GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Newborn Bloodspot Screening Wales Annual Statistical Report 2022-23

Version 1

Mae'r ddogfen yma ar gael yn y Gymraeg/This document is available in Welsh

Publication details

This report is a detailed summary of information on work undertaken by the Newborn Bloodspot Screening Programme for the year April 2022 to the end of March 2023. Results are reported by health board where screening has been carried out. Further details are available on request.

Title: Newborn Bloodspot Screening Wales Annual Statistical Report 2022-23
Date: This report published July 2025
ISBN: 978-1-83766-621-8

Contact details,

Guy Stevens, Deputy Lead Informatics and Data Services Manager,
Informatics Division, Public Health Wales,
Number 2 Capital Quarter, Tyndall Street, Cardiff, CF10 4BZ

Email: screening.information@wales.nhs.uk

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.

QA 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.

Copyright information

© 2025 Public Health Wales NHS Trust.

Material contained in this document may be reproduced under the terms of the Open Government Licence (OGL) www.nationalarchives.gov.uk/doc/open-government-licence/version/3/ provided it is done so accurately and is not used in a misleading context.

Acknowledgement to Public Health Wales NHS Trust to be stated. Copyright in the typographical arrangement, design and layout belongs to Public Health Wales NHS Trust.

This document is also available in Welsh.

Contents

Key messages 7
Introduction 8
Background 8
Programme delivery 10
Screening pathway 11
Headline statistics 14
Data 15
Number of eligible births and number tested 15
Completeness of offer (newborns) 17
Completeness of offer (all babies) 19
Coverage (newborns) 21
Coverage (all babies) 23
Timely collection of sample 25
Timely collection of avoidable repeat sample 27
Timely collection of second congenital hypothyroidism (CHT) sample for pre-term babies 29
Timely collection of second sample for borderline TSH 31
Avoidable repeat rate 33
Poor quality repeat rate 36
NHS number validation 38
Timely receipt of card in laboratory 40
Timely processing of positive samples in laboratory 42
Timely clinical care for positive babies 44
Definitions 46
Production team 51
Programme contact details 52

Tables

Table 1: Number of eligible births and number tested in Wales April 2022 to March 2023 16
Table 2: Eligible newborn babies offered bloodspot screening by day fourteen of life in 2022-23 ..18
Table 3: Eligible newborn babies offered bloodspot screening by day fourteen of life in 2020-21 to 2022-23 18
Table 4: Eligible babies offered bloodspot screening within eighteen days of registration in 2022-23 20
Table 5: Eligible babies offered bloodspot screening within eighteen days of registration - 2020-21 to 2022-23 20
Table 6: Eligible newborn babies with a conclusive result by day seventeen of life in 2022-23 22
Table 7: Eligible newborn babies with a conclusive result by day seventeen of life in 2020-21 to 2022-23 22
Table 8: Eligible babies with a conclusive result within twenty-one days of registration in 2022-23 24
Table 9: Eligible babies with a conclusive result within twenty-one days of registration - 2020-21 to 2022-23 24
Table 10: First sample between day 4 and 6 2022-23 26
Table 11: First sample between day 4 and 6 2020-21 to 2022-23..... 26
Table 12: Day distribution of first sample 2022-23 26
Table 13: Avoidable repeat sample within 3 calendar days of request 2022-23..... 28
Table 14: Avoidable repeat sample within 3 calendar days of request 2020-21 to 2022-23 28
Table 15: Day distribution of avoidable repeat sample 2022-23 28
Table 16: Second CHT sample for pre-term babies by day twenty-eight of life 2020-21 to 2022-23 30
Table 17: Day distribution of testing of second CHT sample for pre-term babies 2022-23..... 30
Table 18: Second sample for borderline TSH between 7 and 10 days of initial sample in 2022-23 ...32
Table 19: Second sample for borderline TSH between 7 and 10 days of initial sample - 2020-21 to 2022-23 32
Table 20: Avoidable repeat rate 2022-23 34
Table 21: Avoidable repeat rate 2020-21 to 2022-23 34
Table 22: Poor quality repeat rate 2022-23 37
Table 23: Poor quality repeat rate 2020-21 to 2022-23..... 37
Table 24: NHS number on card 2022-23 39
Table 25: NHS number on card 2020-21 to 2022-23..... 39
Table 26: Lab receipt of card within 3 working days 2022-23 41
Table 27: Lab receipt of card within 3 working days 2020-21 to 2022-23 41
Table 28: Day distribution of card receipt in the laboratory for 2022-23 41
Table 29: Timely processing of positive samples 2022-23 43
Table 30: Timely processing of positive samples 2020-21 to 2022-23..... 43

Table 31: Timely referral into clinical care for positive babies 2022-23 45
Table 32: Timely referral into clinical care for positive babies 2020-21 to 2022-23.....45

Figures

Figure 1: Pathway for newborn bloodspot screening.....13
Figure 2: Reasons for avoidable repeat, percentage of all repeat samples requested 2020-21 to 2022-23.....35

Key messages

- Newborn bloodspot screening identifies babies who may have rare but serious conditions.
- If a baby is found to have any of the conditions, they will receive early specialist care and treatment.
- Early treatment can improve an affected baby's health and prevent severe disability or even death.
- Screening is not 100% accurate. If the screening test suggests a problem, the baby will need further tests to confirm that they have the condition.
- Newborn bloodspot screening is recommended by the NHS.
- 'Information for Parents', which is online from Newborn Bloodspot Screening - Information for parents - Public Health Wales, explains the conditions screened for and how the sample is taken.

Introduction

Background

The aim of Newborn Bloodspot Screening Wales (NBSW) is to offer all eligible babies timely, quality assured screening for rare but serious conditions that would benefit from early intervention to reduce mortality and/or morbidity.

The NBSW programme was established in 2014. Whilst bloodspot screening for some conditions was available through NHS organisations in Wales prior to that point, transferring responsibility to a nationally managed programme ensured that delivery and performance could be standardised. Additionally, as part of NBSW establishment, the introduction of a national failsafe system ensured that every eligible baby was offered screening and that every bloodspot card reached the laboratory.

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.

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)

Eligibility

All babies up to 1 year of age who are resident in Wales are eligible for NBSW screening. Screening for cystic fibrosis (CF) is not offered to older babies, as the test is unreliable after 8 weeks of age.

The programme aims to screen most babies when they are in the first week of life to support access to treatment at the earliest opportunity. Babies who are resident in Wales at day 5-6 of life, whether they usually live in Wales or are temporarily resident but in receipt of routine midwife care are all eligible for NBSW screening.

Samples cannot be taken before day 4 of life.

Those babies that move into Wales from elsewhere are eligible for NBSW screening up to the age of 1 if there is no evidence that they have been tested or declined screening elsewhere in the UK.

Sources of additional information

Further details about NBSW can be found on the programme web pages: phw.nhs.wales/services-and-teams/screening/newborn-bloodspot-screening-wales/

Screening locations

Screening samples are taken as part of routine postnatal care, usually by community midwives during home visits. Babies in hospital wards or special care units will be offered and receive screening whilst they are an inpatient. In some cases, particularly for older babies, bloodspot samples are taken in clinics run by health visitors.

Summary of activity in reported year

There were a range of developments throughout 2022-23, many of which impacted on programme delivery. The most significant change resulted from the industrial action taken by Royal Mail staff during 2022. Since the inception of NBSW, samples taken from babies across Wales have been transferred to the national laboratory via Royal Mail post. Previously, this has provided generally swift transport, with sample takers able to access a wide network of convenient drop off points via post boxes. The industrial action meant that there was likely to be substantial delays to sample delivery, with increased risk that babies may not be identified in time for effective treatment. As a result, a dedicated sample courier service was rapidly commissioned to ensure timely transport of samples from a network of collection points across NHS Wales premises. Initially, the service was introduced for ten weeks from the end of October 2022. This was further extended to the end of the financial year due to continued uncertainty around Royal Mail provision, and in recognition of the reduced transit times that resulted from the change.

In Autumn 2022, the programme transitioned to a 'digital first' approach to providing pre-screening information. This means that most families access information about NBSW screening via the programme web pages, rather than a paper information leaflet. New animations were developed to support this change, which offers particular advantages around translation in a range of languages and a more engaging format for key information. The digital work was further extended with the launch of a new Information for Professionals website to provide a single place where sample takers and clinical colleagues can access key NBSW information and keep up to date with programme developments.

Service changes within the laboratory and specialist clinical teams took place, including a change in referral pathways to reflect the move of the specialist Inherited Metabolic Disorders service being provided by Birmingham Children's Hospital for babies in South Wales. Additionally, the laboratory implemented a change to the test Cut off Value used in Wales to trigger additional testing for Cystic Fibrosis, to ensure that the risk of babies being missed remains low.

Our ongoing work with sample takers to support the capture of high quality samples has continued. Agreement was reached to use General Medical Council numbers for doctors when registering as sample takers, increasing the likelihood that this data will be recorded on the sample card. The programme approved a process to ensure that samples are only taken by NHS staff, to provide assurance that sample takers are trained and able to access all NBSW resources. We also expanded the use of thank you letters for sample takers who submit consistently high quality samples from intensive care teams to all sample taker staff groups. Finally, the programme completed an evaluation of the acceptance of samples taken from babies at day 4 of life. This was originally introduced during the Covid-19 pandemic, but has been shown to be effective, with no negative impact on the quality of screening and no notable increase in repeat samples required.

Developments since the reported year

In response to the success of the sample courier service implemented in 2022, the programme secured funding to continue with this new way of working. Further improvements to reduce transit time and support sample tracking have been progressed as part of this work.

Continuing focussed efforts to improve and maintain sample quality, programme staff developed resources for sample takers and laboratory staff to ensure a shared and clear understanding of sample acceptance criteria. In addition, the programme has published a sample taker performance and assurance framework to support Health Boards to remedy sample taker practice where standards are not met in a consistent way.

In November 2022, the UK National Screening Committee recommended the introduction of bloodspot screening for a new condition – Hereditary Tyrosinemia Type 1. Throughout 2023-24, work to design detailed proposals for clinical pathways and laboratory testing progressed as part of UK-wide specialist groups. This work continues; refining the UK plans to ensure they fit within the Welsh context.

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. The NBSW Head of Programme manages both NBSW and Newborn Hearing Screening Wales (NBHSW). There are 2 NBSW programme co-ordinators with administration support.

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 any of the conditions screened for. 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 system which 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 fourteen of life. Every baby identified by the failsafe is followed up by the administration failsafe teams. The 3 regional teams across Wales are staffed by newborn screening managers and administrative staff who work across both the NBSW and Newborn Hearing Screening Wales programmes.

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

Screening pathway

Babies who are eligible for screening are identified in each health board from midwife birth notifications. Eligible babies up to 1 year of age who move into 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 inpatients in those units at day 5 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 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 6 weeks of the sample being taken.

For babies who are suspected of having any of the conditions, the results letter is provided in person by the health visitor to give parents an opportunity to discuss the outcome. After the baby has been referred for diagnostic assessment, and once this process is underway the programme contacts the baby's health visitor to inform them of the result. The baby's health visitor is then sent the screening results letter and supporting information about the identified condition to enable an informed discussion of the results with the parents. The results for each baby are also 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/

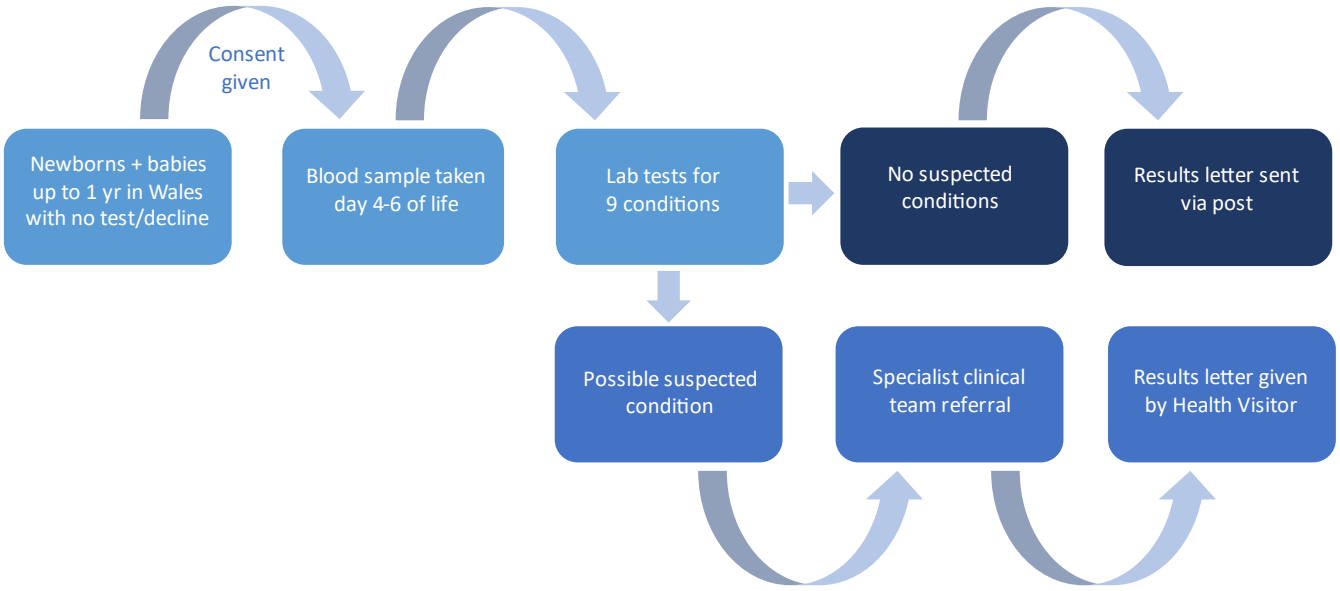

Figure 1: Pathway for newborn bloodspot screening

Headline statistics

This report covers the period from April 2022 to March 2023

- The number of eligible births across Wales was 28,313
- The number of babies tested was 28,178 (99.5%)
- 96.5% of eligible newborn babies had a bloodspot card (for screening or decline) received in the laboratory by day fourteen of life (completeness of offer)
- 95.0% of eligible newborn babies had conclusive bloodspot screening results by day seventeen of life (coverage)
- 95.2% of first blood spot samples were taken between days 4 and 6 of life
- 54.4% of samples were taken on day 5 of life, in line with programme recommendations
- The avoidable repeat rate was 2.8%
- 99.9% of bloodspot cards received in the laboratory had a valid NHS number for the baby recorded
- 85.1% of bloodspot cards were received within 3 working days of sample collection
- The number of screen positive babies detected in the year was as follows: phenylketonuria (4), maple syrup urine disease (0), medium-chain acyl-CoA dehydrogenase deficiency (6), homocystinuria (1), glutaric aciduria type 1 (0), isovaleric acidaemia (4), congenital hypothyroidism (27), cystic fibrosis (16) and sickle cell disorders (4). These are screen positive results, not all babies were diagnosed with the suspected condition.

Data

Number of eligible births and number tested

Definition

The total number of babies meeting the NBSW eligibility criteria during the period, and number of those that were screened.

Result for 2022-23

28,313 eligible births, with 28,178 tested.

Three-year trend

There were 28,313 eligible births (28,178 tested) in 2022-23, 29,716 eligible births (29,587 tested) in 2021-22, and 28,612 eligible births (28,445 tested) in 2020-21.

Comment

There were 135 babies that were not tested in this period. Parents declined screening in 42 newborns and 46 babies that moved into Wales from outside the UK. Sadly, there were 23 deaths after day 5.

A suspended status was recorded for 24 babies. These were movements into Wales from outside the UK that, 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.

Figures and tables

Table 1: Number of eligible births and number tested in Wales April 2022 to March 2023

Health Board	Births	Tested	Rate (%)
Aneurin Bevan	5,806	5,781	99.6
Betsi Cadwaladr	5,869	5,841	99.5
Cardiff & Vale	4,718	4,694	99.5
Cwm Taf	4,237	4,226	99.7
Hywel Dda	3,019	2,993	99.1
Powys	987	984	99.7
Swansea Bay	3,270	3,254	99.5
Wales	28,313	28,178	99.5

The Wales total above and in all subsequent tables includes some babies who do not map to a health board.

Completeness of offer (newborns)

Definition and standard

The percentage of eligible newborn babies who have a notification of receipt of the bloodspot card in the laboratory by day fourteen of life.

Standard: 99%

Results for 2022-23

All Wales rate: 96.5%

Three-year trend

All Wales rates: 96.5% in 2022-23, 97.2% in 2021-22, and 96.9% 2020-21.

Geographic overview

At an all-Wales level, this standard was not met during the year. Whilst the standard was achieved in some Health Board areas during individual months, performance across the year fell below standard for all organisations. As has been the case before, performance during December and January were particularly poor because of the impact of Christmas postal delays on sample transit via Royal Mail.

Comment

Performance against this standard is affected by the timeliness of the screening offer and dispatch of the sample card. The effectiveness of sample transport arrangements and laboratory booking in processes also play a key part.

Delays in sample transport were apparent from late August to November because of industrial action by Royal Mail staff, which caused delays in sample transit times. The sample courier service was found to minimise the impact of routine Christmas delays along with mitigating further disruption linked to industrial action. By contrast, improved performance was seen during February and March once use of the sample courier service had become well embedded.

More generally, the programme routinely emphasises the importance of timely sample capture and dispatch. This includes the need to complete and submit sample cards when the offer of screening is declined.

Figures and tables

Table 2: Eligible newborn babies offered bloodspot screening by day fourteen of life in 2022-23

Health Board	Births	Offer in time	Rate (%)
Aneurin Bevan	5,806	5,600	96.5
Betsi Cadwaladr	5,869	5,680	96.8
Cardiff & Vale	4,718	4,523	95.9
Cwm Taf	4,237	4,136	97.6
Hywel Dda	3,019	2,947	97.6
Powys	987	956	96.9
Swansea Bay	3,270	3,096	94.7
Wales	28,313	27,332	96.5

Table 3: Eligible newborn babies offered bloodspot screening by day fourteen of life - 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	96.8	97.3	96.5
Betsi Cadwaladr	96.6	96.8	96.8
Cardiff & Vale	96.2	96.3	95.9
Cwm Taf	98.0	98.2	97.6
Hywel Dda	97.6	97.9	97.6
Powys	97.7	97.8	96.9
Swansea Bay	96.7	96.8	94.7
Wales	96.9	97.2	96.5

Completeness of offer (all babies)

Definition and standard

The percentage of all babies who have a notification of receipt of the bloodspot card in the laboratory within eighteen days of registration of birth.

Standard: 99%

Results for 2022-23

All Wales rate: 98.6%

Three-year trend

All Wales rates: 98.6% in 2022-23, 98.7% in 2021-22, and 98.9% in 2020-21.

Geographic overview

Only one Health Board area achieved this standard, with the remaining organisations and the all Wales position narrowly missing the required level.

Comment

As previously noted, performance against this standard was directly impacted by delays in sample transit between sample taker and laboratory linked to industrial action by Royal Mail staff. This is shown in reduced performance overall for 2022-23 compared with previous years. The potential continued use of the sample courier service, introduced in late 2022, provides opportunities to improve performance against this standard.

Figures and tables

Table 4: Eligible babies offered bloodspot screening within eighteen days of registration in 2022-23

Health Board	Babies	Offer in time	Rate (%)
Aneurin Bevan	11,786	11,634	98.7
Betsi Cadwaladr	12,077	11,927	98.8
Cardiff & Vale	9,791	9,592	98.0
Cwm Taf	8,640	8,563	99.1
Hywel Dda	6,137	6,066	98.8
Powys	2,104	2,080	98.9
Swansea Bay	6,708	6,560	97.8
Wales	58,079	57,241	98.6

Table 5: Eligible babies offered bloodspot screening within eighteen days of registration - 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	98.9	98.8	98.7
Betsi Cadwaladr	98.9	98.9	98.8
Cardiff & Vale	98.6	98.4	98.0
Cwm Taf	99.2	99.1	99.1
Hywel Dda	98.9	98.9	98.8
Powys	98.8	98.9	98.9
Swansea Bay	98.6	98.1	97.8
Wales	98.9	98.7	98.6

Coverage (newborns)

Definition and standard

The percentage of newborn babies who have a conclusive bloodspot screening result by day seventeen of life.

Standard: 95%

Results for 2022-23

All Wales rate: 95.0%

Three-year trend

All Wales rates: 95.0% in 2022-23, 95.9% in 2021-22, and 96.0% in 2020-21.

Geographic overview

Five of the seven Health Boards achieved this standard, with three organisations maintaining performance in every month other than for babies born in December.

Comment

Coverage performance is affected by the timeliness and quality of sample capture and dispatch, effectiveness of sample transport arrangements and speed of laboratory testing. As such, performance generally falls below standard for babies born in December, where timeliness is affected by both postal delays and the extended bank holiday weekends. This pattern reflects performance in previous years.

Figures and tables

Table 6: Eligible newborn babies with a conclusive result by day seventeen of life in 2022-23

Health Board	Births	Result in time	Rate (%)
Aneurin Bevan	5,806	5,540	95.4
Betsi Cadwaladr	5,869	5,590	95.2
Cardiff & Vale	4,718	4,448	94.3
Cwm Taf	4,237	4,065	95.9
Hywel Dda	3,019	2,878	95.3
Powys	987	950	96.3
Swansea Bay	3,270	3,028	92.6
Wales	28,313	26,890	95.0

Table 7: Eligible newborn babies with a conclusive result by day seventeen of life - 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	96.1	96.4	95.4
Betsi Cadwaladr	95.9	95.4	95.2
Cardiff & Vale	95.2	95.2	94.3
Cwm Taf	96.7	96.5	95.9
Hywel Dda	96.6	96.7	95.3
Powys	97.3	96.0	96.3
Swansea Bay	95.5	95.4	92.6
Wales	96.0	95.9	95.0

Coverage (all babies)

Definition and standard

The percentage of all babies who have a conclusive bloodspot screening result within twenty-one days of registration.

Standard: 95%

Results for 2022-23

All Wales rate: 96.8%

Three-year trend

All Wales rates: 96.8% in 2022-23, 97.2% in, 2021-22, 97.2% in 2020-21.

Geographic overview

This standard was met at an all Wales level and by each individual Health Board. Variations in performance are noted, with corresponding impact of an elevated avoidable repeat rate and delayed capture of repeat samples of note.

Comment

Performance against this standard is affected by the timeliness and quality of the first sample. In addition, where repeat samples are required, delays in capturing these can contribute to reduced performance. As is the case for other standards, delays between sample capture and arrival in the laboratory are also a factor.

For older babies, some Health Boards run clinics to take bloodspot samples to support high quality sample capture by a smaller group of specialist staff. The timing of these clinics can affect performance against this standard.

Figures and tables

Table 8: Eligible babies with a conclusive result within twenty-one days of registration in 2022-23

Health Board	Babies	Result in time	Rate (%)
Aneurin Bevan	11,786	11,481	97.4
Betsi Cadwaladr	12,077	11,671	96.6
Cardiff & Vale	9,791	9,436	96.4
Cwm Taf	8,640	8,414	97.4
Hywel Dda	6,137	5,955	97.0
Powys	2,104	2,049	97.4
Swansea Bay	6,708	6,435	95.9
Wales	58,079	56,247	96.8

Table 9: Eligible babies with a conclusive result within twenty-one days of registration - 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	97.3	97.6	97.4
Betsi Cadwaladr	97.1	96.8	96.6
Cardiff & Vale	97.1	97.0	96.4
Cwm Taf	97.5	97.6	97.4
Hywel Dda	97.5	97.5	97.0
Powys	97.7	97.2	97.4
Swansea Bay	96.8	96.8	95.9
Wales	97.2	97.2	96.8

Timely collection of sample

Definition and standard

The first bloodspot sample should be taken between day 4 and 6 of life (counting day of birth as day zero).

Standard: 95%

Results for 2022-23

All Wales rate: 95.2%

Three-year trend

All Wales rates: 95.2% in 2022-23, 95.2% in 2021-22, 98.0% in 2020-21.

Geographic overview

Performance against this standard is variable and can be significantly affected by staffing shortages. In some Health Boards, shift patterns and a focus on continuity of care by a named midwife result in slight delays in sample capture.

Comment

Table 12 shows a breakdown of the day of sample capture, which illustrates that most Health Boards elect to prioritise sample capture on day 5 of life. A significant number of samples were taken on day 7, falling just outside the standard. A notable proportion were taken on day ten or later, suggesting that these babies were initially missed.

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.

Figures and tables

Table 10: First sample between day 4 and 6 2022-23

Health Board	Samples	Collection in time	Rate (%)
Aneurin Bevan	5,765	5,492	95.3
Betsi Cadwaladr	5,711	5,406	94.7
Cardiff & Vale	4,693	4,384	93.4
Cwm Taf	4,221	4,051	96.0
Hywel Dda	2,997	2,903	96.9
Powys	933	904	96.9
Swansea Bay	3,256	3,124	95.9
Wales	27,806	26,479	95.2

Table 11: First sample between day 4 and 6 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	NA	96.2	95.3
Betsi Cadwaladr	NA	92.6	94.7
Cardiff & Vale	NA	93.9	93.4
Cwm Taf	NA	96.4	96.0
Hywel Dda	NA	97.1	96.9
Powys	NA	96.3	96.9
Swansea Bay	NA	96.4	95.9
Wales	NA	95.2	95.2

Table 12: Day distribution of first sample 2022-23

<4	4	5	6	7	8	9	10	>10	Total
34	8,206	15,123	3,150	456	133	68	37	599	27,806
0.1%	29.5%	54.4%	11.3%	1.6%	0.5%	0.2%	0.1%	2.2%	

Timely collection of avoidable repeat sample

Definition and standard

Repeat testing for insufficient/poor quality samples or incomplete/incorrect sample card information should be conducted within three calendar days of the request.

Standard: 95%

Results for 2022-23

All Wales rate: 71.3%

Three-year trend

All Wales rates: 71.3% in 2022-23, 72.4% 2021-22, and 72.6% in 2020-21.

Geographic overview

Performance against this standard at a Health Board level remains variable. Small numbers in Powys have a disproportionate impact on their rate of performance compared to other areas.

Comment

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. The primary focus for the programme team is on reducing the avoidable repeat rate, which reduces the demand for this time pressured activity.

Figures and tables

Table 13: Avoidable repeat sample within 3 calendar days of request 2022-23

Health Board	Rate (%)
Aneurin Bevan	81.6
Betsi Cadwaladr	62.6
Cardiff & Vale	69.9
Cwm Taf	76.4
Hywel Dda	76.0
Powys	76.5
Swansea Bay	66.9
Wales	71.3

Table 14: Avoidable repeat sample within 3 calendar days of request 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	80.0	88.9	81.6
Betsi Cadwaladr	52.7	33.6	62.6
Cardiff & Vale	69.7	69.6	69.9
Cwm Taf	75.3	82.5	76.4
Hywel Dda	84.0	80.9	76.0
Powys	91.7	88.0	76.5
Swansea Bay	73.4	84.5	66.9
Wales	72.6	72.4	71.3

Table 15: Day distribution of avoidable repeat sample 2022-23

<=3	4	5	6	7	8	9	10	>10	Total
570	24	23	14	13	6	7	0	143	800

Timely collection of second congenital hypothyroidism (CHT) sample for pre-term babies

Definition and standard

Pre-term babies should have a second bloodspot sample taken on day twenty-eight of life or day of discharge if earlier.

Standard: 95%

Results for 2022-23

All Wales rate: 62.5%

Three-year trend

All Wales rates: 62.5% in 2022-23, 59.7% in 2021-22, and 50.2% in 2020-21.

Geographic overview

At an all Wales level, performance did not reach the standard throughout the year. No individual Health Board consistently achieved the standard but most did meet it for some months. The comparatively small number of second samples required each month per unit means that delayed capture for individual babies can have a large impact on performance figures.

Comment

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 with representatives from neonatal units exists to share good practice and explore service improvement.

Figures and tables

Table 16: Second CHT sample for pre-term babies by day twenty-eight of life 2020-21 to 2022-23

	Rate 2020-21	Rate 2021-22	Rate 2022-23
Wales	50.2	59.7	62.5

Table 17: Day distribution of testing of second CHT sample for pre-term babies 2022-23

<28	28	29	30	31	32	33	34	35	>35	Total
45	100	34	13	4	4	3	4	4	21	232

Timely collection of second sample for borderline TSH

Definition and standard

Babies with a borderline thyroid stimulating hormone (TSH) result who have a second bloodspot card for TSH received in the laboratory which was collected between 7and 10 days after the initial borderline sample.

Standard: 95%

Results for 2022-23

All Wales rate: 79.0%

Three-year trend

All Wales rates: 79.0% in 2022-23, 71.4% in 2021-22, 58.3% in 2020-21 (58.3%).

Comment

This data is only available at an all Wales level. As with other second sample testing, the overall number of babies that require this additional test is comparatively small, which impacts on the percentage figures.

The 3year trend shows a continued improvement in performance. Close engagement between the programme team, laboratory colleagues and Health Board staff have contributed to this position.

Figures and tables

Table 18: Second sample for borderline TSH between 7 and 10 days of initial sample in 2022-23

Health Board	Second TSH	Second in time	Rate (%)
Wales	81	64	79.0

Table 19: Second sample for borderline TSH between 7 and 10 days of initial sample - 2020-21 to 2022-23

	Rate 2020-21	Rate 2021-22	Rate 2022-23
Wales	58.3	71.4	79.0

Avoidable repeat rate

Definition and standard

Repeat cards that are required because of poor quality bloodspots or incomplete/incorrect information recorded.

Standard: <=2%

Results for 2022-23

All Wales rate: 2.8%

Three-year trend

All Wales rates: 2.8% in 2022-23, 2.4% in 2021-22, and 1.9% in 2020-21.

Geographic overview

Performance against this standard was variable throughout the year and between Health Board areas. Cardiff and Vale and Swansea Bay Health Boards both experienced particularly elevated avoidable repeat rates. More generally, an increased avoidable repeat rate is observed to coincide with peak holiday periods, suggesting a link between reduced staffing levels and decreased quality of sample capture.

Comment

Repeat rate rises are primarily due to increase in poor quality samples, which includes samples with an insufficient amount of blood captured. There was a notable reduction in the number of samples rejected due to documentation errors with the NHS number (Figure 2).

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.

Figures and tables

Table 20: Avoidable repeat rate 2022-23

Health Board	Cards	Repeats	Rate (%)
Aneurin Bevan	5,988	98	1.6
Betsi Cadwaladr	6,009	147	2.4
Cardiff & Vale	4,997	196	3.9
Cwm Taf	4,356	106	2.4
Hywel Dda	3,144	100	3.2
Powys	940	17	1.8
Swansea Bay	3,461	136	3.9
Wales	28,895	800	2.8

Table 21: Avoidable repeat rate 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	2.0	1.9	1.6
Betsi Cadwaladr	1.5	2.0	2.4
Cardiff & Vale	2.4	3.0	3.9
Cwm Taf	2.0	2.6	2.4
Hywel Dda	1.6	2.1	3.2
Powys	2.3	2.3	1.8
Swansea Bay	1.9	2.9	3.9
Wales	1.9	2.4	2.8

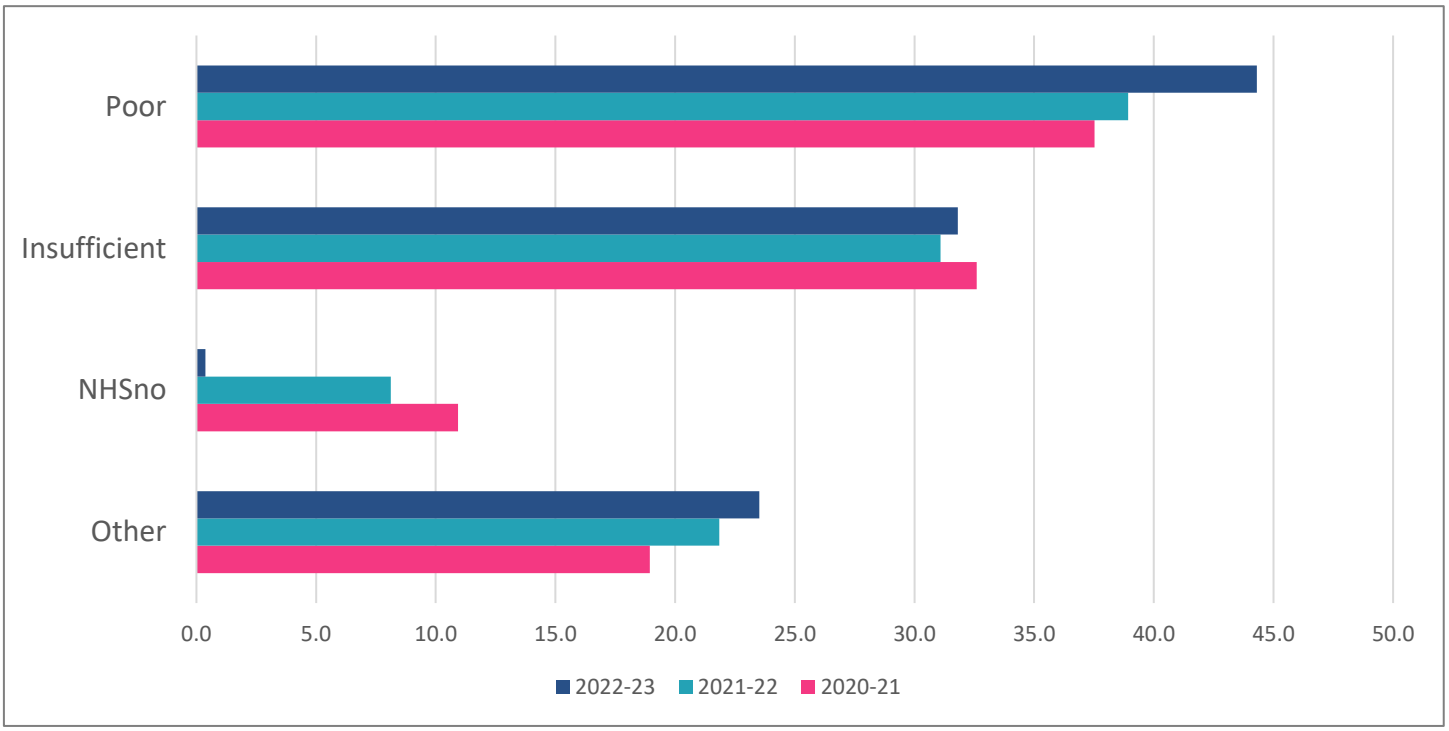

Figure 2: Reasons for avoidable repeat, percentage of all repeat samples requested 2020-21 to 2022-23

'Poor' refers to poor quality samples which are caused by samples which are not captured in line with guidance. This includes compressed samples, samples containing multiple drops of blood in the same spot, blood applied to the front and back of the card rather than a single drop being allowed to soak through.

'Insufficient' samples are those which do not contain the required amount of blood.

Sample cards submitted with missing or inaccurate NHS numbers are classified as 'NHS no'.

'Other' includes contaminated samples, samples taken too early or too late and the use of expired cards.

Poor quality repeat rate

Definition and standard

Repeat cards that are required because of poor quality bloodspots or incomplete/incorrect information recorded.

Standard: <=1.5% (formerly <=2% until November 2021)

Results for 2022-23

All Wales rate: 2.1%

Three-year trend

All Wales rates: 2.1% in 2022-23, 1.7% in 2021-22, 1.3% in 2020-21.

Geographic overview

As also seen in the avoidable repeat rate, there are variances between performance across different Health Board areas. Only Aneurin Bevan Health Board achieved the standard for the year.

Comment

This standard is a sub-set of the avoidable repeat rate and is the main reason for repeat samples being requested. A poor quality sample is primarily caused by the sample capture technique, with a range of different issues included in this category. This includes samples with insufficient blood captured to complete the range of tests incorporated into the screening process.

Sample quality is a critical part of providing high quality bloodspot screening. Poor quality samples can result in the screening test result being unreliable.

Figures and tables

Table 22: Poor quality repeat rate 2022-23

Health Board	Cards	Repeats	Rate (%)
Aneurin Bevan	5,988	67	1.1
Betsi Cadwaladr	6,009	113	1.9
Cardiff & Vale	4,997	166	3.3
Cwm Taf	4,356	83	1.9
Hywel Dda	3,144	79	2.5
Powys	940	15	1.6
Swansea Bay	3,461	91	2.6
Wales	28,895	614	2.1

Table 23: Poor quality repeat rate 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	1.4	1.5	1.1
Betsi Cadwaladr	1.0	1.3	1.9
Cardiff & Vale	1.7	2.1	3.3
Cwm Taf	1.5	1.8	1.9
Hywel Dda	1.0	1.8	2.5
Powys	1.7	1.6	1.6
Swansea Bay	1.2	1.7	2.6
Wales	1.3	1.7	2.1

NHS number validation

Definition and standard

Bloodspot cards received in the laboratory that have a valid NHS number for the baby recorded.

Standard: 99% (formerly 100%)

Results for 2022-23

All Wales rate: 99.9%

Three-year trend

All Wales rates: 99.9% in 2022-23, 99.8% in 2021-22, and 99.8% in 2020-21.

Comment

This standard was achieved, with all Health Boards delivering improved performance compared with the previous year. Across the year there were only 3 cards received which did not have a valid NHS number recorded for the baby.

Figures and tables

Table 24: NHS number on card 2022-23

Health Board	Cards	NHS number	Rate (%)
Aneurin Bevan	6,010	6,008	99.9
Betsi Cadwaladr	6,040	6,040	100
Cardiff & Vale	5,018	5,017	99.9
Cwm Taf	4,370	4,370	100
Hywel Dda	3,170	3,170	100
Powys	945	945	100
Swansea Bay	3,489	3,489	100
Wales	29,042	29,039	99.9

Table 25: NHS number on card 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	99.8	99.8	99.9
Betsi Cadwaladr	99.8	99.8	100
Cardiff & Vale	99.7	99.8	99.9
Cwm Taf	99.8	99.7	100
Hywel Dda	99.8	99.9	100
Powys	99.9	99.8	100
Swansea Bay	99.8	99.8	100
Wales	99.8	99.8	99.9

Timely receipt of card in laboratory

Definition and standard

Bloodspot cards received in the laboratory within 3 working days of sample collection.

Standard: 95%

Results for 2022-23

All Wales rate: 85.1%

Three-year trend

All Wales rates: 85.1% in 2022-23, and 85.1% in 2021-22. There was a different standard in place prior to 2021, therefore trend figures are not available for 2020-21.

Comment

This standard was not met during the period, which reflects the position of the previous year. Performance was highest in the Cardiff and Vale area. This is linked to the volume of samples that are dropped off at the University Hospital of Wales site, where the laboratory is based, by sample takers working for the Cardiff & Vale Health Board.

A notable dip in performance occurred between September and November 2022, due to the industrial action taken by Royal Mail staff which caused postal service delays.

Figures and tables

Table 26: Lab receipt of card within 3 working days 2022-23

Health Board	Cards	Receipt in time	Rate (%)
Aneurin Bevan	6,010	4,907	81.6
Betsi Cadwaladr	6,040	4,903	81.2
Cardiff & Vale	5,018	4,723	94.1
Cwm Taf	4,370	3,689	84.4
Hywel Dda	3,170	2,748	86.7
Powys	945	782	82.8
Swansea Bay	3,489	2,966	85.0
Wales	29,042	24,718	85.1

Table 27: Lab receipt of card within 3 working days 2020-21 to 2022-23

Health Board	Rate 2020-21	Rate 2021-22	Rate 2022-23
Aneurin Bevan	NA	83.7	81.6
Betsi Cadwaladr	NA	76.0	81.2
Cardiff & Vale	NA	92.5	94.1
Cwm Taf	NA	89.2	84.4
Hywel Dda	NA	87.5	86.7
Powys	NA	86.6	82.8
Swansea Bay	NA	84.8	85.0
Wales	NA	85.1	85.1

Table 28: Day distribution of card receipt in the laboratory for 2022-23

<=3	4	5	6	7	8	9	10	>10	Total
24,718	2,868	936	249	118	52	30	21	50	29,042

Timely processing of positive samples in laboratory

Definition and standard

Inherited Metabolic Disorders (IMD)/ Congenital Hyperthyroidism (CHT): clinical referral for IMD/CHT screen positive results initiated within three working days of sample receipt.

CF: clinical referral for CF screen positive results initiated within twenty-five days of sample receipt.

SCD: clinical referral for SCD screen positive results initiated within forty-two days of sample receipt.

Standard (IMD/CHT): 100%

Standard (CF): 95%

Standard (SCD): 95%

Results for 2022-23

All Wales rate (IMD/CHT): 100%

All Wales rate (CF): 100%

All Wales rate (SCD): 100%

Three-year trend

All Wales rates (IMD/CHT): 100% in 2022-23, 100% in 2021-22, 100% in 2020-21.

All Wales rates (CF): 100% in 2022-23, 100% in 2021-22, 100% in 2020-21.

All Wales rates (SCD): 100% in 2022-23, 100% in 2021-22, 100% in 2020-21.

Comment

All standards were achieved. As can be seen from the 3year data, the processes within the bloodspot screening laboratory are designed to respond rapidly when there is a potential suspected condition. For all positive samples, the initial laboratory test is followed by further laboratory testing before the screening result is confirmed and the baby referred on for diagnostic assessment.

Figures and tables

Table 29: Timely processing of positive samples 2022-23

Condition	Positive	Referral in time	Rate (%)
IMD & CHT	42	42	100
CF	16	16	100
SCD	4	4	100

Table 30: Timely processing of positive samples 2020-21 to 2022-23

Condition	Rate 2020-21	Rate 2021-22	Rate 2022-23
IMD & CHT	100	100	100
CF	100	100	100
SCD	100	100	100

Timely clinical care for positive babies

Definition and standard

IMD: first clinical appointment attendance for IMD screen positive results by day fourteen of life (excluding HCU).

CHT: first clinical appointment attendance for CHT screen positive results by day fourteen of life or initial borderline results followed by a positive result by day twenty-one.

CF: first clinical appointment attendance for CF screen positive results by day twenty-eight of life.

SCD: first clinical appointment attendance for SCD screen positive results by day ninety of life.

Standard (IMD): 100%

Standard (CHT): 100%

Standard (CF): 95%

Standard (SCD): 90%

Results for 2022-23

All Wales rate (IMD): 100%

All Wales rate (CHT): 84.6%

All Wales rate (CF): 81.3%

All Wales rate (SCD): 100%

Three-year trend

All Wales rates (IMD): 100% in 2022-23, 100% in 2021-22, and 100% in 2020-21.

All Wales rates (CHT): 84.6% in 2022-23, 94.1% in 2021-22, and 76.0% in 2020-21.

All Wales rates (CF): 81.3% in 2022-23, 76.5% in 2021-22, and 95.7% 2020-21.

All Wales rates (SCD): 100% in 2022-23, 100% in 2021-22, and 100% in 2020-21.

Comment

Due to small numbers, this information is only available at an all Wales level. The standards for CHT and CF were not achieved for this period.

Timely capture of high quality samples and Health Board processes to respond to screening referrals both impact on whether babies commence their clinical assessment within standard timescales.

Figures and tables

Table 31: Timely referral into clinical care for positive babies 2022-23

Condition	Positive	Referral in time	Rate (%)
IMD[1]	14	14	100
CHT	26	22	84.6
CF	16	13	81.3
SCD	4	4	100

[1] Excluding homocystinuria (HCU). However, the one baby who received an HCU screen positive result was referred into clinical care by day fourteen of life.

Table 32: Timely referral into clinical care for positive babies 2020-21 to 2022-23

Condition	Rate 2020-21	Rate 2021-22	Rate 2022-23
IMD	66.6	87.5	100
CHT	76.0	94.1	84.6
CF	95.7	76.5	81.3
SCD	100	100	100

Definitions

Eligible babies (newborn)

- A baby who is resident in Wales at day 5-6 of life
- A baby who is resident in Wales at day 5-6 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 5-6 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 1 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 / Suspected condition

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 / Condition not suspected

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.

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.

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.

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.

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.

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.

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.

Production team

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

Jude Kay	Head of Programme, Newborn Screening
Dr Sikha De Souza	Consultant in Public Health
Margaret Birch	Programme Co-ordinator
Dr Sharon Hillier	Director of Screening Division
Helen Clayton	Lead Informatics and Data Services Manager
Guy Stevens	Deputy Informatics and Data Services Manager
Christie Redman	Senior Informatics & Data Analyst
Dr Todd Smith	Consultant in Public Health

Professor Stuart Moat	Consultant Clinical Biochemist, Director of Wales Newborn Screening Laboratory, Cardiff & Vale University Health Board

Translated by NHS Wales Shared Services Partnership on behalf of Public Health Wales

Programme contact details

Newborn Bloodspot Screening Wales

Public Health Wales

Floor 4

2 Capital Quarter

Tyndall Street

CF10 4BZ

Email: newborn-bloodspot-screening@wales.nhs.uk

Phone: 02920 104 427

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Gweithio gyda'n gilydd i greu Cymru iachach

Working together for a healthier Wales