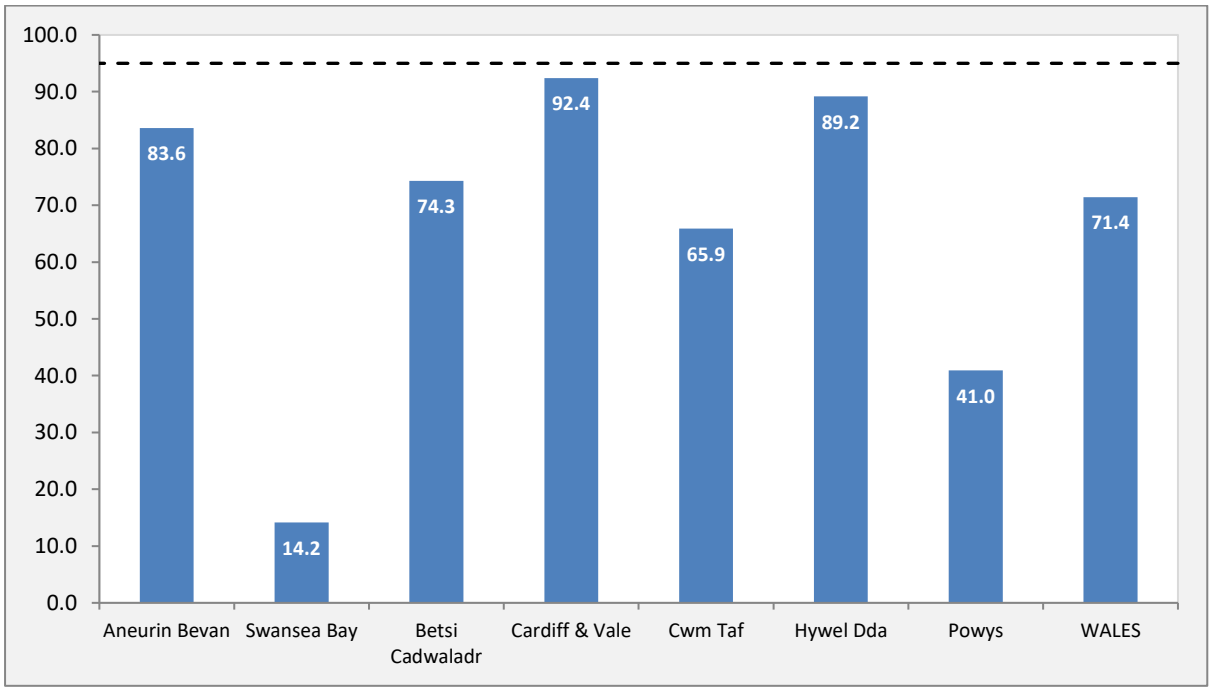
Graph 1a: Timely collection of samples (day five–eight of life)



This standard has only been met in one health board area. Across Wales, 74.1% of samples were taken between day five and day eight of life, which is a 4.9% drop in performance against this standard compared to the previous year.

During 2021-22, the 3A standard associated with timely sample collection was revised from covering days five to eight of life to days five to six of life. This step was taken to align more closely with other UK nations and improve timely collection of samples. Graph 1b shows performance against the revised standard for the full year.

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



Performance against standard 3A has been directly affected by the adoption of a contingency measure at the start of the pandemic and continuing throughout all of 2021-22. From March 2020, 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. The contingency standard 3J is routinely monitored. On an all-Wales basis, 97.8% of samples were collected between day four and six of life, which is a 0.2% drop compared to 2020-21.

This means that 19% of samples were collected on day four. It should be noted that some health boards actively prioritised sample capture on day four, adversely affecting their performance against standard 3A, but not delaying the timeliness of screening for those babies.

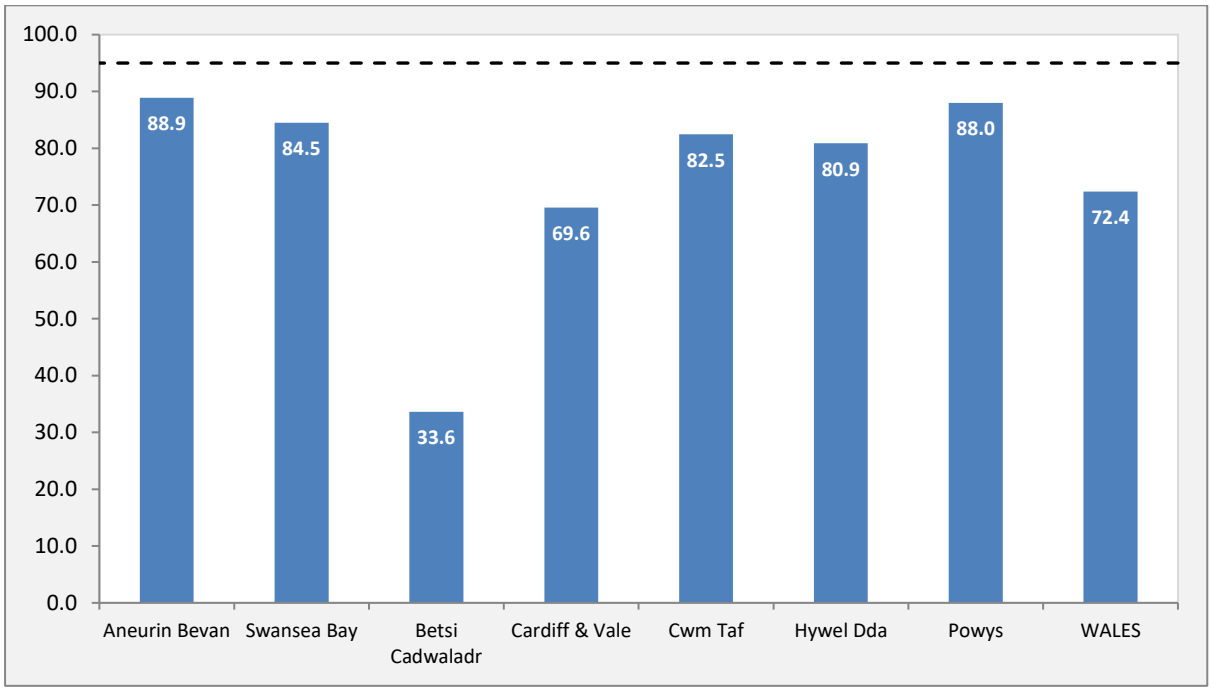
Table 4: Standard 3J performance by Health Board

Health Board	% Samples captured day 4-6 of life
Aneurin Bevan UHB	96.2
Swansea Bay UHB	96.4
Betsi Cadwaladr UHB	92.6
Cardiff and Vale UHB	93.9
Cwm Taf Morgannwg UHB	96.4
Hywel Dda UHB	97.1
Powys Teaching Health Board	96.3
All Wales	95.2

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 2: Timely collection of avoidable repeat samples



Across Wales, 72.4% of avoidable repeat samples were taken within three calendar days of the request. The standard has not been met but most health boards showed an improvement or maintained their performance from the previous year.

All requests for repeat samples are emailed to designated generic email addresses in the maternity and neonatal units. Regular reviews of the process have taken place so that improvements can be made. 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. 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*
28	107	32	9	6	5	3	1	5	27	223

* Across Wales 3 (1.3%) CHT second samples were taken too early, meaning before day 28 but not discharged. This compares to 2 samples (0.9%) in 2020-21. CHT second samples that are taken too early are not accepted for testing and require a repeat sample to be taken.

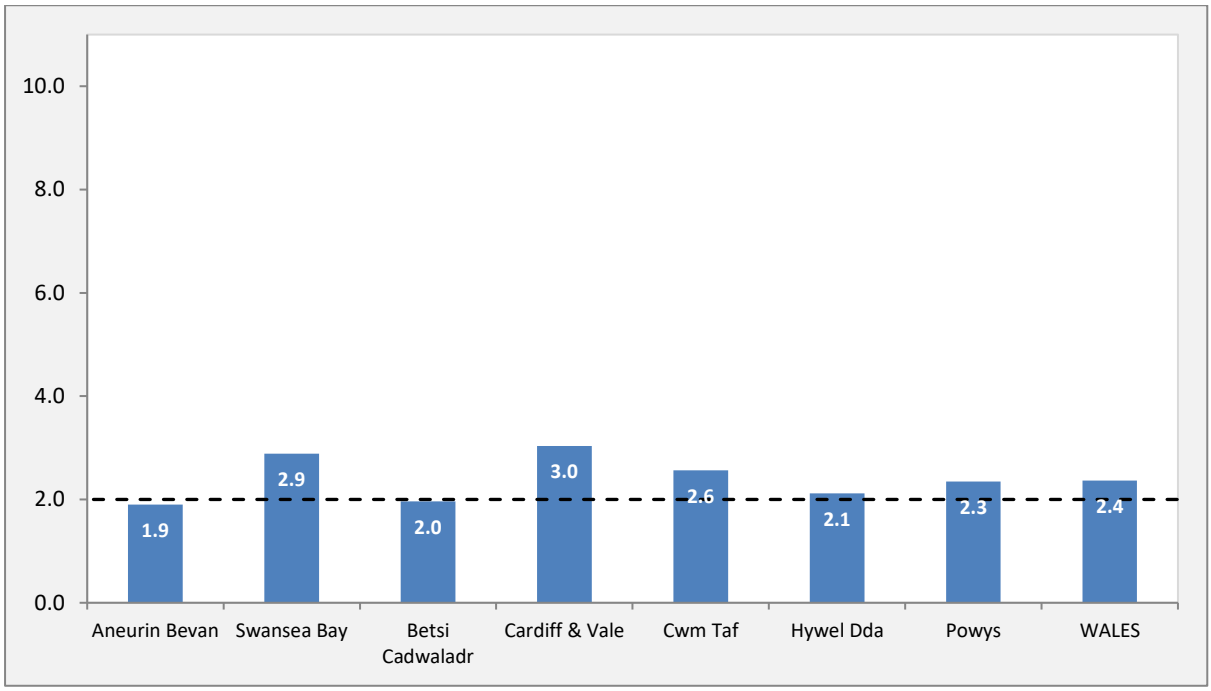
The programme continues to work closely with the neonatal units across Wales to improve performance so that CHT second samples are taken at the correct time. NBSW education resources for neonatal unit staff include the short film 'Newborn bloodspot screening in neonatal units'. An all Wales task and finish group was established in 2021-22 with representatives from neonatal units to share good practice and explore service improvement.

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 amended from November 2021)*
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 amended from November 2021)*

Standard 4A

Graph 3: Avoidable repeat rate



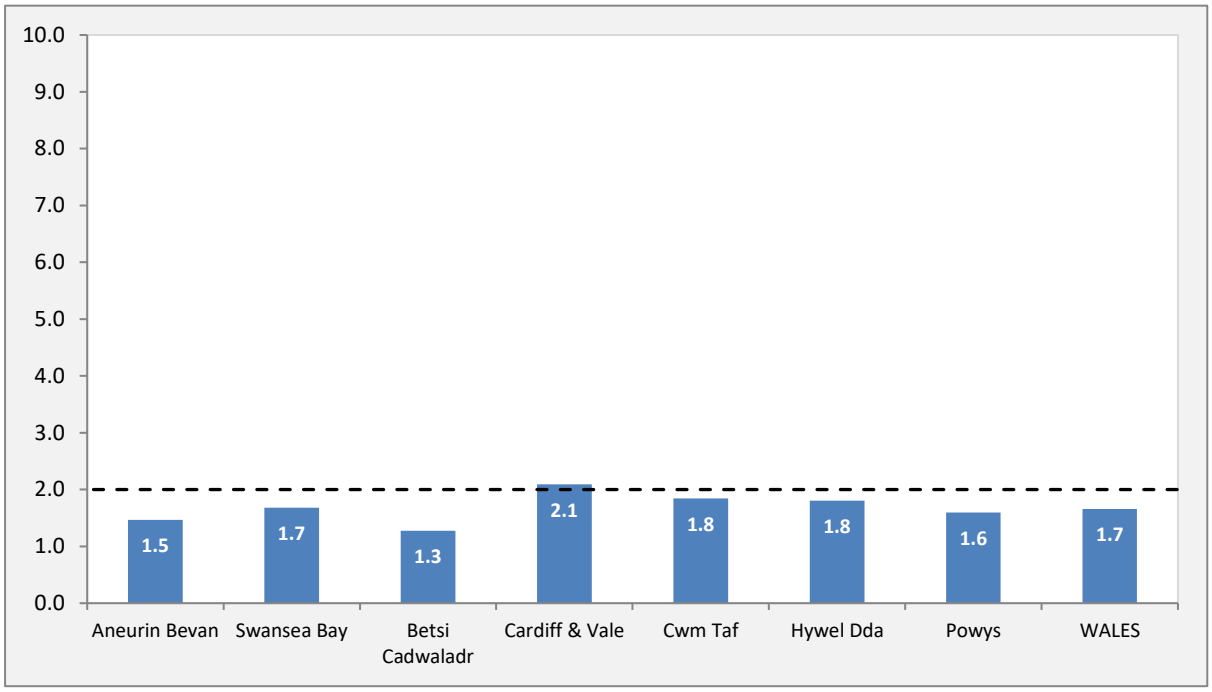
The avoidable repeat rate in Wales is 2.4%. Continuing to achieve the standard of ≤2% is a high priority for the programme to avoid delays in the referral of babies and to avoid the other costs associated with repeating samples. Whilst the standard was achieved again during 2021-22, the all Wales figure masks reduced performance in most health boards compared to the previous year.

The programme undertakes a wide range of actions to improve sample quality and minimise the number of babies requiring repeat samples. These include a range of training videos, resources and sessions delivered by programme staff. Sample takers are encouraged to register with the programme which ensures that they receive programme updates directly, and allows them to access individual reports on the quality of the samples they have taken.

Each month, 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 4: Poor quality repeat rate

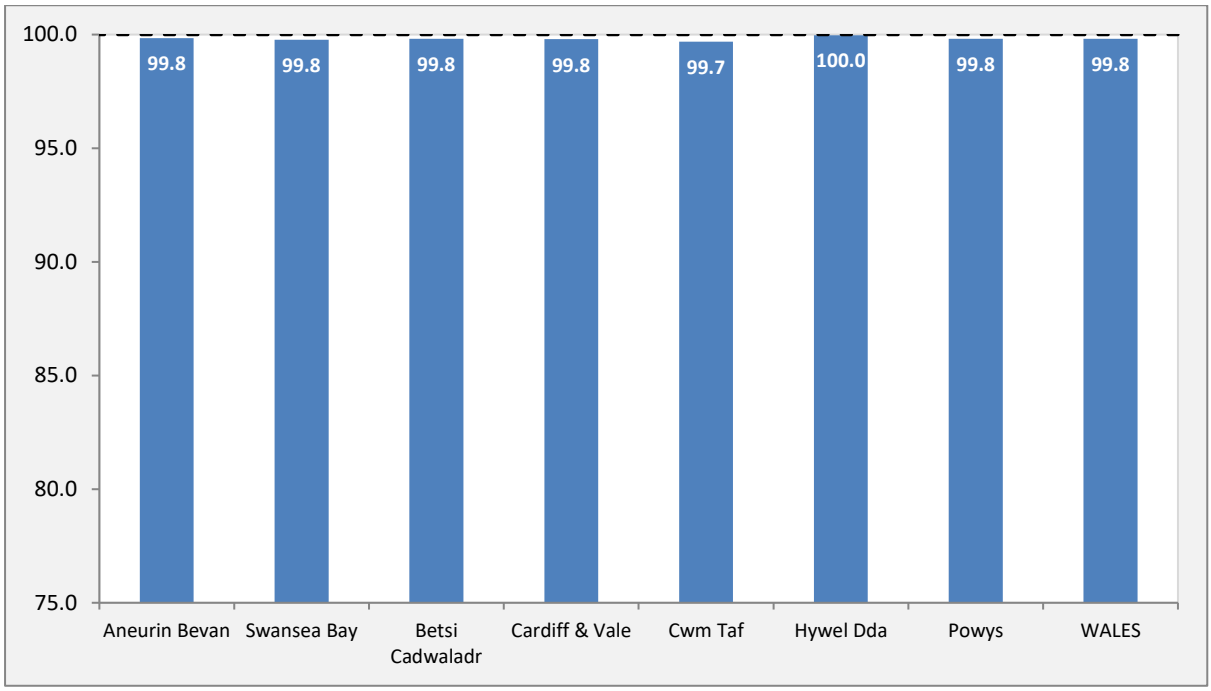


Across Wales, 1.7% of samples required repeating due to poor quality bloodspots. Unfortunately, compared with the previous year, performance dropped in all health board areas other than Powys .

Ensuring high quality sample capture is a key quality indicator and remains a high priority for the programme. The standards review resulted in a strengthening of the threshold for this indicator, reducing from ≤2% to ≤1.5%, with the programme committed to continued work with sample takers to produce high quality samples for all babies.

Standard 4C

Graph 5: NHS number on bloodspot card



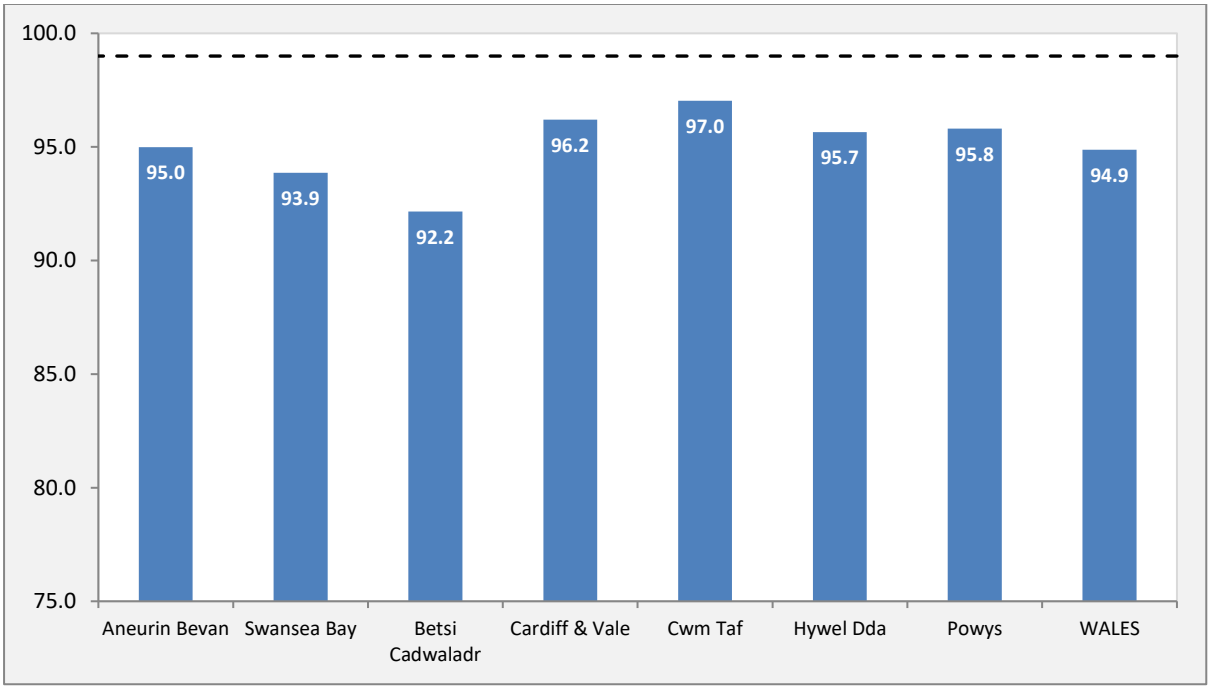
A valid NHS number for the baby was recorded on 99.8% bloodspot cards received in the laboratory, the same as last year. Illegibility of NHS numbers, inaccuracy or mixing up the numbers with another baby are the main reasons for non-compliance. Work continues with the health boards to improve performance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

The standards review adjusted the threshold for this standard from 100% to 99% in recognition of the potential for human error, and thus unrealistic expectation of consistent 100% achievement.

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 (standard amended from November 2021)*

Graph 6a: Timely receipt of card in laboratory (four working days)



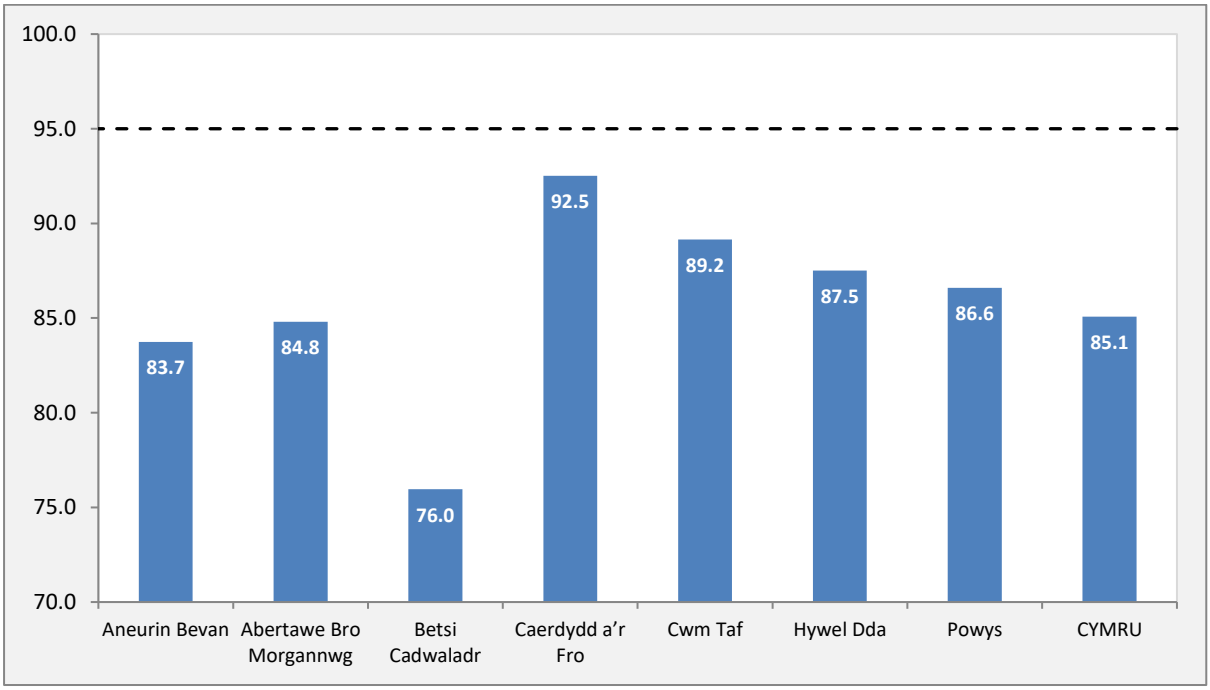
Across Wales, 94.9% of samples were received in the laboratory within four working days. This is an increase of 0.6% compared with the previous year. Continued performance improvement in this standard remains a high priority to enable timely referral of screen positive babies into clinical care.

Work continues with the health boards and Royal Mail to minimise delays in samples reaching the laboratory. 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 has been highlighted to sample takers. The use of internal mail systems has been associated with delays in the receipt of samples.

Delays in sample receipt are significantly increased during the Christmas period and addressing these delays is a high priority for the programme. Information for Royal Mail staff to highlight the importance of timely receipt of samples in the laboratory is distributed prior to the Christmas period each year. The programme works with the laboratory, health boards, failsafe teams and Royal Mail to identify issues so that they can be investigated and action can be taken to minimise delays.

The programme standard review adjusted this standard to receipt within three working days of the sample being taken, but slightly reduced the performance threshold from 99% to 95%. Performance against the revised standard is shown on graph 6b. Work continues to explore ways to reduce the transit time of samples, with a task and finish group established to consider a range of potential options.

Graph 6b: Timely receipt of card in laboratory (three working days)



3.6 Laboratory processing and referral

The current standard for timely processing of specimens was met for all conditions. For the inherited metabolic disorders; a total of 7 infants were referred for follow up; 1 case of PKU, 2 cases of isovaleric acidaemia (IVA) and 4 cases of medium-chain acyl-CoA dehydrogenase deficiency (MCADD). Confirmation of receipt into clinical care was available for all 7 cases (100%). The average age of timely receipt of the 4 MCADD cases into care was day 12 of life (range 8-18), for the 2 IVA cases (day 9 & 10) and day 3 for the PKU case (family history sample).

Of the 17 congenital hypothyroidism (CHT) screen positive cases identified and referred for follow up, 13 were identified as having a raised TSH ≥20mU/L on the initial sample and 4 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 17 cases (100%). Of the 13 CHT cases with a raised TSH ≥20mU/L on the initial screening sample, 13 (100%) were received into care by day 14 of life. The average age at the first clinic appointment was day 11 of life (range 9-14 days). Of the 4 cases identified following an initial borderline test result, 3 (75%) cases were received into care by day 21 of life (the average age at the first clinic appointment was day 20 of life, (range 17-24 days)). Feedback as to whether or not patients were commenced on thyroxine treatment was good with 17/17 (100%) of cases being reported. A total of 56 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 40 (71.4%) with 12 samples being collected on or after day 11 (range 11-50 days). A few samples (n=4) were collected before day 7.

For CF screening, a total of 17 cases were referred for follow-up. Of the 17 cases referred; 6 had 2 CF pathogenic variants, 10 had 1 pathogenic variant and 1 case had an IRT concentration ≥170ng/ml but with no pathogenic variants detected. A total of 7 cases (6 cases with 2 mutations and 1 case with 1 mutation) were confirmed as having CF, following a positive sweat test. Confirmation of receipt into clinical care was available for all 17 cases (100%). Of these 17 cases referred for follow-up, 13 (76.5%) of cases were seen at the first clinic appointment by day 28 of life. The average age at the first clinic appointment was 25 days (range 20-34).

1 case of sickle cell disorder (SCD) was detected and was referred into clinical care on day 15 of life. This case was also identified via the 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*

* Definitions amended from November 2021 to reflect change to standards. All newborn eligibility definitions adjusted from 'day five-eight of life' to 'day five-six 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 five 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 older 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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Dr Sharon Hillier	Director of Screening Division
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Guy Stevens	Deputy Informatics and Data Services Manager
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