Newborn Bloodspot Screening Wales
Sgrinio Smotyn Gwaed Newydd-anedig Cymru

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Newborn Bloodspot Screening Wales Annual Statistical Report 2017-18

November 2018

Screening Division of Public Health Wales Official Statistics Publication

About us

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

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

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

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

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

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

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

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

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

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

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

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

Further information

Web: www.publichealthwales.org
Email: general.enquiries@wales.nhs.uk
Twitter: @PublichealthW
Facebook: www.facebook.com/PublicHealthWales

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

This report is a detailed summary of information on work undertaken by Newborn Bloodspot Screening Wales for the financial year from April 2017 to the end of March 2018. Results are reported by Health Board where screening has been carried out. Further details are available on request.

Publication Details:

Title: Newborn Bloodspot Screening Wales Annual Statistical Report 2017-18

Date: This report published 22nd November 2018

ISBN: 978-1-78986-008-5

For more information and to feedback about this report contact:
Helen Clayton, Lead Informatics and Data Services Manager,
Informatics Division, Floor 6, Public Health Wales, Number 2 Capital Quarter, Tyndall Street, Cardiff, CF10 4BZ
Tel: 029 2010 4405
Email: Screening.Information@wales.nhs.uk

Quality Assurance Statement

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

© 2018 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.

Contents

Tables and Graphs

1 Introduction

This is the second annual statistical report published by Newborn Bloodspot Screening Wales (NBSW). This report covers data for babies born between 1st April 2017 and 31st March 2018.

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

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

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

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

1.1 Key messages for parents

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

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

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

1.2 Programme delivery

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

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

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

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

NBSWS also identifies babies for whom the programme expects to receive either a bloodspot card or decline for the test(s), and initiates failsafe procedures for possible 'missed' babies. This failsafe system identifies babies in Wales who do not have a newborn bloodspot screening sample in the Newborn Screening Laboratory by day 14 of life. Every baby identified by the failsafe is followed up by the administration failsafe teams. The three regional teams across Wales are staffed by newborn screening managers and administrative staff who work across both the NBSW and Newborn Hearing Screening Wales (NBSHW) programmes.

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

1.3 Screening pathway

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

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

Newborn bloodspot screening samples are sent by prepaid envelopes (first class) to the Wales Newborn Screening Laboratory in Cardiff for testing. The laboratory accepts samples according to the UK bloodspot quality guidelines for screening laboratories that were implemented in April 2015. Babies suspected of having one of the conditions screened for are referred, according to the relevant clinical referral guidelines, to the appropriate specialist clinician for diagnostic tests and treatment. The results for each baby are sent to the local Child Health Department and are entered onto the Child Health System. The baby's health visitor discusses the results with the parents.

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

2 Headline Statistics

April 2017 to March 2018

- The number of eligible births across Wales was 32,159
- The number of these babies tested was 31,932 (99.3%)

Screening

Completeness of offer and coverage (eligible newborns)

- Completeness of offer – 96.5% of babies had a bloodspot card (for screening or decline) received in the laboratory by day 14 of life
- Coverage – 93.8% of babies had conclusive bloodspot screening results by day 17 of life

Timeliness of sample collection

- Timely collection of sample (day 5-8 of life) – 97.9%
- Timely collection of sample (day 5 of life) – 78.4%

Avoidable repeat rate

- Avoidable repeat rate – 4.7%

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

NHS number on bloodspot card

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

Timely receipt of card in laboratory

- 93.9% of bloodspot cards were received within 4 working days of sample collection

Outcomes

The number of screen positive babies detected in the year was as follows: phenylketonuria (4), congenital hypothyroidism (19), medium-chain acyl-CoA dehydrogenase deficiency (4), cystic fibrosis (9), sickle cell disorders (3).

3 Data

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

Table 1: The number of eligible births in Wales in the period April 2017 to March 2018, and the number of these babies tested.

	Aneurin Bevan	Abertawe Bro Morgannwg	Betsi Cadwaladr	Cardiff & Vale	Cwm Taf	Hywel Dda	Powys	Wales
Births	6389	5483	6775	5413	3231	3509	1123	32159
Tested	6347	5460	6716	5370	3216	3477	1111	31932
%	99.3	99.6	99.1	99.2	99.5	99.1	98.9	99.3

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

3.1 Standards

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

Table 2: Programme performance standards

NBSW Standards – Screening Programme					
	Objective	**Criteria**	**Minimum Standard**	**Actual Value**	**Variance from 2016-17**
1A	Completeness of offer (Newborns)	The percentage of eligible newborn babies who have a notification of receipt of the bloodspot card in the laboratory by day 14 of life	99%	96.5%	-0.8
1B	Completeness of Offer (All)	Eligible babies (up to one year of age) who have a notification of receipt of the bloodspot card in the laboratory within 18 days of registration	99%	97.1%	-1.3
1C	Coverage (Newborns)	Eligible newborn babies who have a conclusive bloodspot screening result by day 17 of life	95%	93.8%	-0.2

1D	Coverage (All)	Eligible babies (up to one year of age) who have a conclusive bloodspot screening result within 21 days of registration	95%	94.8%	-0.8
3A	Timely Collection of Sample (Day 5-8 of Life)	The first bloodspot sample should be taken between day 5 and day 8 of life (counting day of birth as day 0)	95%	97.9%	-0.1
3F	Timely Collection of Sample (Day 5 of Life)	The first bloodspot sample should be taken on day 5 of life (counting day of birth as day 0)	90%	78.4%	+3.1
3B	Timely Collection of Avoidable Repeat Samples	Repeat testing for insufficient/poor quality samples or incomplete/incorrect card information should be conducted within 3 calendar days of the request	95%	64.4%	+3.5
3C	Timely CHT Second Sample Collection for Pre-Term Babies	Pre-term babies with a second bloodspot card received in the laboratory which was taken at day 28 of life or on day of discharge	95%	2017-18 performance not reportable	
3D	Timely Second Sample Collection for Borderline TSH	Babies with a borderline TSH result who have a second bloodspot card for TSH received in the laboratory which was collected between 7 and 10 days after the initial borderline sample	95%	75%	-25
4A	Avoidable Repeat Rate	Repeat cards that are required because of poor quality bloodspots or incomplete/incorrect information recorded	<=2%	4.7%	-0.8 (an improvement)
4B	Poor Quality Repeat Rate	Repeat cards that are required because of poor quality bloodspots	<=2%	3.2%	-1.0 (an improvement)

4C	NHS Number on Bloodspot Card	Bloodspot cards received in the laboratory that have a valid NHS number for the baby recorded	100%	99.1%	no change
5	Timely Receipt of Card in Laboratory	Bloodspot cards received within 4 working days	99%	93.9%	-1.7
6A	Timely Processing of IMD and CHT Positive Samples	Clinical referral for IMD/CHT screen positive results initiated within 3 working days of sample receipt	100%	100%	no change
6B	Timely Processing of CF Positive Samples	Clinical referral for CF screen positive results initiated within 25 days of sample receipt	95%	100%	no change
6C	Timely Processing of SCD Positive Samples	Clinical referral for SCD screen positive results initiated within 42 days of sample receipt	95%	100%	no change
7A	Timely Clinical Care Receipt of IMD Positive Babies	First clinical appointment attendance for IMD screen positive results by day 17 of life	100%	100%	no change
7B	Timely Clinical Care Receipt of CHT Positive Babies	First clinical appointment attendance for CHT screen positive results by day 17 of life or initial borderline results followed by a positive by day 24	100%	100%	no change
7C	Timely Clinical Care Receipt of CF Positive Babies	First clinical appointment attendance for CF screen positive results by day 28 of life	95%	88%	+13
7D	Timely Clinical Care Receipt of SCD Positive Babies	First clinical appointment attendance for SCD screen positive results by day 56 of life	90%	100%	no change

3.2 Completeness of Offer and Coverage

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

Table 3: Babies offered and completing newborn bloodspot screening

Health Board	% Offered (Newborn)	% Offered (All)	% Coverage (Newborn)	% Coverage (All)
Abertawe Bro Morgannwg University Health Board	97.4	97.5	94.3	95.3
Aneurin Bevan University Health Board	96.0	97.1	93.2	94.6
Betsi Cadwaladr University Health Board	95.7	97.2	92.9	94.7
Cardiff and Vale University Health Board	96.2	96.3	94.2	94.7
Cwm Taf University Health Board	97.2	98.4	94.6	96.2
Hywel Dda University Health Board	97.5	97.3	94.2	94.7
Powys Teaching Health Board	96.8	96.2	95.0	94.7
All Wales	96.5	97.1	93.8	94.8

The All Wales figures show that the standards for offer of screening have not been met and there is a slight decrease of 0.8% (newborns) and 1.3% (all babies) compared with the previous year. Work to improve timeliness of sample collection and dispatch is continuing.

The standards for coverage have not been met this year. Across Wales, 93.8% of newborn babies had screening completed in the specified

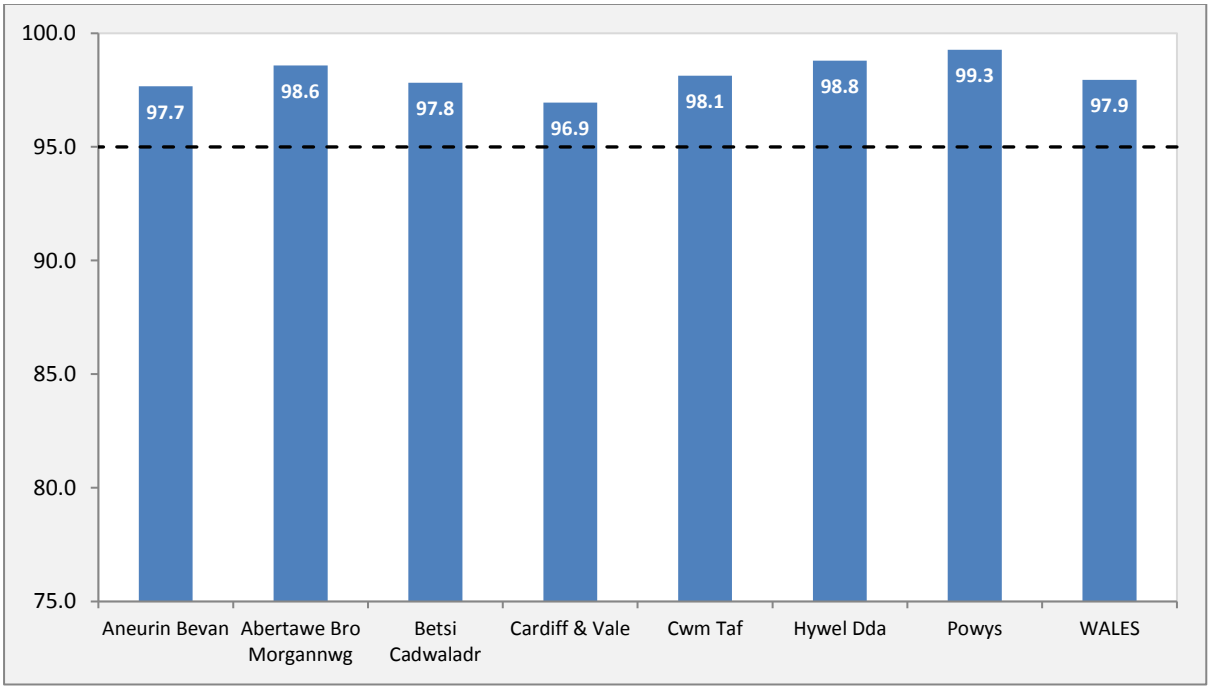
timeframe which is a small decrease of 0.2% compared with the previous year. The relatively high avoidable repeat rate and timeliness issues in collecting repeat samples have an impact on performance in coverage, and addressing these problems continues to be a high priority for the programme.

3.3 Timeliness of testing

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

Standard 3A

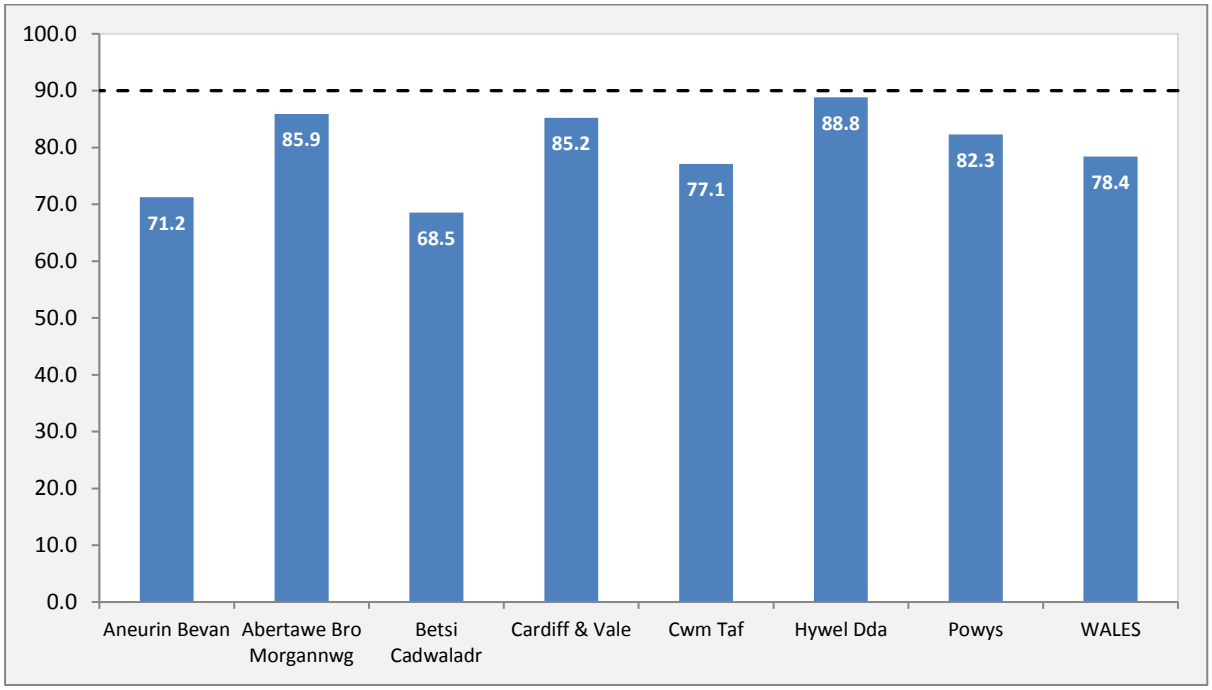
Graph 1: Timely Collection of Samples (Day 5 – 8 of Life)



This standard has been met in all of the health boards. Across Wales, 97.9% of samples were taken between day 5 and day 8 of life. The programme is working with the health boards to improve timeliness of sample collection, with the emphasis on taking the sample on day 5 to enable earlier identification and referral of screen positive babies.

Standard 3F

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



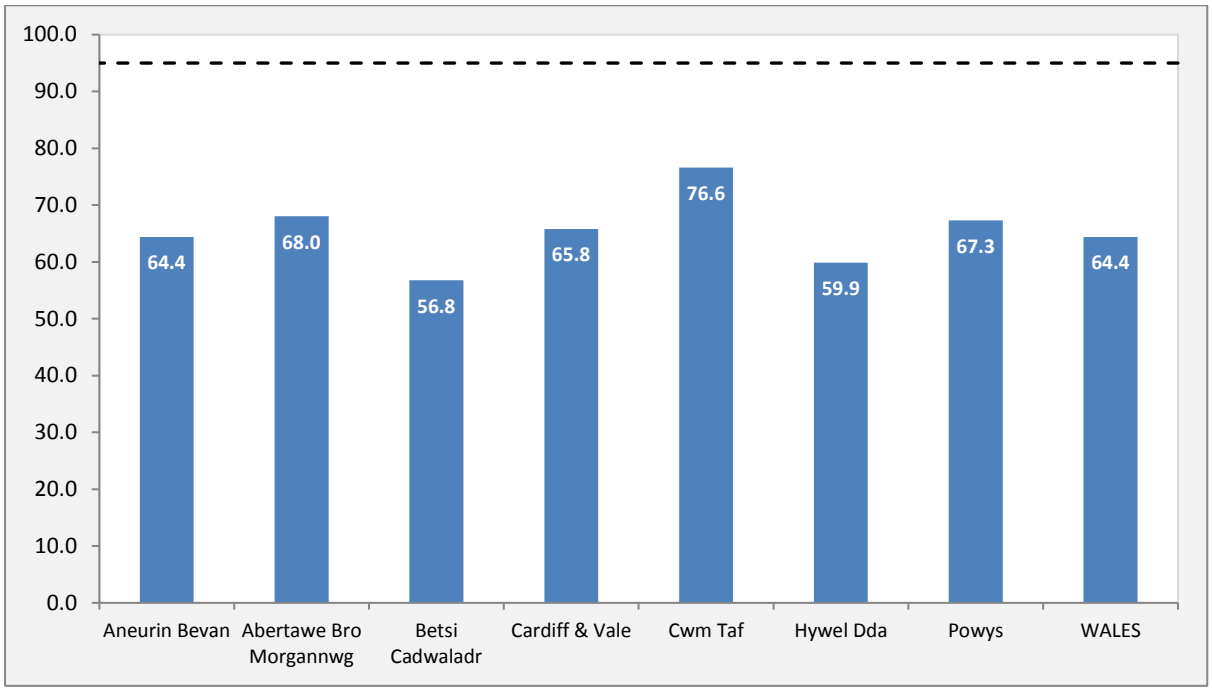
To monitor performance in timely collection of samples more closely, data is collected for sample collection at day 5 of life. Improving performance in this is a high priority for the programme to enable timely referral of screen positive babies to clinical care.

Across Wales, 78.4% of samples were taken at day 5 of life which is an improvement of 3.1% compared with the previous year. This improvement has been across Wales and in six of the seven health boards.

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 improve performance, and this has included the education of sample takers of the importance of taking samples on day 5.

Standard 3B

Graph 3: Timely Collection of Avoidable Repeat Samples



Across Wales, 64.4% avoidable repeat samples were taken within 3 calendar days of the request. The standard has not been met but there is an improvement of 3.5% on the previous year.

To improve timeliness in the collection of avoidable repeat samples, the process for requesting the repeats was reviewed and a new process was implemented on 27th June 2017. The requests had previously been made by fax to the maternity or neonatal unit in the health board, which was problematic and associated with delays in actioning these requests.

All requests for repeat samples are now made to designated generic email addresses in the maternity units and neonatal units. The emails contain an image of the card, details of the sample problem and advice for the sample taker to improve performance. The health board governance lead is copied into the email to enable monitoring of sample taker performance and timely identification of training needs.

The programme is working with the health boards and the Newborn Screening Laboratory to improve performance in timeliness of avoidable repeat sample collection. This includes the review of the process and identification of factors that impact on timeliness. Audits have been

conducted in the laboratory to identify any factors that may result in delays in the repeat requesting process, so that they can be managed to enable timely receipt of requests in the health boards.

Standard 3C

Timely CHT Second Sample Collection for Pre-Term Babies

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

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

Day of life second CHT sample taken										
<28*	28	29	30	31	32	33	34	35	>35	Total
27	79	41	20	10	10	6	5	12	14	224

* Performance by Health Board and all Wales has not been given for this standard as the IT systems were not able to capture all the required data during 2017-18.

Across Wales, 12% of second CHT samples were taken before day 28 of life, compared with 21% in the previous year. This data does not currently show if the samples were correctly taken from babies who were discharged before day 28 of life, or taken too early resulting in a request for a repeat sample. However, laboratory monthly feedback of avoidable repeat samples includes CHT second samples that have been taken too early. This showed a reduction in the number of these samples that were taken too early this year.

The programme is continuing to work with the neonatal units across Wales to improve performance so that CHT second samples are taken at the correct time. Meetings have been held with the Wales Neonatal Network Lead Nurse and Neonatal Unit managers across Wales to discuss newborn bloodspot screening performance and issues.

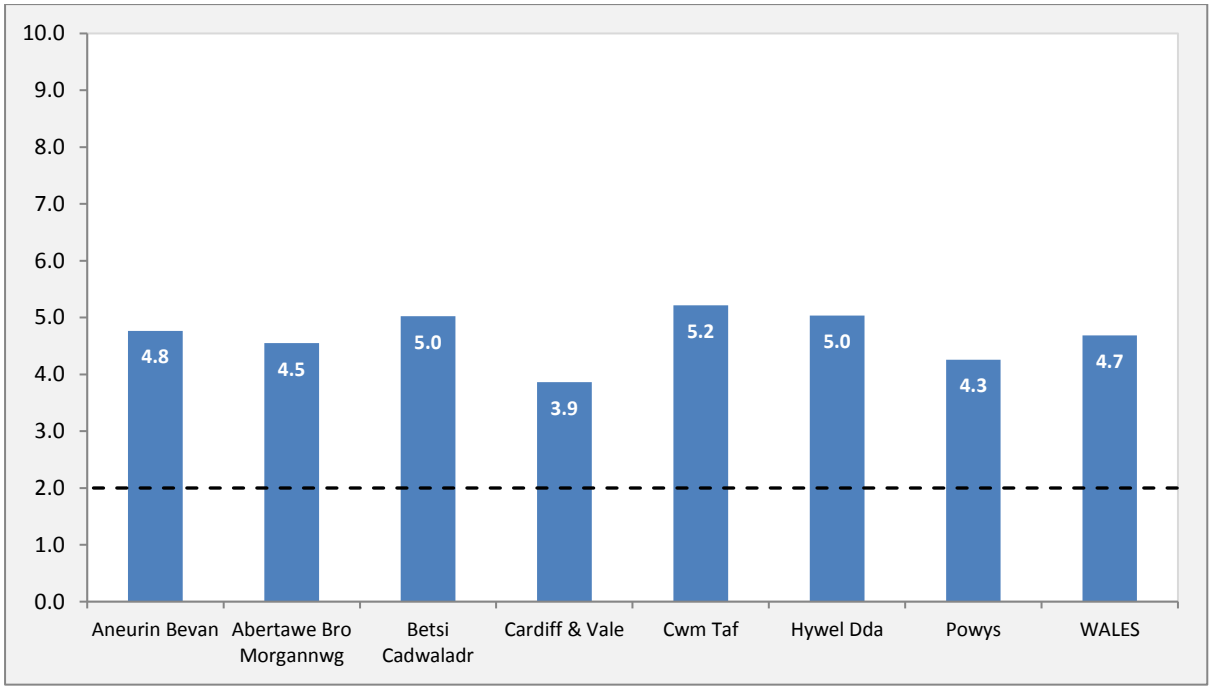
Work has been undertaken to produce a film for neonatal unit staff which looks at aspects of newborn bloodspot screening for babies who are cared for in neonatal units, and the additional requirements needed for screening these babies.

3.4 Poor Quality Repeat Samples Required

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

Standard 4A

Graph 4: Avoidable Repeat Rate



The avoidable repeat rate in Wales is 4.7%, a slight improvement of 0.8% compared with the previous year. Achieving the standard of ≤2% remains a high priority for the programme to avoid delays in the referral of babies and to avoid the other costs associated with repeating samples.

Work is continuing with the health boards to reduce the avoidable repeat rate. The focus is on improving the quality of the bloodspots as poor quality bloodspots are the main reason for an avoidable repeat. The governance leads and Heads of Midwifery are sent monthly sample quality performance reports to enable monitoring and appropriate action to be taken within the health boards. More specific sample quality information is now available to the governance leads in the emails requesting repeat samples. This has

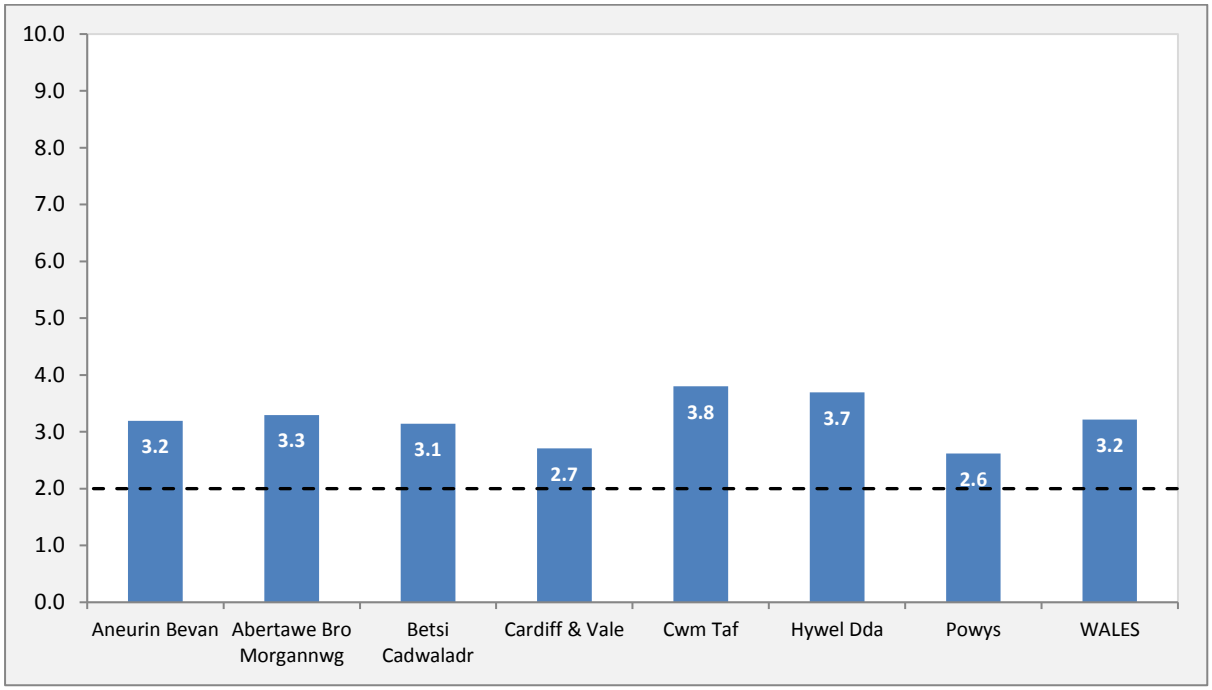
enabled the timely feedback of poor quality samples to specific sample takers so that training needs can be identified and addressed. Interventions in the health boards to improve sample quality have also included the review of lancets used and the review of training for sample takers.

NBSW has provided sample quality training sessions in the health boards and developed new training resources focusing on improving sample quality. A film has been produced which shows samples being processed in the laboratory and highlights why a good quality sample is important. NBSW also provides newborn bloodspot screening education to student midwives in the universities across Wales.

Samples taken on expired cards contribute to the avoidable repeat rate and the importance of checking the expiry date has been highlighted to sample takers. To minimise the use of expired cards, the redesigned bloodspot card now has the expiry date printed in red on the front so it is easily visible. This new card was first sent out to the health boards in April 2018 and so would not have had an impact for this year.

Standard 4B

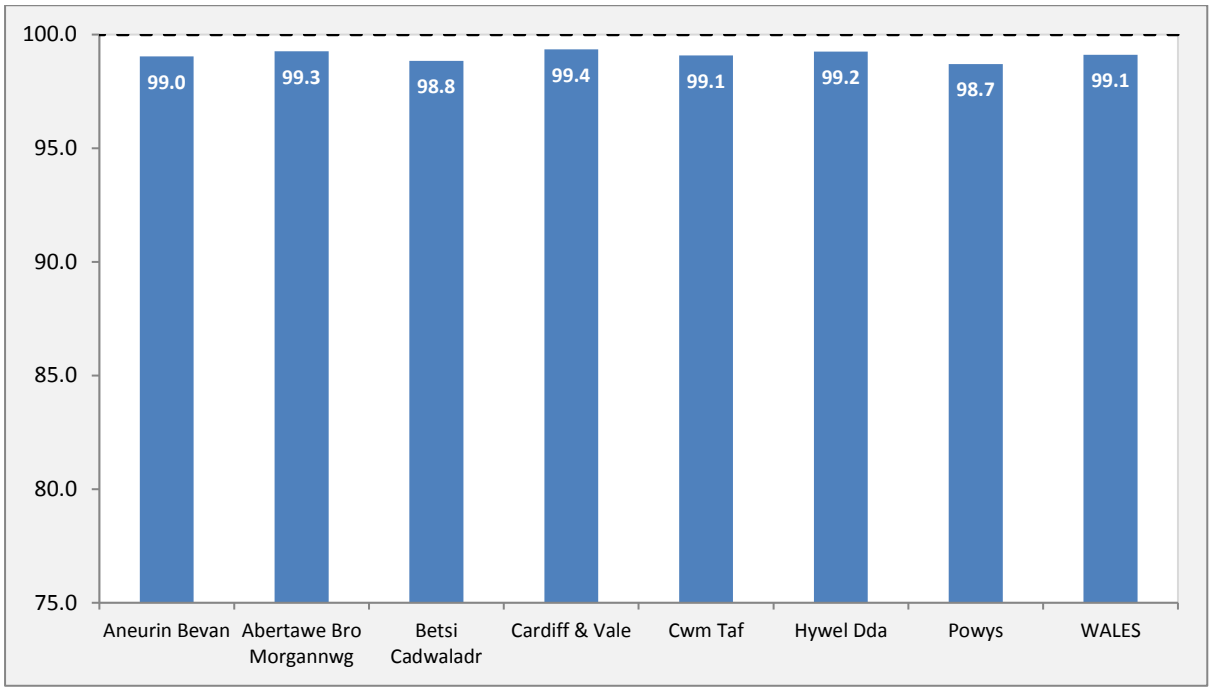
Graph 5: Poor Quality Repeat Rate



Across Wales, 3.2% samples required repeating due to poor quality bloodspots which is an improvement of 1% compared with the previous year. Reducing the poor quality rate further to achieve the standard of ≤2% is a high priority for the programme.

Standard 4C

Graph 6: NHS Number on Bloodspot Card



The valid NHS number for the baby was recorded on 99.1% bloodspot cards received in the laboratory. This rate is unchanged from the previous year. Illegibility of NHS numbers, inaccuracy or mixing up the numbers with another baby are the main reasons for non compliance. Work continues with the health boards to improve performance in this standard.

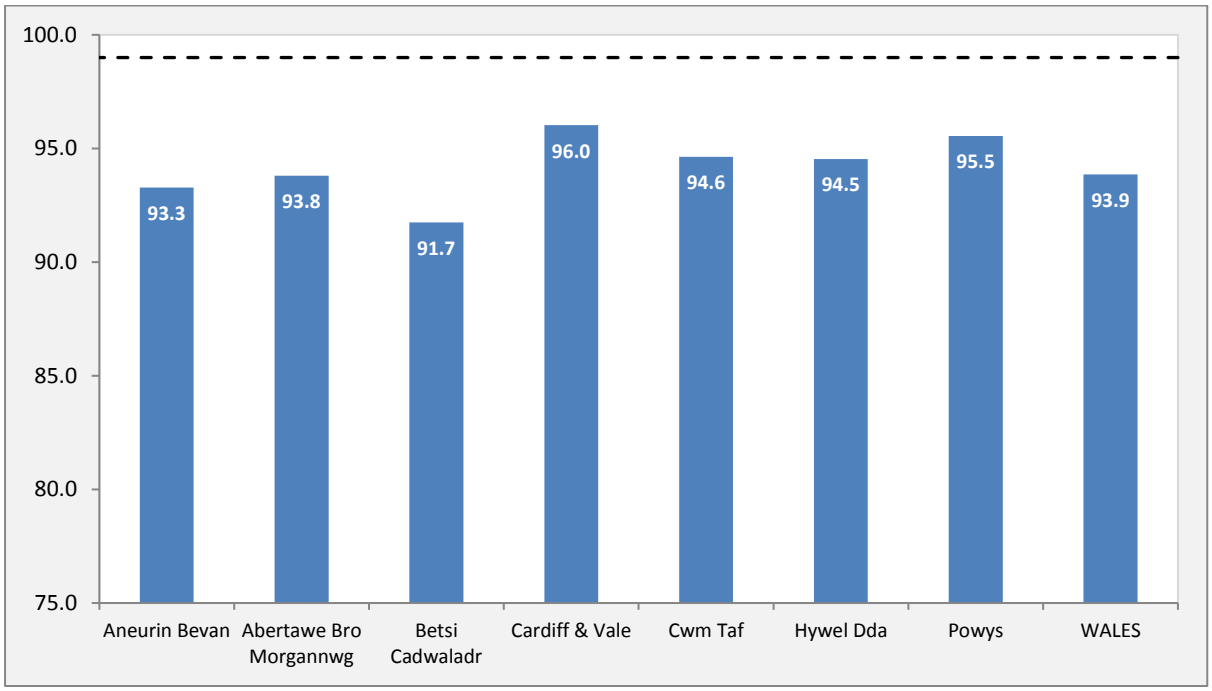
Following a scoping exercise looking at the potential use of bar coded NHS number labels for bloodspot screening in Wales, it was concluded that this is currently not a viable option. This was due to the feedback from health boards that there would be difficulties in the production of standardised labels specifically for use on bloodspot screening cards.

3.5 Timely Receipt of Card in Laboratory

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

Standard 5

Graph 7: Timely Receipt of Card in Laboratory



Across Wales, 93.9% samples were received in the laboratory within 4 working days. This is a decrease of 1.7% compared with the previous year. Improving performance in this standard is a high priority to enable timely referral of screen positive babies into clinical care.

To monitor performance more closely, additional data is collated for receipt of samples received within 3 working days of the sample being taken. This performance data is fed back quarterly to the health board governance leads and Heads of Midwifery.

Work continues with the health boards and Royal Mail to minimise delays in samples reaching the laboratory. Delays in sample receipt were significantly increased during the Christmas period and addressing these delays is a priority for the programme. Information for Royal Mail staff to highlight the importance of timely receipt is planned prior to Christmas 2018.

3.6 Laboratory Processing and Referral

The current standard for timely processing of samples was met for all conditions. For the inherited metabolic disorders a total of 4 phenylketonuria (PKU) cases and 4 medium-chain acyl-CoA dehydrogenase deficiency (MCADD) cases were identified. Confirmation on receipt into clinical care was available for 8 cases, (100%). The average age of timely receipt into clinical care was day 11 of life (range 10-15 days). No cases of maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (HCU) were identified during 2017/18.

Of the 19 congenital hypothyroidism (CHT) cases, 2 were identified as part of the CHT preterm policy, 14 were identified as having a raised TSH ≥20mU/L on the initial sample and 3 cases were identified with a borderline raised TSH on the initial sample with a positive repeat sample. Of the 17 cases identified as having a raised TSH all were referred into clinical care in a timely manner. Confirmation on receipt into clinical care was available for only 16 out of the 17 cases (94%). Of the 14 CHT cases with a raised TSH ≥20mU/L on the day 5 sample, the average age at the first clinic appointment was day 12 of life (range 9-15 days). The remaining 3 cases, identified following an initial borderline result, the average age at the first clinic appointment was day 21 of life (range 19-24 days).

The timely second sample collection for borderline TSH performance was poor at only 75% (standard = 95%), with 20% collected on or after day 11 (range 11-33 days). A few samples n=4 (5%) were collected before day 7.

A total of 9 cystic fibrosis (CF) cases were identified, with the mean age of referral into clinical care being day 21 of life (range 15-33 days). Confirmation on receipt into clinical care was available for only 8 out of the 9 cases (89%). The average age at the first clinic appointment was day 25 of life (range 18-34 days). The case received into clinical care on day 34 of life was due to the fact that the day 5 sample was of such poor quality and that the repeat sample was not collected until day 28 of life.

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

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

4 Definitions

Eligible babies (newborn)

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

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

Eligible babies (all)

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

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

Screen positive result

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

Screen negative result

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

Conclusive result

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

Parent/guardian surveys

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

The Conditions

Congenital hypothyroidism (CHT)

Congenital hypothyroidism (CHT) is a condition where the baby's thyroid gland fails to develop or work properly and fails to make the thyroid hormone called thyroxine. Thyroxine is needed for normal growth and development. Without thyroxine, babies do not grow properly and can develop permanent, serious physical problems and learning disabilities.

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

CHT has been screened for in Wales since 1981.

Cystic fibrosis (CF)

Cystic fibrosis (CF) is one of the UK's most common inherited life-limiting diseases. CF is a disease in which abnormal movement of salt and water into and out of cells causes a build-up of thick, sticky mucous. This occurs particularly in the lungs and digestive system. Babies with CF may not gain weight well, have frequent chest infections and a limited life span.

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

CF has been screened for in Wales since December 1996.

Inherited metabolic disorders (IMDs):

- **Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)**

  MCADD is a rare inherited condition in which there is a deficiency in the enzyme medium-chain acyl-CoA dehydrogenase which is needed for the breakdown of certain stored fats (medium-chain fatty acids). This makes it difficult for the body to break down fatty acids and produce energy, and can cause sudden death in infants. Fatty acids are an important energy reserve during periods of poor calorie intake, prolonged periods between meals or during infections and sickness. In these situations people with MCADD have high levels of partially broken down fatty acids and low blood glucose concentrations which can result in a metabolic crisis. Most of the time children are well, but an infection or relatively long period without food upsets their metabolism causing coma and sometimes death.

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

  MCADD has been screened for in Wales since June 2012.

- **Phenylketonuria (PKU)**

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

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

  PKU has been screened for in Wales since 1970.

- **Maple syrup urine disease (MSUD)**

  Maple syrup urine disease (MSUD) is a rare inherited disorder that prevents the breakdown of some of the building blocks of protein, the amino acids leucine, isoleucine and valine in the blood. For people with MSUD, eating normal amounts of protein can cause a harmful build-up of these amino acids in the blood. Many babies with MSUD become unwell when they are a few days old. Without treatment, this leads to a coma and permanent brain damage. In older children a minor illness,

such as a chest infection or a tummy upset, can lead to serious problems. As in babies, this can lead to a coma unless treated correctly.

MSUD can be treated with a protein-restricted diet. A different regime is required when the child is ill, and they may need to be hospitalised. The condition is named maple syrup urine disease because high levels of these amino acids can cause an unusual sweet smell in the urine and sweat.

MSUD has been screened for in Wales since January 2015.

- **Isovaleric acidaemia (IVA)**

Isovaleric acidaemia (IVA) is a rare inherited disorder that prevents the breakdown of a building block of protein, the amino acid leucine. This then causes a harmful build-up of a substance called isovaleric acid in the blood. Children with IVA can become severely unwell. Without treatment, this can lead to a coma and permanent brain damage. Some babies with IVA have problems within a few days of birth; other children become unwell at a few months or years of age, maybe during a minor illness, such as a chest infection or a tummy upset.

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

IVA has been screened for in Wales since January 2015.

- **Glutaric aciduria type 1 (GA1)**

Glutaric aciduria type 1 (GA1) is a rare inherited disorder that prevents the breakdown of certain building blocks of protein, in particular the amino acids lysine and tryptophan. For people with GA1, eating normal amounts of protein can cause harmful substances to build up in the blood and urine. In children with GA1, a minor illness, such as a chest infection or a tummy upset, can lead to serious problems. Without treatment, the child can go into a coma. Though most children come out of the coma, they usually have brain damage that affects their ability to control their muscles and movements. This means that they may be unable to sit, walk, talk or swallow.

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

GA1 has been screened for in Wales since January 2015.

- **Homocystinuria (HCU)**

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

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

  HCU has been screened for in Wales since January 2015.

Sickle cell disorders (SCD)

Sickle cell disorders (SCD) is a term that describes a group of conditions in which haemoglobin in red blood cells is abnormal in structure. This causes red blood cells to take up a shape like a crescent moon or farmer's sickle when de-oxygenated. Sickled red blood cells are not as flexible as normal red blood cells and can cause blockages within small blood vessels. Babies who have these conditions will need specialist care throughout their lives.
People with SCD can have attacks of severe pain, get serious, life threatening infections and are usually anaemic (their bodies have difficulty carrying oxygen).

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

SCD has been screened for in Wales since 2013.

5 Editorial team

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

Ruth Lawler	Head of Maternal and Child Screening
Catherine Boyce	Programme Co-ordinator
Professor Stuart Moat	Consultant Clinical Biochemist, Director of Wales Newborn Screening Laboratory
Dr Sikha de Souza	Consultant in Public Health
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	Informatics & Data Analyst
Dan Owens	Communications Officer
Chris Jones	Communications Co-ordinator

6 Pre-Release List

These Official Statistics were sent to the people on this pre-release list five working days prior to publication in accordance with the Pre-publication Official Statistics Order Access (Wales) 2009.

Public Health Wales

Jan Williams	Chair
Dr Tracey Cooper	Chief Executive
Dr Quentin Sandifer	Executive Director of Public Health Services and Medical Director
Leah Morantz	Head of Communications

Welsh Government

Dr Frank Atherton	Chief Medical Officer
Dr Andrew Goodall	Director General - Health and Social Services
Rebekah Tune	Head of Strategic Communications and Marketing
Prof Chris Jones	Deputy Chief Medical Officer / Medical Director NHS Wales
Dr Heather Payne	Senior Medical Officer for Maternal & Child Health
Helen Tutt	Senior Executive Manager – Health Protection Services
Stephen Thomas	Head of Health Protection Branch