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# Newborn Bloodspot Screening Wales Annual Statistical Report 2023-24

Version 1

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## Publication details

This report is a detailed summary of information on work undertaken by the Newborn Bloodspot Screening Programme for the year April 2023 to the end of March 2024. 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 2023-24

Date: This report published January 2026

ISBN: 978-1-83766-770-3

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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 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.



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This document is also available in Welsh.

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## 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:

<https://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

During 2023–24, several service changes and developments were implemented, building on previous initiatives and the launch of new workstreams. Following the introduction of the courier service used to transport samples in late 2022, further refinements were made to enhance service quality. Sample collection schedules were harmonised to improve consistency; previously, midweek collections aimed to ensure daily sample deliveries to the lab. However, analysis revealed that samples collected on certain days were taking longer to reach the lab, affecting result turnaround times. Standardising the collection schedule was shown to improve timeliness.

A pilot was conducted to assess the impact of adjusting collection routes, after which lab staff agreed to modify their working patterns to increase availability on key days. These changes were then made permanent, resulting in improved testing turnaround.

Additionally, the programme introduced 'individual sample tracking' via the courier. This required the design and distribution of new sample envelopes that allow barcode scanning without revealing the baby's personal details, maintaining confidentiality. As a result, the risk of lost samples has been reduced, and processes for tracking delayed or misplaced samples have been strengthened.

We maintained a strong focus on ensuring consistently high-quality sample capture throughout the year. This resulted in the publication of the NBSW sample acceptance criteria for staff, this tool includes a visual guide to rejected samples and practical guidance of how to avoid common mistakes.

In addition, the 'Sample Taker Performance and Assurance Framework' was introduced, offering health boards a consistent approach to manage sample taker practice. It includes guidance on

addressing repeated errors and supporting staff who have spent extended time away from sample capture.

To further support 'Sample Takers' the programme launched twice-monthly virtual 'tips and tricks' sessions led by Programme Co-ordinators. These sessions provide a refresher on core aspects of high quality sample capture, plus offer tailored support to attendees. Existing quick reference guides were updated, and new materials were created in response to feedback from 'sample takers', ensuring resources remain relevant and user-informed.

Following the adoption of a 'digital first' approach to providing pre-screening information in 2022, providing most families access information about NBSW screening via the programme web pages, rather than a paper information leaflet; an evaluation of the impact of the change was completed. The change was well received, with recommendations made to inform future digital content developments based on the data gathered from viewing patterns for the programme animations.

In January 2024 changes were made to the process for managing babies who are identified as carriers of sickle cell disorders or other unusual haemoglobin following targeted Antenatal Screening Wales (ASW) early testing. These changes aim to minimise potential parental confusion caused by the different tests used for antenatal and newborn bloodspot screening. While NBSW does not actively test for carrier status, carriers may be identified incidentally. In contrast, the targeted ASW test actively aims to identify both carriers and babies with sickle cell disorder. As a result, parents may receive different screening results from the 2 programmes. To address this, NBSW introduced a process which mirrors that used to provide results for babies identified as sickle cell disorder/other unusual haemoglobin carriers through NBSW testing. This ensures parents receive clear information explaining the difference between the two sets of results and provides an opportunity to discuss the findings with their Health Visitor.

The positive impact of reduced sample transit time, coupled with the ability to better track individual samples via the courier service created an opportunity to further improve sample timeliness. To improve further and ensure that no baby is missed, programme administrators manage a failsafe process to flag any baby who reaches day 14 of life without a 'sample card' being booked into the lab. In October 2023, the trigger day for this process was brought forward from day 14 to day 12. This change supports earlier identification and testing for babies whose screening has been missed or delayed, helping improve the overall screening coverage and timeliness.

There have been some changes to the management structure and roles within Newborn Hearing Screening that were implemented between September 2023 and January 2024 and affected NBSW. Most notably, lead responsibility for NBSW failsafe moved to the newly created Newborn Screening Operations Manager in North Wales. Additionally, a new post was developed 'Newborn Screening Pathway Co-ordinator' to manage all Newborn Screening Administrators under a single line management structure, and to provide additional capacity for process improvement and assurance.

## Developments since the reported year

Work continued at a UK level to design detailed proposals for clinical pathways and laboratory testing to enable the implementation of screening for Hereditary Tyrosinemia Type 1 (HT1). In August 2023, Welsh Government approved the recommendation previously made by the UK National Screening Committee to introduce HT1 screening as part of NBSW. An implementation group was established and work continues to adapt the plans for use in Wales.

Further progress has been made in reducing the delayed sample failsafe trigger day, which was re-set at day 11 in January 2025. It is planned to reduce the timeframe further, but this will require additional service capacity to progress.

Significant work is underway to improve the stability of the 'Newborn Bloodspot Screening Wales System' (NBSWS) clinical IT system, with further plans to then adapt system functionality. This will be critical to enable a range of service improvements and developments.

## 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 7 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 14 of life (by day 12 from October 2023). 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 operations 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:

<https://phw.nhs.wales/services-and-teams/screening/newborn-bloodspot-screening-wales/>

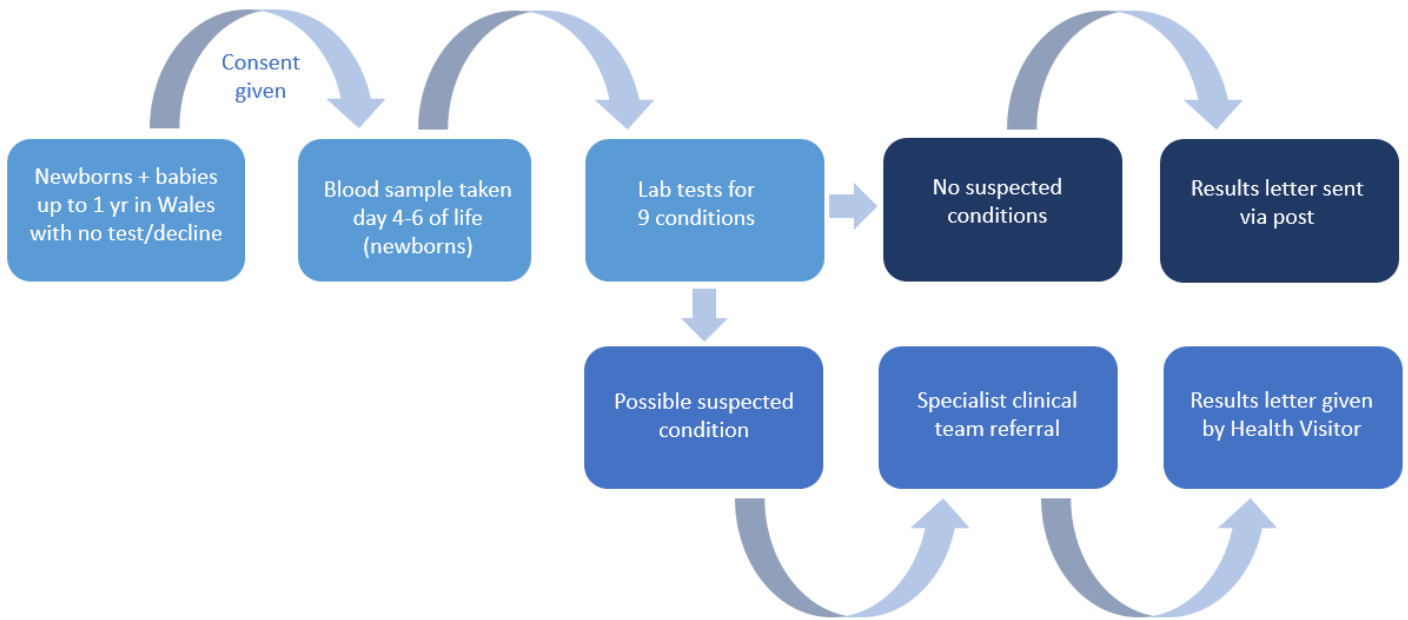


Figure 1: Pathway for newborn bloodspot screening

# Headline statistics

This report covers the period from April 2023 to March 2024

- The number of eligible births across Wales was 28,014
- The number of babies tested was 27,856 (99.4%)
- 97.5% of eligible newborn babies had a bloodspot card (for screening or decline) received in the laboratory by day 14 of life (completeness of offer)
- 95.2% of eligible newborn babies had conclusive bloodspot screening results by day 17 of life (coverage)
- 95.1% of first blood spot samples were taken between days 4 and 6 of life
- The avoidable repeat rate was 4.7%, representing repeat samples that are required because of poor quality bloodspots or incomplete/incorrect information recorded
- 99.3% of bloodspot cards received in the laboratory had a valid NHS number for the baby recorded
- 91.4% of bloodspot cards were received within three working days of sample collection
- The number of screen positive babies detected in the year was as follows: phenylketonuria (3), maple syrup urine disease (0), medium-chain acyl-CoA dehydrogenase deficiency (4), homocystinuria (0), glutaric aciduria type 1 (0), isovaleric acidaemia (1), congenital hypothyroidism (33), cystic fibrosis (23) and sickle cell disorders (3). 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 2023-24

28,014 eligible births, with 27,856 tested.

### Three-year trend

There were 28,014 eligible births (27,856 tested) in 2023-24, 28,313 eligible births (28,178 tested) in 2022-23 and 29,716 eligible births (29,587 tested) in 2021-22.

### Comment

There were 158 babies that were not tested in this period. Parents declined screening in 59 newborns and 59 babies that moved into Wales from outside the UK. Sadly, there were 22 deaths after day 5.

A suspended status was recorded for 18 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 2023 to March 2024

Health Board	Births	Tested	Rate (%)
Aneurin Bevan	5,697	5,678	99.7
Betsi Cadwaladr	5,782	5,750	99.4
Cardiff & Vale	4,655	4,621	99.3
Cwm Taf	4,044	4,032	99.7
Hywel Dda	3,077	3,052	99.2
Powys	983	971	98.8
Swansea Bay	3,358	3,336	99.3
Wales	28,014	27,856	99.4

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 14 of life.

Standard: 99%

### Results for 2023-24

All Wales rate: 97.5%

### Three-year trend

All Wales rates: 97.5% in 2023-24, 96.5% in 2022-23 and 97.2% in 2021-22.

### 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.

### 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.

The sample courier service introduced during the previous year was found to have a positive impact on transit times. Improved transit times were apparent during the Christmas period; this compared favourably to previous years where substantial delays were commonplace. Performance in April was particularly poor, linked to disruption of routine services during the Easter period.

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 14 of life in 2023-24

Health Board	Births	Offer in time	Rate (%)
Aneurin Bevan	5,697	5,576	97.9
Betsi Cadwaladr	5,782	5,676	98.2
Cardiff & Vale	4,655	4,490	96.5
Cwm Taf	4,044	3,958	97.9
Hywel Dda	3,077	3,022	98.2
Powys	983	952	96.8
Swansea Bay	3,358	3,235	96.3
Wales	28,014	27,313	97.5

Table 3: Eligible newborn babies offered bloodspot screening by day 14 of life – A comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	97.3	96.5	97.9
Betsi Cadwaladr	96.8	96.8	98.2
Cardiff & Vale	96.3	95.9	96.5
Cwm Taf	98.2	97.6	97.9
Hywel Dda	97.9	97.6	98.2
Powys	97.8	96.9	96.8
Swansea Bay	96.8	94.7	96.3
Wales	97.2	96.5	97.5



## Completeness of offer (all babies)

### Definition and standard

The percentage of all babies up to 1 year of age who have a notification of receipt of the bloodspot card in the laboratory within 18 days of registration.

Standard: 99%

### Results for 2023-24

All Wales rate: 98.4%

### Three-year trend

All Wales rates: 98.4% in 2023-24, 98.6% in 2022-23 and 98.7% in 2021-22.

### Geographic overview

No Health Boards achieved this standard, with all narrowly missing the required level.

### Comment

The sample courier service provides opportunities to improve performance against this standard. Additionally, the reduced time before a sample is chased following a change to the delayed sample failsafe trigger may support future improvements, with a slight improvement noted between January and March 2024.

Older babies who move into Wales are disproportionately represented in the cohort of babies whose bloodspot card does not arrive within the standard timeframe. 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 4: Eligible babies offered bloodspot screening within 18 days of registration in 2023-24

Health Board	Babies	Offer in time	Rate (%)
Aneurin Bevan	11,570	11,424	98.7
Betsi Cadwaladr	11,739	11,587	98.7
Cardiff & Vale	9,420	9,216	97.8
Cwm Taf	8,237	8,145	98.9
Hywel Dda	6,127	6,054	98.8
Powys	1,932	1,900	98.3
Swansea Bay	6,645	6,483	97.6
Wales	56,397	55,519	98.4

Table 5: Eligible babies offered bloodspot screening within 18 days of registration – a comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	98.8	98.7	98.7
Betsi Cadwaladr	98.9	98.8	98.7
Cardiff & Vale	98.4	98.0	97.8
Cwm Taf	99.1	99.1	98.9
Hywel Dda	98.9	98.8	98.8
Powys	98.9	98.9	98.3
Swansea Bay	98.1	97.8	97.6
Wales	98.7	98.6	98.4



## Coverage (newborns)

### Definition and standard

The percentage of newborn babies who have a conclusive bloodspot screening result by day 17 of life.

Standard: 95%

### Results for 2023-24

All Wales rate: 95.2%

### Three-year trend

All Wales rates: 95.2% in 2023-24, 95.0% in 2022-23 and 95.9% in 2021-22.

### Geographic overview

Three of the seven Health Boards achieved this standard, with the others narrowly missing the required level. Performance varied across the year for all organisations, with no Health Board consistently achieving 95% and all either maintaining or decreasing performance compared with the previous year. During June, September and March every Health Board achieved the standard. Performance dipped most significantly during April, with only one Health Board achieving the standard.

### 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 when timeliness is affected by extended bank holiday weekends. This was apparent during April where service delivery was affected by Easter. However, the positive effect of the new sample courier service was observed for babies born in December, with much improved performance compared to previous years despite the 4-day weekend.

Going forward, the continued use of the sample courier and changes to the delayed sample failsafe will provide opportunities for improvement.

## Figures and tables

Table 6: Eligible newborn babies with a conclusive result by day 17 of life in 2023-24

Health Board	Births	Result in time	Rate (%)
Aneurin Bevan	5,697	5,467	96.0
Betsi Cadwaladr	5,782	5,532	95.7
Cardiff & Vale	4,655	4,398	94.5
Cwm Taf	4,044	3,838	94.9
Hywel Dda	3,077	2,966	96.4
Powys	983	930	94.6
Swansea Bay	3,358	3,162	94.2
Wales	28,014	26,674	95.2

Table 7: Eligible newborn babies with a conclusive result by day 17 of life – a comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	96.4	95.4	96.0
Betsi Cadwaladr	95.4	95.2	95.7
Cardiff & Vale	95.2	94.3	94.5
Cwm Taf	96.5	95.9	94.9
Hywel Dda	96.7	95.3	96.4
Powys	96.0	96.3	94.6
Swansea Bay	95.4	92.6	94.2
Wales	95.9	95.0	95.2



## Coverage (all babies)

### Definition and standard

The percentage of all babies who have a conclusive bloodspot screening result within 21 days of registration.

Standard: 95%

### Results for 2023-24

All Wales rate: 96.5%

### Three-year trend

All Wales rates: 96.5% in 2023-24, 96.8% in 2022-23 and 97.2% in 2021-22.

### 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 contributing for some health boards.

### 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 21 days of registration in 2023-24

Health Board	Babies	Result in time	Rate (%)
Aneurin Bevan	11,570	11,247	97.2
Betsi Cadwaladr	11,739	11,331	96.5
Cardiff & Vale	9,420	9,041	96.0
Cwm Taf	8,237	7,960	96.6
Hywel Dda	6,127	5,942	97.0
Powys	1,932	1,870	96.8
Swansea Bay	6,645	6,344	95.5
Wales	56,397	54,423	96.5

Table 9: Eligible babies with a conclusive result within 21 days of registration – a comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	97.6	97.4	97.2
Betsi Cadwaladr	96.8	96.6	96.5
Cardiff & Vale	97.0	96.4	96.0
Cwm Taf	97.6	97.4	96.6
Hywel Dda	97.5	97.0	97.0
Powys	97.2	97.4	96.8
Swansea Bay	96.8	95.9	95.5
Wales	97.2	96.8	96.5



## 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 0).

Standard: 95%

### Results for 2023-24

All Wales rate: 95.1%

### Three-year trend

All Wales rates: 95.1% in 2023-24, 95.2% in 2022-23 and 95.2% in 2021-22.

### 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. In most cases, performance fluctuated through the year, with some dips during school holidays where staffing levels can be reduced. One Health Board consistently achieved the standard throughout the year, with one other Health Board failing to meet the standard every month.

### 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, in line with recommended practice. A significant number of samples were taken on day 7, falling just outside the standard. A notable proportion were taken on day 10 or later, suggesting that these babies were initially missed.

Performance data for sample collection timeliness is fed back each month 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 in 2023-24

Health Board	Samples	Collection in time	Rate (%)
Aneurin Bevan	5,664	5,397	95.3
Betsi Cadwaladr	5,613	5,346	95.2
Cardiff & Vale	4,626	4,318	93.3
Cwm Taf	4,025	3,864	96.0
Hywel Dda	3,069	2,997	97.7
Powys	905	863	95.4
Swansea Bay	3,336	3,128	93.8
Wales	27,549	26,205	95.1

Table 11: First sample between day 4 and 6 – a comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	96.2	95.3	95.3
Betsi Cadwaladr	92.6	94.7	95.2
Cardiff & Vale	93.9	93.4	93.3
Cwm Taf	96.4	96.0	96.0
Hywel Dda	97.1	96.9	97.7
Powys	96.3	96.9	95.4
Swansea Bay	96.4	95.9	93.8
Wales	95.2	95.2	95.1

Table 12: Day distribution of first sample in 2023-24

<4	4	5	6	7	8	9	10	>10	Total
53	6,617	16,329	3,259	510	129	63	39	550	27,549
0.2%	24.0%	59.3%	11.8%	1.9%	0.5%	0.2%	0.1%	2.0%	



## 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 3 calendar days of the request.

Standard: 95%

### Results for 2023-24

All Wales rate: 79.8%

### Three-year trend

All Wales rates: 79.8% in 2023-24, 71.3% in 2022-23 and 72.4% 2021-22.

### Geographic overview

Performance against this standard at a Health Board level remains variable. Small numbers in Powys (Table 20) 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.

Table 15 shows that a significant proportion of delayed samples are captured on the fourth day, or that there is an extended time before they are obtained (over 10 days).



## Figures and tables

Table 13: Avoidable repeat sample within three calendar days of request in 2023-24

Health Board	Rate (%)
Aneurin Bevan	83.7
Betsi Cadwaladr	75.2
Cardiff & Vale	77.8
Cwm Taf	74.2
Hywel Dda	86.7
Powys	86.4
Swansea Bay	83.5
Wales	79.8

Table 14: Avoidable repeat sample within three calendar days of request – a comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	88.9	81.6	83.7
Betsi Cadwaladr	33.6	62.6	75.2
Cardiff & Vale	69.6	69.9	77.8
Cwm Taf	82.5	76.4	74.2
Hywel Dda	80.9	76.0	86.7
Powys	88.0	76.5	86.4
Swansea Bay	84.5	66.9	83.5
Wales	72.4	71.3	79.8

Table 15: Day distribution of avoidable repeat sample in 2023-24

<=3	4	5	6	7	8	9	10	>10	Total
1,106	43	19	25	12	5	8	4	154	1,376
80.4%	3.1%	1.4%	1.8%	0.9%	0.4%	0.6%	0.3%	11.2%	



# 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 28 of life or day of discharge if earlier.

Standard: 95%

## Results for 2023-24

All Wales rate: 63.1%

## Three-year trend

All Wales rates: 63.1% in 2023-24, 62.5% in 2022-23 and 59.7% in 2021-22.

## Geographic overview

At an all Wales level, performance did not reach the standard throughout the year. One individual Health Board consistently achieved the standard, with the remaining organisations meeting it for at least 1 month. 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

Figures for the current year are improved in comparison to previous years, but further work is required. 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 28 of life – a comparison across previous years 2021-22 to 2023-24

	Rate 2021-22	Rate 2022-23	Rate 2023-24
Wales	59.7	62.5	63.1

Table 17: Day distribution of testing of second CHT sample for pre-term babies in 2023-24

<28	28	29	30	31	32	33	34	35	>35	Total
46	106	28	16	6	5	6	1	8	16	238



## 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 7 and 10 days after the initial borderline sample.

Standard: 95%

### Results for 2023-24

All Wales rate: 74.6 %

### Three-year trend

All Wales rates: 74.6 % in 2023-24, 79.0% in 2022-23 and 71.4% in 2021-22.

### 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 three-year trend shows variable performance. Close engagement between the programme team, laboratory colleagues and Health Board staff continues, in order to improve this position.

## Figures and tables

Table 18: Second sample for borderline TSH between 7 and 10 days of initial sample in 2023-24

	Second TSH	Second in time	Rate (%)
Wales	71	53	74.6

Table 19: Second sample for borderline TSH between 7 and 10 days of initial sample a comparison across previous years 2021-22 to 2023-24

	Rate 2021-22	Rate 2022-23	Rate 2023-24
Wales	71.4	79.0	74.6



## Avoidable repeat rate

### Definition and standard

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

Standard:  $\leq 2\%$

### Results for 2023-24

All Wales rate: 4.7%

### Three-year trend

All Wales rates: 4.7% in 2023-24, 2.8% in 2022-23 and 2.4% in 2021-22.

### Geographic overview

Performance against this standard was consistently poor throughout the year. Although there was variability between Health Board areas, standard achievement was sporadic with no organisation managing to sustain performance. Three Health Boards experienced particularly elevated avoidable repeat rates.

### Comment

Repeat rate rises are primarily due to poor quality samples, which includes samples with an insufficient amount of blood captured. Figure 2 shows that, compared to previous years, there was an increase in the number of samples rejected due to documentation errors with the NHS number. This is attributed to a tightening of the protocol associated with these criteria, following high adherence to this standard in previous years.

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.

The publication of both a sample taker performance and assurance framework and visual guide sample acceptance criteria in January 2024 provide helpful tools for Health Boards to support improvements to sample taker practice.

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.

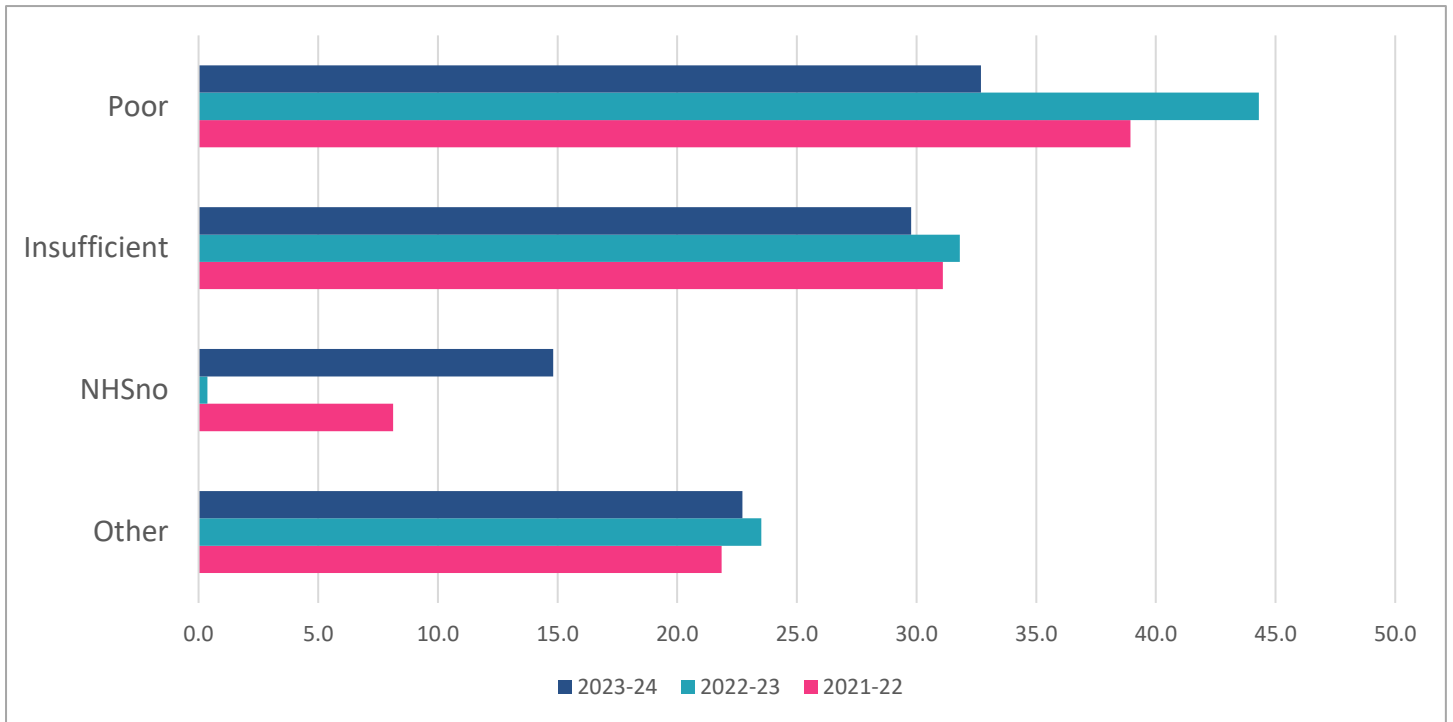
## Figures and tables

**Table 20: Avoidable repeat rate in 2023-24**

Health Board	Cards	Repeats	Rate (%)
Aneurin Bevan	6,036	209	3.5
Betsi Cadwaladr	6,043	274	4.5
Cardiff & Vale	4,965	275	5.5
Cwm Taf	4,307	217	5.0
Hywel Dda	3,245	181	5.6
Powys	964	44	4.6
Swansea Bay	3,557	176	4.9
Wales	29,117	1,376	4.7

**Table 21: Avoidable repeat rate – a comparison across previous years 2021-22 to 2023-24**

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	1.9	1.6	3.5
Betsi Cadwaladr	2.0	2.4	4.5
Cardiff & Vale	3.0	3.9	5.5
Cwm Taf	2.6	2.4	5.0
Hywel Dda	2.1	3.2	5.6
Powys	2.3	1.8	4.6
Swansea Bay	2.9	3.9	4.9
Wales	2.4	2.8	4.7



**Figure 2: Reasons for avoidable repeat, percentage of all repeat samples requested 2021-22 to 2023-24**

‘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.

Note: an individual sample card can be rejected for more than one reason. For example, if a sample is both poor quality and the NHS number is missing, the same sample card is recorded against 2 separate reasons in figure 2.

## Poor quality repeat rate

### Definition and standard

Repeat cards that are required because of poor quality bloodspots (including insufficient sampling).

Standard:  $\leq 1.5\%$  (formerly  $\leq 2\%$  until November 2021)

### Results for 2023-24

All Wales rate: 3.0%

### Three-year trend

All Wales rates: 3.0% in 2023-24, 2.1% in 2022-23 and 1.7% in 2021-22.

### Geographic overview

As also seen in the avoidable repeat rate, there was consistently poor performance against this standard in all months, and for all Health Boards.

### 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.

Note: a single card can have multiple reasons for repeat. The large increase in missing or inaccurate NHS numbers in 2023-24 explains why the poor quality repeat reason as a percentage of all recorded reasons appears proportionally lower than previous years (Figure 2) but the overall poor quality repeat rate has increased.



## Figures and tables

Table 22: Poor quality repeat rate in 2023-24

Health Board	Cards	Repeats	Rate (%)
Aneurin Bevan	6,036	125	2.1
Betsi Cadwaladr	6,043	164	2.7
Cardiff & Vale	4,965	174	3.5
Cwm Taf	4,307	119	2.8
Hywel Dda	3,245	139	4.3
Powys	964	31	3.2
Swansea Bay	3,557	123	3.5
Wales	29,117	875	3.0

Table 23: Poor quality repeat rate – a comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	1.5	1.1	2.1
Betsi Cadwaladr	1.3	1.9	2.7
Cardiff & Vale	2.1	3.3	3.5
Cwm Taf	1.8	1.9	2.8
Hywel Dda	1.8	2.5	4.3
Powys	1.6	1.6	3.2
Swansea Bay	1.7	2.6	3.5
Wales	1.7	2.1	3.0



GIG  
CYMRU  
NHS  
WALES

Iechyd Cyhoeddus  
Cymru  
Public Health  
Wales

## 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% until November 2021)

### Results for 2023-24

All Wales rate: 99.3%

### Three-year trend

All Wales rates: 99.3% in 2023-24, 99.9% in 2022-23 and 99.8% in 2021-22.

### Comment

This standard was achieved at an all Wales and individual Health Board level. Performance was down slightly compared to the previous year, which is attributed to a tightening of the criteria application in response to exceptional performance in 2022-23.



## Figures and tables

Table 24: NHS number on card in 2023-24

Health Board	Cards	NHS number	Rate (%)
Aneurin Bevan	6,057	6,028	99.5
Betsi Cadwaladr	6,073	6,020	99.1
Cardiff & Vale	5,002	4,962	99.2
Cwm Taf	4,322	4,287	99.2
Hywel Dda	3,278	3,254	99.3
Powys	976	968	99.2
Swansea Bay	3,586	3,567	99.5
Wales	29,294	29,086	99.3

Table 25: NHS number on card – a comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	99.8	99.9	99.5
Betsi Cadwaladr	99.8	100	99.1
Cardiff & Vale	99.8	99.9	99.2
Cwm Taf	99.7	100	99.2
Hywel Dda	99.9	100	99.3
Powys	99.8	100	99.2
Swansea Bay	99.8	100	99.5
Wales	99.8	99.9	99.3



GIG  
CYMRU  
NHS  
WALES

Iechyd Cyhoeddus  
Cymru  
Public Health  
Wales

## 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 2023-24

All Wales rate: 91.4%

### Three-year trend

All Wales rates: 91.4% in 2023-24, 85.1% in 2022-23 and 85.1% in 2021-22.

### Comment

This standard was not met during the period, which reflects the position of previous years. 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.

From December onwards there was a marked improvement across all Health Boards utilising the sample courier service. This reflects harmonisation of the sample collection schedule across all sites as part of ongoing improvement activity linked to this service change.

## Figures and tables

Table 26: Lab receipt of card within 3 working days in 2023-24

Health Board	Cards	Receipt in time	Rate (%)
Aneurin Bevan	6,057	5,348	88.3
Betsi Cadwaladr	6,073	5,788	95.3
Cardiff & Vale	5,002	4,824	96.4
Cwm Taf	4,322	3,656	84.6
Hywel Dda	3,278	2,964	90.4
Powys	976	880	90.2
Swansea Bay	3,586	3,325	92.7
Wales	29,294	26,785	91.4

Table 27: Lab receipt of card within 3 working days – a comparison across previous years 2021-22 to 2023-24

Health Board	Rate 2021-22	Rate 2022-23	Rate 2023-24
Aneurin Bevan	83.7	81.6	88.3
Betsi Cadwaladr	76.0	81.2	95.3
Cardiff & Vale	92.5	94.1	96.4
Cwm Taf	89.2	84.4	84.6
Hywel Dda	87.5	86.7	90.4
Powys	86.6	82.8	90.2
Swansea Bay	84.8	85.0	92.7
Wales	85.1	85.1	91.4

Table 28: Day distribution of card receipt in the laboratory for 2023-24

<=3	4	5	6	7	8	9	10	>10	Total
26,785	1,842	445	88	51	24	13	18	28	29,294
91.4%	6.3%	1.5%	0.3%	0.2%	0.1%	0.04%	0.1%	0.1%	

## 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 25 days of sample receipt.

SCD: clinical referral for SCD screen positive results initiated within 42 days of sample receipt.

Standard (IMD/CHT): 100%

Standard (CF): 95%

Standard (SCD): 95%

### Results for 2023-24

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 2023-24, 100% in 2022-23 and 100% in 2021-22.

All Wales rates (CF): 100% in 2023-24, 100% in 2022-23 and 100% in 2021-22.

All Wales rates (SCD): 100% in 2023-24, 100% in 2022-23 and 100% in 2021-22.

### Comment

All standards were achieved. As can be seen from the three-year 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 in 2023-24

Condition	Positive	Referral in time	Rate (%)
IMD & CHT	41	41	100
CF	23	23	100
SCD	3	3	100

Table 30: Timely processing of positive samples – a comparison across previous years 2021-22 to 2023-24

Condition	Rate 2021-22	Rate 2022-23	Rate 2023-24
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 14 of life (excluding HCU).

CHT: first clinical appointment attendance for CHT screen positive results by day 14 of life or initial borderline results followed by a positive result by day 21.

CF: first clinical appointment attendance for CF screen positive results by day 28 of life.

SCD: first clinical appointment attendance for SCD screen positive results by day 90 of life.

Standard (IMD): 100%

Standard (CHT): 100%

Standard (CF): 95%

Standard (SCD): 90%

### Results for 2023-24

All Wales rate (IMD): 100%

All Wales rate (CHT): 93.3%

All Wales rate (CF): 78.3%

All Wales rate (SCD): 100%

### Three-year trend

All Wales rates (IMD): 100% in 2023-24, 100% in 2022-23 and 100% in 2021-22.

All Wales rates (CHT): 93.3% in 2023-24, 84.6% in 2022-23 and 94.1% in 2021-22.

All Wales rates (CF): 78.3% in 2023-24, 81.3% in 2022-23 and 76.5% in 2021-22.

All Wales rates (SCD): 100% in 2023-24, 100% in 2022-23 and 100% in 2021-22.



## 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.

The CHT standard was not achieved for 2 babies due to the late arrival (after day 14 of life) of the sample cards at the laboratory.

All babies receiving a screen positive result for CF were referred to a Health Board for diagnostic assessment by day 23 of life. Some Health Boards book referred babies into a specialist clinic for their diagnostic sweat test, rather than performing the test as a separate assessment. The timing of these clinics can mean that babies are not seen by 28 days of age.

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 provision of clinical care for screen positive babies in 2023-24

Condition	Positive	Referral in time	Rate (%)
IMD <sup>1</sup>	8	8	100
CHT	30	28	93.3
CF	23	18	78.3
SCD	3	3	100

<sup>1</sup> Excluding homocystinuria (HCU).

Table 32: Timely provision of clinical care for screen positive babies – a comparison across previous years 2021-22 to 2023-24

Condition	Rate 2021-22	Rate 2022-23	Rate 2023-24
IMD	87.5	100	100
CHT	94.1	84.6	93.3
CF	76.5	81.3	78.3
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.

## 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 mucus. 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



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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.

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