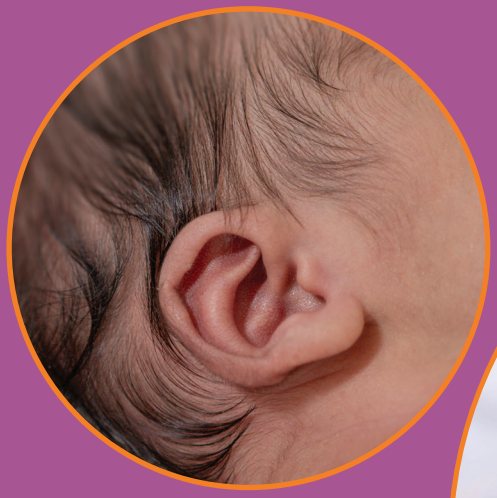
National Healthy Childhood Programme



Screening Programmes Report 2024

- National Newborn Bloodspot Screening Programme
- National Universal Newborn Hearing Screening Programme

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Summary

This report provides a detailed summary of the activity and performance of the Health Service Executive's (HSE) National Healthy Childhood Programme screening programmes – the National Newborn Bloodspot Screening Programme and the National Universal Newborn Hearing Screening Programme, for 2024.

Target audience: This report is relevant to all stakeholders with an interest in newborn screening in Ireland, including people living with rare conditions or permanent childhood hearing loss (PCHL), parents/ guardians, and everybody with a role in the delivery of the screening programmes and the management and support of babies diagnosed with rare conditions or PCHL through the screening programmes.

This report is available electronically from the National Newborn Screening Programme website or upon request from the programme manager by contacting child.screening@hse.ie

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Foreword from Chief Clinical Officer, Health Service Executive

On behalf of the Health Service Executive and the National Healthy Childhood Programme (NHCP), I am pleased to present to you the report of the HSE's NHCP Screening Programmes for 2024. In Ireland, the HSE NHCP delivers two population level screening programmes, both for newborn babies - the National Newborn Bloodspot Screening Programme (NNBSP) and the National Universal Newborn Hearing Screening Programme (UNHSP).

These programmes are delivered as integrated core components of the NHCP, the universal child health programme offered to all children in Ireland. The key focus of the screening programmes is early detection and intervention for rare conditions and permanent childhood hearing loss (PCHL), with the aim of reducing morbidity and mortality in our population.

The HSE is committed to the ongoing delivery and expansion of the NNBSP. In 2023, the Minister for Health approved the addition of two new conditions to the NNBSP - severe combined immunodeficiency (SCID) and spinal muscular atrophy (SMA). Implementation of quality assured population-based screening for new conditions requires significant time and resources. In 2024, the HSE NNBSP commenced the complex process of recruiting the necessary staff and procuring the required laboratory equipment to enable implementation of screening for SCID and SMA. Work continues in 2025, with validation of the screening test, development of appropriate clinical screening pathways, and updates to programme policies, procedures, protocols and guidelines and we look forward to programme implementation in 2026.

In addition to working towards the implementation of newborn screening for SCID and SMA, the HSE will continue to support the work of the National Screening Advisory Committee (NSAC) and the Health Information and Quality Authority (HIQA) in considering further expansion of the NNBSP to include a wider range of rare conditions.

The UNHSP is continuing to perform well in detecting children at an increased risk of permanent childhood hearing loss, and ensuring they are referred for diagnostic evaluation and appropriate intervention in a timely manner. It was reassuring to note the awarding of a successful tender in 2024 to ensure continued provision of this essential service.

I wish to acknowledge the hard work and dedication of all staff who contribute to the delivery of these two excellent screening programmes across the country. Their commitment to the programmes is testament to their dedication to children's health and well-being. I look forward to working with these teams in further developing our newborn screening programmes in accordance with the needs of our population.

Míle buíochas d'achan dhuine sna foirne uilig.

Dr Colm Henry
Chief Clinical Officer, HSE

Introduction from National Director of Public Health and National Clinical Lead Child Health Public Health

On behalf of the HSE's National Public Health function we welcome this third report of the HSE's National Healthy Childhood Programme (NHCP) screening programmes.

Dr John Cuddihy

Early detection and intervention are core tenets of public health and are key to improving clinical outcomes. Through newborn bloodspot and hearing screening, the HSE supports early detection of infants with very serious clinical conditions and permanent childhood hearing loss (PCHL) at the earliest possible opportunity, thereby enabling early intervention and reducing morbidity and mortality in our population.

Dr Abigail Collins

Each year in Ireland the National Newborn Bloodspot Screening Programme (NNBSP) detects approximately 130 babies with rare conditions, enabling early intervention and significantly improving outcomes for these babies and their families. The number of babies with rare conditions who are detected through the programme is increasing, and the HSE is committed to expanding the NNBSP to include other serious conditions that impact our population, which are requested of the HSE by the Minister for Health. SCID and SMA are currently undergoing the complex work required to add these new conditions to the screening programme, and we look forward to their implementation in 2026.

Through the Universal Newborn Hearing Screening Programme (UNHSP), between 80 and 90 newborns with PCHL are detected each year. These babies are referred to the audiology services for diagnostic evaluation and treatment. Early intervention, including offering hearing aids and speech and language therapy, helps to ensure best possible outcomes for babies affected by hearing loss.

This report describes the activity of the HSE's NHCP screening programmes for 2024, including numbers of babies screened and numbers of babies with rare conditions and PCHL detected through the programmes, and provides information on programme quality and performance.

We would like to sincerely express our gratitude to our many colleagues with a role in the delivery and quality assurance of the NHCP screening programmes, and commend their ongoing support for and commitment to children and their families.

Dr John Cuddihy
National Director of Public Health

Dr Abigail Collins
National Clinical Lead Child Health Public Health

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List of Abbreviations

ABBREVIATION	DEFINITION
AABR	Automated Auditory Brainstem Response
ADA-SCID	Adenosine Deaminase Deficiency Severe Combined Immunodeficiency
AOAE	Automated Otoacoustic Emissions
CGAL	Classical Galactosaemia
CCO	Chief Clinical Officer
CCMV	Congenital Cytomegalovirus
CF	Cystic Fibrosis
CHI	Children's Health Ireland
CHT	Congenital Hypothyroidism
CLN	Clinical Liaison Nurse
GA1	Glutaric Aciduria Type 1
HCU	Homocystinuria
HIQA	Health Information Quality Authority
HSE	Health Service Executive
HTA	Health Technology Assessment
KPI	Key Performance Indicator
MCADD	Medium Chain Acyl-CoA Dehydrogenase Deficiency
MSUD	Maple Syrup Urine Disease
NEC	Nippon Electric Company
NHCP	National Healthy Childhood Programme
NICO	Neonatal Intensive Care Unit
NIMS	National Incident Management System
NNBSL	National Newborn Bloodspot Screening Laboratory
NNBSP	National Newborn Bloodspot Screening Programme
NSAC	National Screening Advisory Committee
OAE	Otoacoustic Emissions
PCHL	Permanent Childhood Hearing Loss
PHN	Public Health Nurse
PICU	Paediatric Intensive Care Unit
PKU	Phenylketonuria
PPV	Positive Predictive Value
S4H	Smart 4 Hearing
SCBU	Special Care Baby Unit
SCID	Severe Combined Immunodeficiency
SMA	Spinal Muscular Atrophy
NUNHSP	National Universal Newborn Hearing Screening Programme
UPI	Unique Perinatal Identifier

Glossary of Terms

Sensitivity refers to a screening method's ability to designate an individual with a condition as screen positive. A highly sensitive method minimises the occurrence of false negatives.

Specificity is the ability of the screen to designate an individual without a condition as screen negative. A highly specific screen minimises the occurrence of false positives.

Positive predictive value (PPV) is the likelihood that the screening participant has the condition screened for when the screen is positive.

Negative predictive value (NPV) is the likelihood that the screening participant does not have the condition screened for when the screen is negative.

A **false positive** is a positive screen in a person who does not have the condition being screened for.

A **false negative** is a negative screen in a person who has the condition being screened for.

Figure 1:
How to calculate sensitivity, specificity, PPV and NPV

Test result	Has the condition (cases)	Does not have the condition (healthy)
Positive	True positive	False positive
Negative	False negative	True negative

These values can be used to calculate:

Sensitivity = $\dfrac{\text{true positive}}{\text{true positive + false negative}}$

Specificity = $\dfrac{\text{true negative}}{\text{true negative + false positive}}$

Positive predictive value = $\dfrac{\text{true positive}}{\text{true positive + false positive}}$

Negative predictive value = $\dfrac{\text{true negative}}{\text{true negative + false negative}}$

The **detection rate** of a screening programme is the number of actual cases of the condition detected in a population that has been screened, expressed as a proportion or per a certain number of people screened.

The **yield** is the measure of previously unrecognised disease, diagnosed as the result of screening and brought to treatment.

Introduction

In Ireland, the HSE National Healthy Childhood Programme (NHCP)[1] provides two population level screening programmes:

- the National Newborn Bloodspot Screening Programme (NNBSP) and
- the National Universal Newborn Hearing Screening Programme (UNHSP).

The key focus of both screening programmes is early identification of serious medical conditions and appropriate interventions to reduce mortality and/or morbidity and improve the quality of life of our population. The programmes are delivered as integrated components of the NHCP, the universal programme of clinical care offered to all children in Ireland to support them and their families from birth[2]. The NHCP is delivered under the governance of the National Public Health function of the Chief Clinical Officer (CCO) of the HSE.

A key strength of the NHCP screening programmes is the fact that they are integrated within the NHCP. Integrated care is a collaborative approach to healthcare where different services work together to deliver coordinated, continuous, and person-centered care, aiming to improve patient outcomes and experience. Integrated care is key to the HSE's strategic reform and innovation agenda. Integrated care, such as that provided by the NHCP, is an example of how the HSE is implementing Sláintecare.

National Newborn Bloodspot Screening Programme

Newborn Bloodspot Screening has been embedded in universal health services for the children of Ireland since 1966, when the programme commenced with screening for phenylketonuria (PKU). The NNBSP now offers screening for a total of nine rare conditions, with Ministerial approval to expand the programme to include Spinal Muscular Atrophy (SMA) and all forms of Severe Combined Immunodeficiency (SCID). Recommendations for expansion of the NNBSP are made by the National Screening Advisory Committee (NSAC), an independent advisory committee which advises the Minister and Department of Health on all new proposals for population-based screening programmes and revisions to existing programmes.[3]

Every year in Ireland, approximately 130 babies with rare conditions are detected through the NNBSP. Newborn screening enables early diagnosis and treatment of these babies, resulting in better outcomes for them and their families.

Universal Newborn Hearing Screening Programme

Universal newborn hearing screening commenced in Ireland in 2011, following publication of the report of the National Audiology Review Group[4]. It has been implemented on a national basis since the end of 2013. The national UNHSP aims to detect infants who may have moderate or severe permanent childhood hearing loss (PCHL) and who require further evaluation.

1 https://www.hse.ie/eng/health/child/newbornscreening/
2 https://www.hse.ie/eng/about/who/healthwellbeing/our-priority-programmes/child-health-and-wellbeing/national-healthy-childhood-programmenew.html
3 https://www.gov.ie/en/campaigns/nsac/?referrer=https://www.nsacommittee.gov.ie/
4 https://www.lenus.ie/handle/10147/128291?show=full

Every year in Ireland, approximately 80 babies with PCHL are detected through the UNHSP. Early diagnosis and appropriate intervention for PCHL is vital for these children to approach school entry with age-appropriate language and communication skills, so that the development of literacy, numeracy and knowledge acquisition is on a typically-developing trajectory, rather than the child, the family and educators having to endeavour to 'catch up'. Late diagnosis and consequent delayed development have potential long-term implications for a child's socioemotional wellbeing, educational achievement, employment prospects, and mental health[5].

5 https://www.infanthearing.org/nhstc/docs/Year%202019%20JCIH%20Position%20Statement.pdf

Section 1

National Newborn Bloodspot Screening Programme

Introduction – National Newborn Bloodspot Screening Programme

Overview of the National Newborn Bloodspot Screening Programme and conditions screened for

The overall aim of the HSE NNBSP is to reduce morbidity and mortality by screening newborn babies for rare but clinically serious conditions that benefit from early detection and intervention. Each year in Ireland, approximately 130 babies are diagnosed with a rare condition through the NNBSP.

Newborn bloodspot screening involves taking a small sample of blood from a newborn baby's heel, (also referred to as the 'heel-prick test'), between 72 and 120 hours after birth and placing the blood on a screening card. The blood sample is taken by either a Midwife or a Public Health Nurse (PHN) as part of routine care for mothers and babies after delivery. The screening card is then sent to the National Newborn Bloodspot Laboratory (NNBSL) at Children's Health Ireland (CHI) Temple Street, where samples are analysed and from where onward care is organised. This includes referral of screen detected babies to the appropriate paediatric service for further testing and treatment, if indicated.

The NNBSP provides a quality assured screening programme for the following nine conditions, listed in order of when screening commenced in Ireland:

- Phenylketonuria (PKU) - 1966
- Homocystinuria (HCU) - 1971
- Classical Galactosaemia (CGAL) - 1972
- Maple Syrup Urine Disease (MSUD) - 1972
- Congenital Hypothyroidism (CHT) - 1979
- Cystic Fibrosis (CF) - 2011
- Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD) - 2018
- Glutaric Aciduria Type 1 (GA1) - 2018
- Adenosine Deaminase Deficiency Severe Combined Immunodeficiency (ADA-SCID) - 2022

Newborn bloodspot screening is available to all babies born in the Republic of Ireland and to any baby (up to one year of age) who arrives in Ireland before any screening has been performed or if conditions that are screened for in Ireland are not screened for in their country of birth. The screening method for cystic fibrosis (CF) is not reliable in babies over six weeks of age, so screening for CF is not offered to babies over six weeks of age.

Constituted in 2019, the National Screening Advisory Committee (NSAC) is an independent advisory committee which advises the Minister and Department of Health on all new proposals for population-based screening programmes and revisions to existing programmes in Ireland.[6] In order to recommend a condition for inclusion in the NNBSP, NSAC must be satisfied that the condition fulfils internationally accepted criteria pertaining to the viability, effectiveness and appropriateness

6 Further information on NSAC is available at: https://www.gov.ie/en/campaigns/nsac/

of screening.[7] These criteria include a stipulation that the condition should be an important health problem as judged by its frequency and/or severity.

All of the conditions screened for under the NNBSP are important health problems by virtue of their severity, and most have a relatively high incidence in the Irish population compared to global incidence rates as described in Table 1.

Table 1:
Conditions included in the National Newborn Bloodspot Screening Programme

Condition	Date Started	Irish Incidence (2017-2024)	Worldwide Incidence*
Phenylketonuria (PKU)	1966	1:4,078	1:12,000
Homocystinuria (HCU)	1971	1:66,417	1:120,000
Classical Galactosaemia (CGAL)	1972	1:10,566	1:45,000
Maple Syrup Urine Disease (MSUD)	1972	1:464,916	1:225,000
Congenital Hypothyroidism (CHT)	1979	1:845	1:3,500
Cystic Fibrosis (CF)	2011	1:2,152	1:3,500
Glutaric Aciduria Type 1 (GA1)¥	2018	1:85,379	1:100,000
Medium Chain Acyl Co-A Dehydrogenase Deficiency (MCADD) ¥	2018	1:17,076	1:14,600
Adenosine Deaminase Deficiency Severe Combined Immunodeficiency (ADA-SCID)$	2022	1:137,072	1:200,000

*CHI Temple Street (2022). A Practical Guide to Newborn Bloodspot Screening in Ireland 9th edition

¥Screening for GA1 and MCADD commenced in December 2018. Table 1 analysis for these conditions reflects six years of screening (2019-2024)

$Screening for ADA-SCID commenced in May 2022, Table 1 analysis for these conditions reflects 2.5 years of screening

In January and November 2023, following recommendations from the NSAC based on the findings of detailed Health Technology Assessments (HTAs) carried out by the Health Information and Quality Authority (HIQA), the Minister for Health, approved the addition of SCID and SMA respectively to the NNBSP.

Following successful service plan bids for the required resources to enable this expansion, work is now progressing to enable implementation of quality assured population-based screening for SCID and SMA in Ireland.

7 https://www.gov.ie/en/department-of-health/publications/about-the-national-screening-advisory-committee-nsac/#nsac-criteria

Operational delivery of the National Newborn Bloodspot Screening Programme

The HSE is responsible for the operational delivery of the NNBSP.

While the operational and clinical governance of the NNBSP is through the National Public Health function of the Office of the CCO, services responsible for newborn bloodspot sample taking, analysis and reporting lie outside National Public Health. As such, delivery of the NNBSP is a complex process necessitating collaborative working across services and disciplines, with programme leadership and clinical governance provided through National Public Health/NHCP, sample taking delivered by the PHN and midwifery services, and sample receipt, analysis, reporting and referral of screen detected infants undertaken by the NNBSL on behalf of the HSE.

There are two key operational elements to the provision of the NNBSP – (1) sample taking and (2) laboratory services.

Sample taking

As described above, newborn bloodspot screening involves taking a small sample of blood from a newborn baby's heel between 72 and 120 hours after birth and placing the blood on a screening card. Sample taking for the NNBSP is delivered through the PHN and midwifery services. Bloodspot samples are taken by either a midwife or a PHN, in the maternity hospital/unit or at home, as part of routine care provided to mothers and babies after delivery. Samples are sent by registered post, or transferred by courier, to the NNBSL at CHI Temple Street. A national procedure titled "*Procedure for all Community and Hospital Services Providing the National Newborn Bloodspot Screening Programme (NNBSP)*" is in place to provide a standardised approach for the implementation and operation of the NNBSP in all relevant hospital and community settings.

Informed consent

Signed consent for screening is obtained by the sample taker (midwife/PHN) from the parent(s)/guardian(s) (hereafter referred to as 'parents') of the baby being offered screening. The consent process is carried out just before the bloodspot sample is taken. A parent must sign the newborn screening card to indicate their consent for their baby to undergo screening. To facilitate informed consent, parents are provided with a parent information leaflet both antenatally and at the time of sample taking. The parent information leaflet has been approved by the National Adult Literacy Agency (NALA) and has been translated from English into 14 different languages.

The parent information leaflet includes the following core information:

- the purpose of newborn bloodspot screening is to help identify babies that may be at risk of having one or more of the conditions screened for
- most babies will not have any of these conditions
- for the small number of babies who do have one of these conditions, the benefits of screening are significant, with screen detected babies benefitting from early diagnosis and treatment

- no screening programme is 100% accurate - some babies with a positive screen will not have the condition being screened for (false positives), and, very rarely, some babies with a negative/ normal screen will have the condition being screened for (false negatives)
- the screening programme is recommended for all babies.

There is also an information sheet as a cover page on the newborn bloodspot screening card that the sample taker gives to the parents at the time of sample collection.

Information for parents and the general public is also available on the HSE website.

Sample receipt, analysis, reporting and referral of babies who screen positive for a rare condition

The NNBSL at CHI Temple Street, provides a single, centralised laboratory service for the HSE NNBSP. The NNBSL is responsible for the receipt, processing, analysis and reporting of all newborn bloodspot screening samples taken in the Republic of Ireland, and for initiating the referral of babies who screen positive for a rare condition to the relevant clinical service as required.

The NNBSL has a bespoke laboratory information management system and provides electronic reports to each PHN area to allow for reconciliation that screening has been undertaken, or declined, and results reported for all babies within their service.

Quality Assurance of the NNBSL

The NNBSL is accredited by the Irish National Accreditation Board (INAB) to ISO Standard 15189. The NNBSL engages with a number of external quality assurance schemes such as the US Centre for Disease Control and Prevention (CDC), NEQAS in the UK and ERNDIM a European scheme.

Governance for the National Newborn Bloodspot Screening Programme

Overall programme ownership, clinical governance and quality assurance for the NNBSP lies with the National Public Health Function of the Chief Clinical Officer (CCO) of the HSE. As described above, the NHCP screening programmes are delivered as integrated components of the NHCP, which sits within the National Public Health function of the CCO.

Clinical governance, including quality assurance, for the NNBSP is provided through the NNBSP Governance Group. This is a multidisciplinary group chaired by the National Clinical Lead Child Heath Public Health, with representation from key stakeholder groups including paediatrics, public health nursing, midwifery, laboratory, public health medicine and administrative staff. Subgroups of the NNBSP progress actions relevant to specific conditions screened for.

The NNBSP Governance Group provides quality assurance for the NNBSP through monitoring the performance of the programme against a suite of metrics, including key performance indicators (KPIs). The Governance Group met quarterly in 2024, with quality assurance (including the metrics and programme evaluations/ reviews presented in this report), operational issues and programme updates discussed at each meeting, and relevant actions agreed upon and progressed between meetings.

Further, the HSE has convened a Child Screening Oversight Group, chaired by Dr Orla Healy, National Clinical Director HSE Quality and Patient Safety. The purpose of the Oversight Group is to provide strategic oversight and direction to the NNBSP through review and monitoring the performance and governance of the NNBSP. The NNBSP provided updates to three meetings of the Oversight Group in 2024.

Figure 2 describes the governance structure for the NNBSP.

Figure 2:
Governance structure for the NNBSP

NNBSP activity and performance, 2024 – Summary overview

This report presents data for babies who completed their newborn bloodspot screening under the NNBSP during 2024, and references activity and performance in previous years for comparison purposes/ trend analysis. Note that yearly data may be subject to change.

Summary metrics

Table 2 outlines key headline metrics (including KPIs) for the NNBSP, 2020-2024 inclusive.

Table 2: NNBSP summary metrics

Metric	2020	2021	2022	2023	2024	Programme Target (KPIs only)
Number of babies screened	57,016	60,985	54,775	54,820	54,319	
Programme acceptability/update	99.9%	99.9%	99.9%	99.8%	99.8%	
Number of opt-outs	37	60	81	106	115	
Number of samples analysed~	66,122	70,420	62,519	66,455	64,860	
% samples taken between 72-120 hours after birth	96.5%	96.0%	96.1%	96.8%	97.1%	95%
Avoidable repeats – repeat samples required due to quality issues (% of babies screened)	2,172 (3.8%)	2,349 (3.85%)	2,755 (5.0%)	3,030 (5.5%)	2,654 (4.9%)	
% samples received by NNBSL within 3 working days	99.2%	98.6%*	98.8%	98.9%	98.7%	98.2% (100%**)
% of screening cards with Unique Perinatal Identifier (UPI) recorded	99.4%	99.5%	99.5%	99.4%	99.5%	99%
% 'screen positive' clinical referrals performed within 10 days of sample receipt (all conditions except Cystic Fibrosis)	100%	100%	100%	100%	100%	100%
% referrals sent to Cystic Fibrosis Centres by 4th week of life	91.9%¥	93.8%$	97.7%	100%	100%	95%

~ includes repeat samples
* The NNBSL ICT system was not available during Q2 2021 due to the HSE cyber-attack. This figure was calculated using Q1, Q3 and Q4 only
** KPI target amended to 98.2% from Q1 2024
¥Analytical issues in the genetics laboratory in Q4 2020 impacted the achievement of this KPI
$Analyser failure in the genetics laboratory in Q3 2021 impacted the achievement of this KPI
Note: in-depth analysis of individual performance measures is provided later in this report

Programme acceptability/uptake

As outlined in Table 2, the acceptability/uptake rate of the NNBSP amongst parents is consistently high at over 99.8% annually between 2020 and 2024.

While the proportion of parents opting out of NBS remains very low, between 0.1 and 0.2% annually, absolute numbers are increasing year on year, with 115 parents opting out in 2024. The majority (82%) of parents who opted out in 2024 were parents of babies born in Ireland. While the number and overall proportion of parents opting out of the NNBSP annually remains small, the increasing trend in recent years is of concern to the programme and requires further investigation. Parents who opt out of the programme are informed of the implications and requested to sign an opt-out form. The NNBSP added an additional data field to the opt-out form in 2024 to try and capture information on the reasons why parents choose not to participate in the programme. This information will be used to better understand parental concerns and design interventions to maximise programme uptake. Information on reasons why parents choose to opt-out of the NNBSP will be included in the NHCP screening programmes annual report for 2025.

Yield and detection rate

Of the 54,319 babies screened in 2024, 143 were confirmed as having one of the rare conditions screened for under the NNBSP. Table 3 describes numbers of screen detected cases, by condition and year (2020-2024), and rates per 100,000 babies screened for the combined time period 2020-2024 inclusive.

Table 3:
Confirmed positive cases 2020, 2021, 2022, 2023 and 2024 and 2020-2024 combined

Condition	Number of confirmed positive cases					Total Number confirmed positive cases 2020-2024 inclusive	Rate per 100,000 babies screened 2020-2024 inclusive
	2020	2021	2022	2023	2024		
CHT	64	62	75	87	89	377	133.7
CF	29	27	21	30	26	133	47.2
PKU	11	16	15	17	13	72	25.5
GAL	4	8	3	5	8	28	9.9
MCADD	2	7	3	4	3	19	6.7
HCU	1	1	2	0	1	5	1.8
GA1	0	0	1	0	2	3	1.1
ADA-SCID	-	-	0	0	1	1	0.7
MSUD	0	0	0	0	0	0	0
Total	**111**	**121**	**120**	**143**	**143**	**638**	**226.3**

Key Performance Indicators for the NNBSP

This section outlines the KPIs used by the NNBSP to monitor the performance of the screening programme.

Newborn bloodspot screening is a time sensitive process, ensuring conditions screened for are identified and treatment commenced as soon as possible. The NNBSP monitors five KPIs relevant to timely sample collection, dispatch and processing. These KPIs are reported to the NNBSP Governance Group on a quarterly basis and a summary is noted in Table 4.

Table 4:
Summary of KPIs for the NNBSP 2020-2024

	2020	2021	2022	2023	2024	Programme Target
KPI 1 Timeliness of sample collection (sample collected between 72 and 120 hours after birth)	96.5%	95.8%	96.1%	96.8%	97.1%	95%
KPI2 Timeliness of sample dispatch (sample received in NNBSL within 3 working days)	98.2%	98.6%	98.8%	98.9%	98.7%	98.2%
KPI 3: Enhanced tracking abilities (% of samples with UPI recorded)	99.4%	99.5%	99.5%	99.4%	99.5%	99%
KPI 4: Timely processing of a screen positive sample (all except CF) (clinical referral made within 10 days of sample receipt in NNBSL)	100%	100%	100%	100%	100%	100%
KPI 5: Timely processing of a screen positive sample for CF (clinical referral made by 4th week of life)	91.9%¥	93.8%$	97.7%	100%	100%	95%

¥ Q4 2020 Analytical issues in the genetics laboratory impacted the achievement of this KPI

$Q3 2021 Analyser failure in the genetics laboratory impacted the achievement of this KPI

KPI 1: Timeliness of sample collection

Target: 95% of samples are taken between 72 and 120 hours of birth.

Rationale: This KPI is to ensure that all babies, where consent is obtained, are screened within the recommended timeframe of between 72 and 120 hours after birth. Timeliness of sample taking is key consideration in newborn bloodspot screening. If a sample is taken before 72 hours, there is an increased risk of false negative screening results for some conditions, e.g. babies may not have received an adequate intake of protein for metabolite accumulation in the case of metabolic conditions. If the sample is taken after 120 hours there is a chance that babies may present clinically unwell before the results of the screening are available.

The target of 95% for KPI was exceeded in 2024, at 97.1% and a comparison across the five-year period 2020-2024 is shown in Table 4.

KPI 2: Timeliness of sample dispatch

Target: 98.2% of samples are received by the NNBSL within 3 working days of the sample being taken.

Rationale: This KPI is to ensure that all bloodspot screening cards are received by the NNBSL in a timely manner. As the conditions screened for are all clinically serious and can cause symptoms at/ shortly after birth, it is important that babies are identified, diagnosed and commenced on appropriate treatment as soon as possible. Therefore, screening cards must be sent to the NNBSL without delay.

From Q1 2024, the target for this KPI was revised from 100% to 98.2%. This maintains an appropriately stringent target while acknowledging the potential impact of weekends, bank holidays etc. on transit times for samples. The target of 98.2% was exceeded in 2024 (98.7%). The NNBSP Governance Group will continue to monitor this KPI against this revised target. A comparison across the five-year period 2020-2024 is shown in Table 4.

KPI 3: Enhanced tracking abilities

Target: 99% of newborn bloodspot screening cards contain the unique perinatal identifier (UPI).

Rationale: The UPI is a unique identifier generated by the sample takers and is crucial to track babies throughout the screening process. This KPI monitors performance with regard to the recording of the UPI on the screening card.

The target for this KPI was exceeded in 2024, at 99.5% and a comparison across the five-year period 2020-2024 is shown in Table 4.

KPI 4 and KPI 5: Timely processing of a screen positive sample

Rationale: Given the potential for rapid and irreversible clinical deterioration in infants with conditions screened for under the NNBSP, timely referral of screen detected babies to the appropriate clinical team is required to ensure optimal outcomes.

a. Target: 100% of clinical referrals for PKU, MSUD, HCU, CHT, GAL, MCADD and GA1 are initiated within 10 working days of sample receipt.

The NNBSP target of 100% for this KPI has been consistently met by the programme across the time period 2020-2024 inclusive (Table 4). Of note, at the point of clinical referral, appointments and admissions are organised typically for the same day or the following day.

b. Target: 95% of clinical referrals for CF to a CF specialist centre by 4th week of life.

The target of 95% for this KPI was met in 2024 (Table 4).

Note:

In Q2 and Q4 of 2020, analytical issues in the laboratory at the Department of Clinical Genetics (DCG) impacted turnaround times for genetic analysis, resulting in subsequent delays of up to two weeks in clinical referrals to CF centres for a small number of screen positive cases.

In Q3 2021, the failure of an analyser in the laboratory at the Department of Clinical impacted turnaround times for genetic analysis.

NNBSP activity and performance, 2024 – individual conditions

No screening test is 100% accurate. False positive and false negative cases are an accepted reality of population-based screening. The NNBSP monitors the performance of screening tests for each condition screened through monitoring the sensitivity, specificity and PPV of the screening tests.

Sensitivity, specificity, PPV

Sensitivity refers to a screening method's ability to designate an individual with a condition as screen positive. It is calculated by dividing the number of true positive cases by the number of true positive and false negative cases combined. A highly sensitive method minimises the occurrence of false negatives.

Specificity is the ability of the screen to designate an individual without a condition as screen negative. It is calculated by dividing the number of true negative cases by the number of true negative and false positive cases combined. A highly specific screen minimises the occurrence of false positives.

Positive predictive value (PPV) is the likelihood that the screening participant has the condition screened for when the screen is positive. It is calculated by dividing the number of true positive cases by the number of true positive and false positive cases combined.

Test result	Has the condition (cases)	Does not have the condition (healthy)
Positive	True positive	False positive
Negative	False negative	True negative

These values can be used to calculate:

Sensitivity = $\dfrac{\text{true positive}}{\text{true positive + false negative}}$

Specificity = $\dfrac{\text{true negative}}{\text{true negative + false positive}}$

Positive predictive value = $\dfrac{\text{true positive}}{\text{true positive + false positive}}$

Negative predictive value = $\dfrac{\text{true negative}}{\text{true negative + false negative}}$

Undetected cases, including false negatives

The NNBSP takes a proactive approach to the investigation of clinically diagnosed cases, undetected through the screening programme. This includes false negatives and also cases that do not meet the screening case definition and which are not considered false negative cases, e.g. mild or atypical forms of conditions screened for. The identification and review of these cases is considered by the NNBSP to be an integral part of the programme's quality assurance. The NNBSP takes a two-pronged approach to identification of undetected cases:

1. An annual formal communication is sent to relevant paediatric services, requesting that any cases of conditions screened for that were diagnosed clinically and not through the screening programme are notified to the NNBSP.
2. Additionally, paediatrician colleagues are requested to immediately inform the NNBSP if a case of a condition screened for is diagnosed clinically, as opposed to through the screening programme, at any stage throughout the year.

The NNBSP has an Incident Review Policy, which includes the steps to be taken by the programme in investigating undetected cases. This involves a thorough review of the case to determine whether there was any issue or non-compliance within the screening programme. A final report, including key findings and recommendations, is reviewed through the appropriate governance lines, including by the NNBSP Governance Group, and appropriate action agreed upon. If an undetected case is found to be a false negative (i.e. meets the case definition for screening but was undetected through the screening programme), data relating to the sensitivity of the screening programme for the relevant condition for the year the baby underwent screening are updated to reflect this, and appropriate action is taken to mitigate risk of future occurrences.

To date, the NNBSP has not been informed of any undetected cases for babies screened between 2020 and 2024 inclusive. As such, the sensitivity of the NNBSP for each condition screened for is reported as 100% for all conditions across this time period.

In the years 2020-2024 inclusive, the NNBSP completed investigations of five cases pertaining to babies screened outside that time period. In each of the cases, the investigation did not identify any clinical concerns or non-compliance within the screening programme.

Sensitivity, specificity and PPV – NNBSP 2020-2024

The sensitivity, specificity and PPV of screening for each condition screened for under the NNBSP is noted below. For some years it was not possible to calculate sensitivity as there were no positive cases diagnosed in those years. Due to the small number of cases that are detected by the NNBSP, PPV data was reviewed for 2020-2024 inclusive with the exception of ADA-SCID that used Q2 & Q3 2022 only, 2023 and 2024.

Congenital hypothyroidism (CHT)

Definition: Congenital hypothyroidism (CHT) is a disorder present at birth where the thyroid gland is either absent, underdeveloped, or unable to produce enough thyroid hormone (thyroxine), which is crucial for normal growth and brain development. Most cases of CHT happen by chance/ sporadically and only a very small number of cases are inherited.

Screening Test: The screening test for CHT is carried out by measuring blood thyroid stimulating hormone in the blood spot sample.

Yield, sensitivity, specificity, PPV: CHT is the most commonly diagnosed condition screened for by the NNBSP. In 2024, 122 babies screened positive for CHT, of whom 89 were ultimately confirmed to have the condition ('true positive') and 33 were found to be false positive cases (Table 5). There were no false negative cases identified.

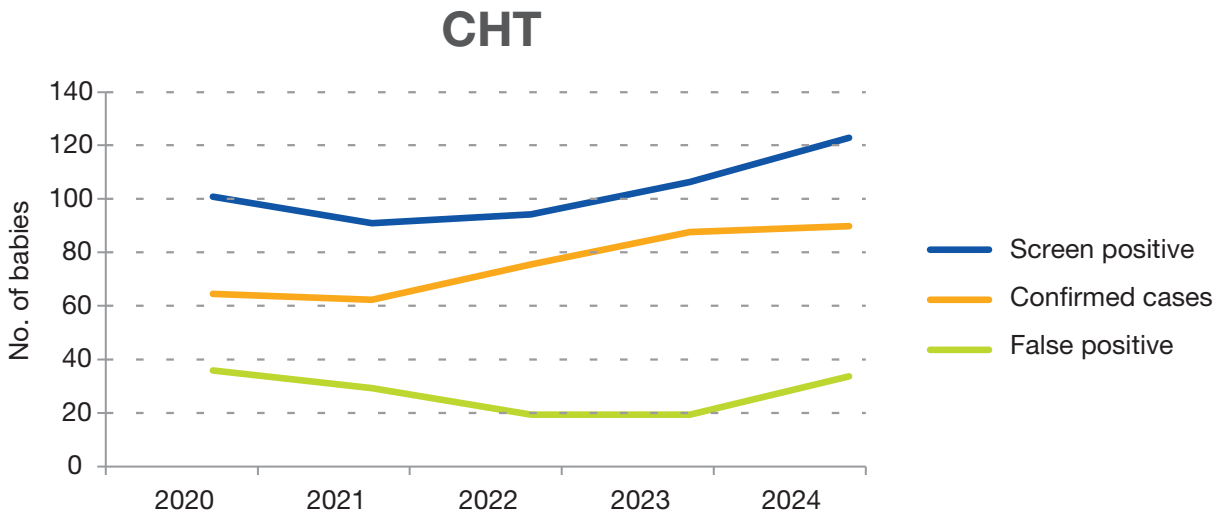
In the five-year period 2020-2024 inclusive, a total of 513 babies screened positive for CHT, of whom 377 were true positive/confirmed cases and 136 babies were ultimately determined to be false positive cases (Table 5). No false negative cases were identified. The sensitivity of the screening programmes for CHT was 100% annually and the specificity exceeded 99.9% each year. The overall PPV for the CHT screening programme for 2020-2024 inclusive was 73.49%.

Table 5:
Congenital hypothyroidism 2020-2024

Congenital hypothyroidism	2020	2021	2022	2023	2024	Total (2020-2024)
Screen Positive cases	100	91	94	106	122	513
Confirmed cases	64	62	75	87	89	377
Dysgenesis	*16*	*8*	*6*	*19*	*20*	*69*
Dyshormonogenesis	*15*	*26*	*33*	*24*	*42*	*140*
Unclassified	*32*	*24*	*25*	*27*	*3*	*111*
*Transient hypothyroidism**	*1*	*4*	*11*	*17*	*24*	*57*
False positive cases	36	29	19	19	33	136
False negative cases	0	0	0	0	0	0
Sensitivity (%)	100	100	100	100	100	100
Specificity (%)	99.94	99.95	99.97	99.97	99.94	99.95
PPV (%)	-	-	-	-	-	73.49

*In 2020, the NNBSP introduced the term 'transient hypothyroidism' to more accurately describe and record a cohort of babies with CHT who were previously described as false positive screens. This diagnosis describes a transient abnormality of thyroid function, which later reverts to normal. In the years 2020 and 2021, some babies who would have been recorded as having 'transient hypothyroidism' were recorded as 'false positive' by the screening programme. This had no impact on the management of the baby. They are now included in the cohort of confirmed cases of CHT.

Figure 3:
CHT data 2020-2024



Cystic fibrosis (CF)

Definition: CF is an inherited condition that causes some organs in the body, such as the lungs and pancreas, to produce thick mucus. This mucus can build up in the lungs and cause infections and these infections can cause lung damage over time. CF can cause digestive problems and people with CF can find it hard to gain weight. CF is an autosomal recessive condition with both parents carrying a pathogenic variant of the cystic fibrosis transmembrane conductance regulator (CFTR) gene.

Screening test: The initial screening test measures the levels of immunoreactive trypsinogen (IRT) in the bloodspot sample. If the blood level of IRT is high, the sample will be referred for CFTR genetic analysis which looks for the presence of 38 possible pathogenic genetic variants of the CFTR gene.

- If two genetic variants are detected, the likelihood of CF is high and the baby will be referred to one of six CF centres where a diagnostic sweat test will be carried out to confirm the diagnosis of CF.
- If one genetic variant is found, the baby will also be referred to a CF centre and a sweat test will be performed to determine whether the baby has CF or is a carrier of the CF gene variant. If the sweat test is positive, this confirms a diagnosis of CF and further genetic testing will be undertaken to identify the second genetic variant. If the sweat test is negative they do not have CF but are a carrier of the CF gene variant or may be assigned as a case of CF-screen positive inconclusive diagnosis (CF-SPID).

Yield, sensitivity, specificity, PPV: In 2024, 68 babies screened positive for CF, of whom 26 were ultimately confirmed to have the condition ('true positive') and 40 were found to be carriers of the CF gene and there were two CF-SPID cases. (Table 6).

In the five-year period 2020-2024 inclusive, 3,884 samples were sent to the associated referral laboratory for genetic analysis following an initial screen positive result. This represented almost 1.4% of all initial newborn samples received in this time period. Following genetic analysis, 439

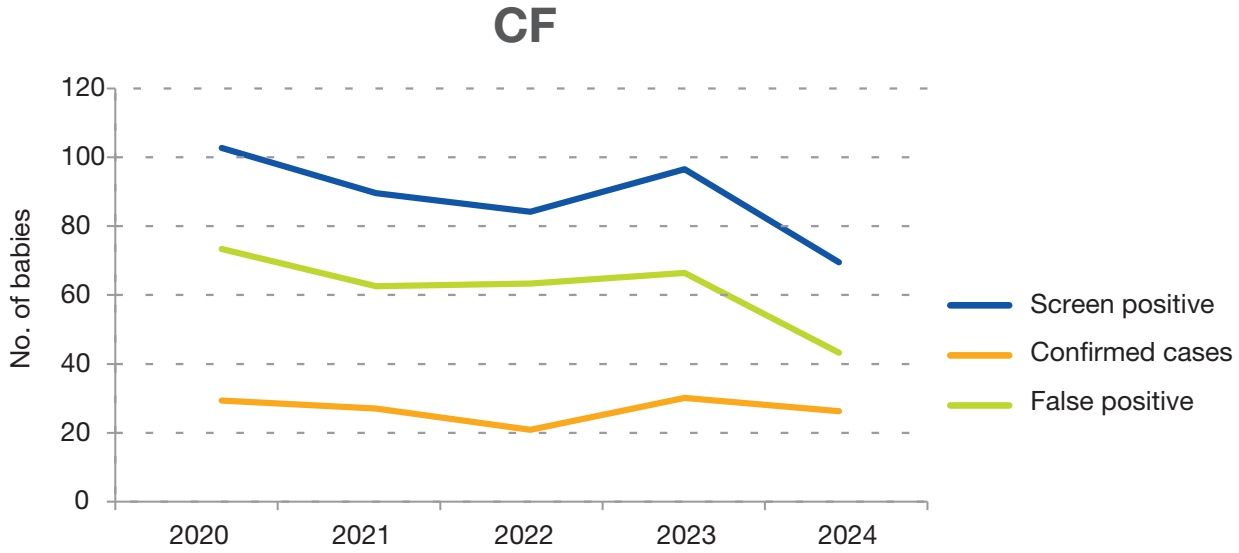
babies were found to have CF causing pathogenic variants and they were referred to one of the six CF centres for sweat testing. Of these 439 babies, 133 were diagnosed with CF following sweat testing – refer to Table 6 and Figure 4.

The sensitivity of the screening programmes for CF was 100% annually and the specificity exceeded 99.8% each year. The overall PPV for the CF screening programme for 2020-2024 inclusive was 30.3%.

Table 6:
Cystic fibrosis 2020-2024

Cystic fibrosis	2020	2021	2022	2023	2024	Total (2020-2024)
No. referred for sweat test (screen positives)	102	89	84	96	68	439
No. with two CFTR genetic variants	*22*	*24*	*20*	*20*	*22*	*108*
No. with one CFTR genetic variant	*80*	*65*	*64*	*76*	*46*	*331*
Confirmed cases	29	27	21	30	26	133
CF carriers/CF-SPID	73	62	63	66	42	306
False negative cases	0	0	0	0	0	0
Sensitivity (%)	100	100	100	100	100	100
Specificity (%)	99.87	99.9	99.88	99.88	99.92	99.89
PPV (%)	-	-	-	-	-	30.3

Figure 4:
CF data 2020-2024



Phenylketonuria (PKU)

Definition: Phenylketonuria (PKU) is a rare but potentially serious inherited disorder. PKU causes high levels of an amino acid called phenylalanine to build up in the blood and brain which can lead to brain damage and learning disabilities.

Screening test: The screening test measures the levels of phenylalanine in the bloodspot sample.

Yield, sensitivity, specificity, PPV: In 2024, 13 babies screened positive for PKU, all of which were ultimately confirmed to have the condition ('true positive') and there were no false positive cases.

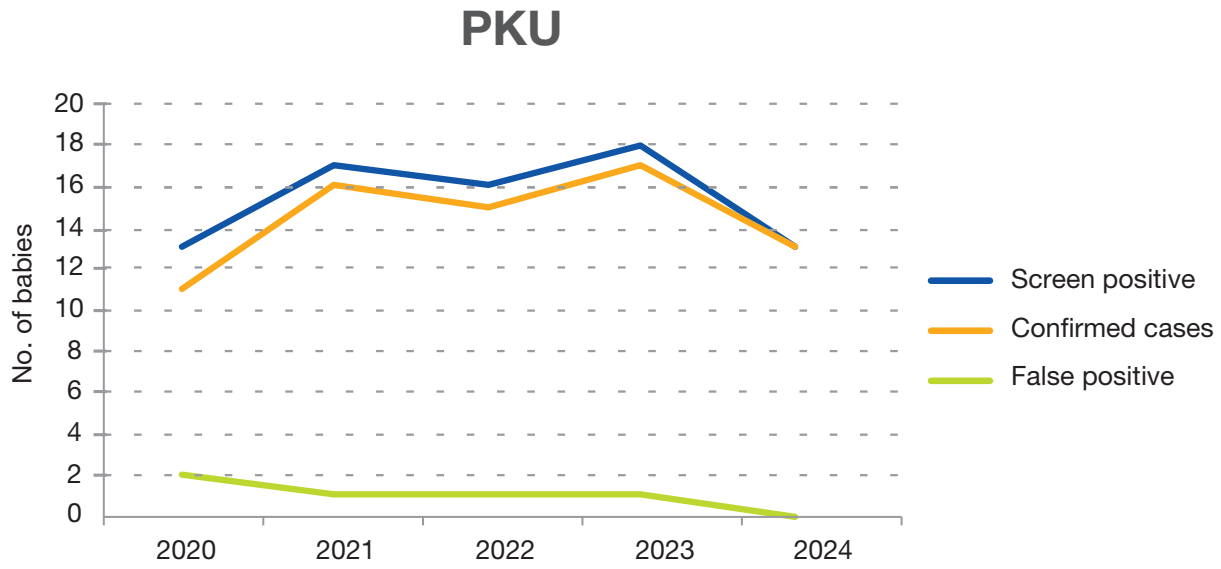
In the five-year period 2020-2024 inclusive, 77 babies screened positive for PKU, of whom 72 were true positive/confirmed cases and 5 babies were ultimately determined to be false positive cases – refer to Table 7 and Figure 5.

The sensitivity of the screening programmes for PKU was 100% annually and the specificity exceeded 99.9% each year. The overall PPV for the PKU screening programme for 2020-2024 inclusive was 93.51%.

Table 7:
Phenylketonuria 2020-2024

Phenylketonuria	2020	2021	2022	2023	2024	Total (2020-2024)
Screen positive cases	13	17	16	18	13	77
Confirmed cases	11	16	15	17	13	72
False positive cases	2	1	1	1	0	5
False negative cases	0	0	0	0	0	0
Sensitivity (%)	100	100	100	100	100	100
Specificity (%)	99.99	99.99	99.99	99.99	100	99.99
PPV (%)	-	-	-	-	-	93.51

Figure 5:
PKU cases 2020-2024



Classical galactosaemia (CGAL)

Definition: Classical galactosaemia (CGAL) is caused by a deficiency of the enzyme that breaks down galactose, a sugar found in milk, and can be life threatening for babies, or lead to liver damage or sepsis. CGAL is more common in babies born to Irish Traveller parents.

Screening test: The screening test for CGAL measures the levels of galactose in the bloodspot sample. If the levels of galactose are high then analysis of the levels of the galactose-1-phosphate uridyl transferase (GALT) enzyme is carried out (Beutler test). If there is no or very low levels of GALT enzyme activity present in the blood sample then a diagnosis of CGAL is likely.

As CGAL is more common in babies born to Irish Traveller parents and to siblings of known cases, the Beutler test is offered to these babies at birth (preferably day 1 of life). Parents/guardians are advised to keep the baby on a galactose free feed (Soya-based) until the result of the Beutler test is available. This protects the baby should they have the condition.

In 2024, 12 babies screened positive for CGAL, of whom 8 were ultimately confirmed to have the condition ('true positive') and 4 were found to be false positive cases.

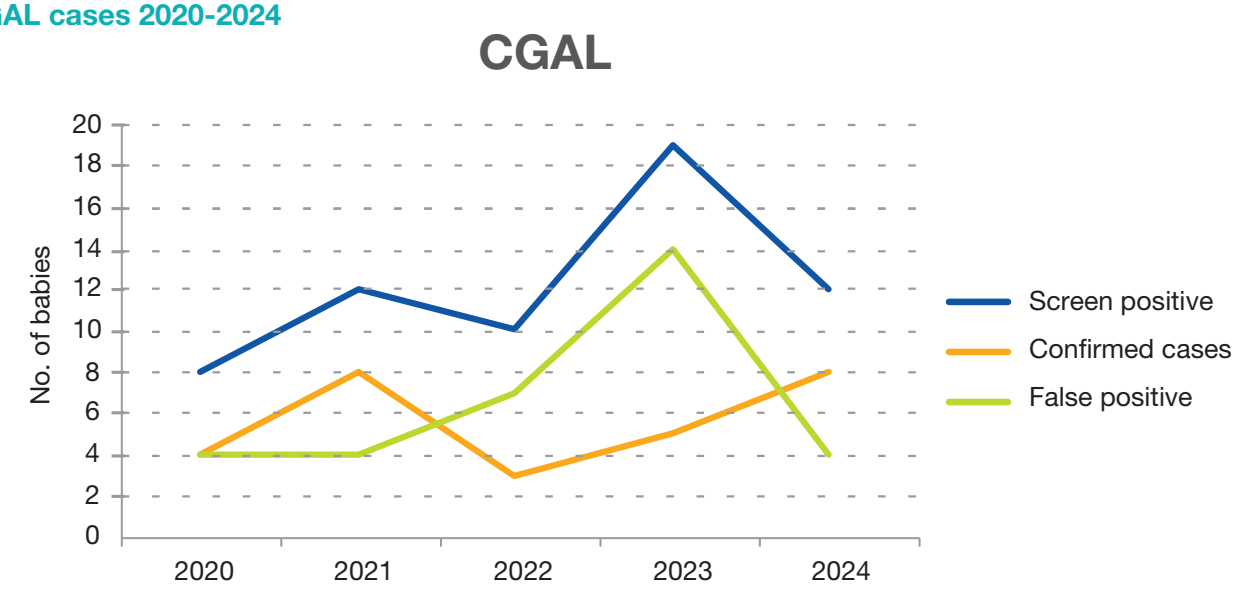
In the five-year period 2020-2024 inclusive, 61 babies screened positive for CGAL, of whom 28 were true positive/confirmed cases and 33 babies were ultimately determined to be false positive cases – refer to Table 8 and Figure 6.

The sensitivity of the screening programmes for CGAL was 100% annually and the specificity exceeded 99.9% each year. The overall PPV for the CGAL screening programme for 2020-2024 inclusive was 45.9.

Table 8:
Classical galactosaemia 2020-2024

Classical galactosaemia	2020	2021	2022	2023	2024	Total (2020-2024)
Screen positive cases	8	12	10	19	12	61
Confirmed cases	4	8	3	5	8	28
False positive cases	4	4	7	14	4	33
False negative cases	0	0	0	0	0	0
Sensitivity (%)	100	100	100	100	100	100
Specificity (%)	99.99	99.99	99.98	99.98	99.99	99.99
PPV (%)	-	-	-	-	-	45.9

Figure 6:
CGAL cases 2020-2024



Medium chain acyl coenzyme A dehydrogenase deficiency (MCADD)

Definition: Medium chain acyl coenzyme A dehydrogenase deficiency (MCADD) is an inherited condition where affected individuals cannot break down fats from food quickly enough to make energy when they are ill. This can cause toxins and low blood sugar to build up which can lead to serious complications such as brain damage, coma or even death if it is not treated.

Screening test: The screening test for MCADD measures the levels of a type of acylcarnitine, C8, in the blood spot sample. Acylcarnitines are fatty acid derivatives that are crucial in transporting fatty acids into the mitochondria to help generate energy. If the levels of C8 acylcarnitine are high this is indicative of MCADD and urgent diagnostic testing is required.

Yield, sensitivity, specificity, PPV: Screening for MCADD commenced in Ireland in December 2018. In 2024, 13 babies screened positive for MCADD, of whom 3 were ultimately confirmed to have the condition ('true positive') and 10 were found to be false positive cases.

In the five-year period 2020-2024 inclusive 76 babies screened positive for MCADD, of whom 19 were true positive/confirmed cases and 57 babies were ultimately determined to be false positive cases – refer to Table 9 and Figure 7.

The sensitivity of the screening programmes for MCADD was 100% annually and the specificity exceeded 99.9% each year. The overall PPV for the MCADD screening programme for 2020-2024 inclusive was 25.0.

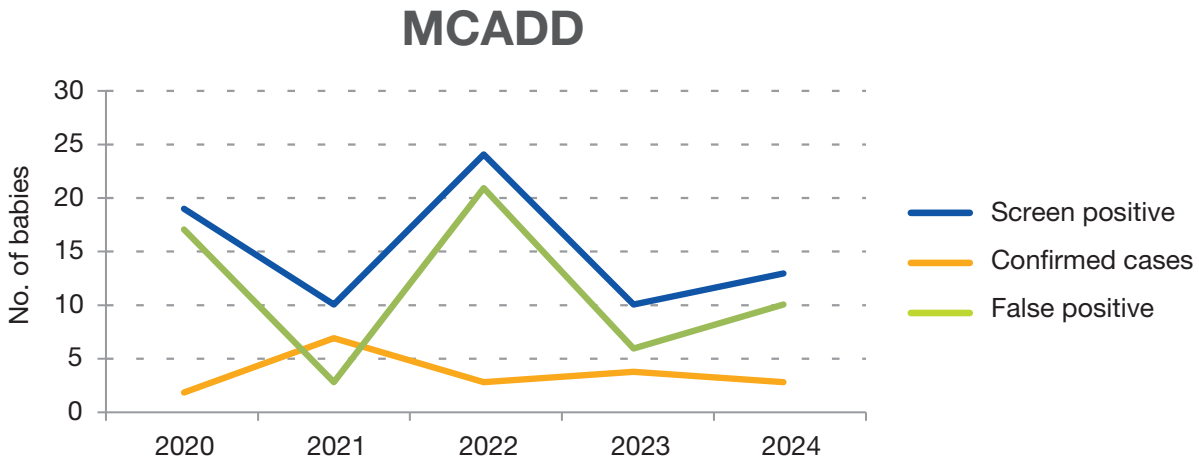
Table 9:

Medium chain acyl coenzyme A dehydrogenase deficiency 2020-2024

Medium chain acyl coenzyme A dehydrogenase deficiency	2020	2021	2022	2023	2024	Total (2020-2024)
Screen positive cases	19	10	24	10	13	76
Confirmed cases	2	7	3	4	3	19
False positive cases	17	3	21	6	10	57
False negative cases	0	0	0	0	0	0
Sensitivity (%)	100	100	100	100	100	100
Specificity (%)	99.97	99.99	99.96	99.99	99.98	99.98
PPV (%)	-	-	-	-	-	25.0

Figure 7:

MCADD cases 2020-2024



Homocystinuria (HCU)

Definition: Homocystinuria (HCU) is an inherited condition caused by a pathogenic gene variant that can cause a build-up of amino acids that can lead to eye problems, impaired brain development and bone disorders. HCU results from the accumulation in the blood of the amino acid methionine and one of its metabolites, homocysteine.

Screening test: The screening test for HCU detects high levels of methionine in the bloodspot sample. This can be challenging, as blood methionine may not be raised initially after birth. Methionine concentration is low in many baby foods, in particular breast milk, and it is important that babies are feeding well before the bloodspot sample is taken to minimise the risk of false negative HCU screening results.

Yield, sensitivity, specificity, PPV: In 2024, 21 babies screened positive for HCU, of whom 1 was ultimately confirmed to have the condition ('true positive') and 20 were found to be false positive cases.

In the five-year period 2020-2024 inclusive, 182 babies screened positive for HCU, of whom 5 were true positive/confirmed cases and 177 babies were ultimately determined to be false positive cases – refer to Table 10 and Figure 8.

The sensitivity of the screening programmes for HCU was 100% annually, except 2023 when there were no positive HCU cases detected, and the specificity exceeded 99.9% each year. The overall PPV for the HCU screening programme for 2020-2024 inclusive was 2.75%.

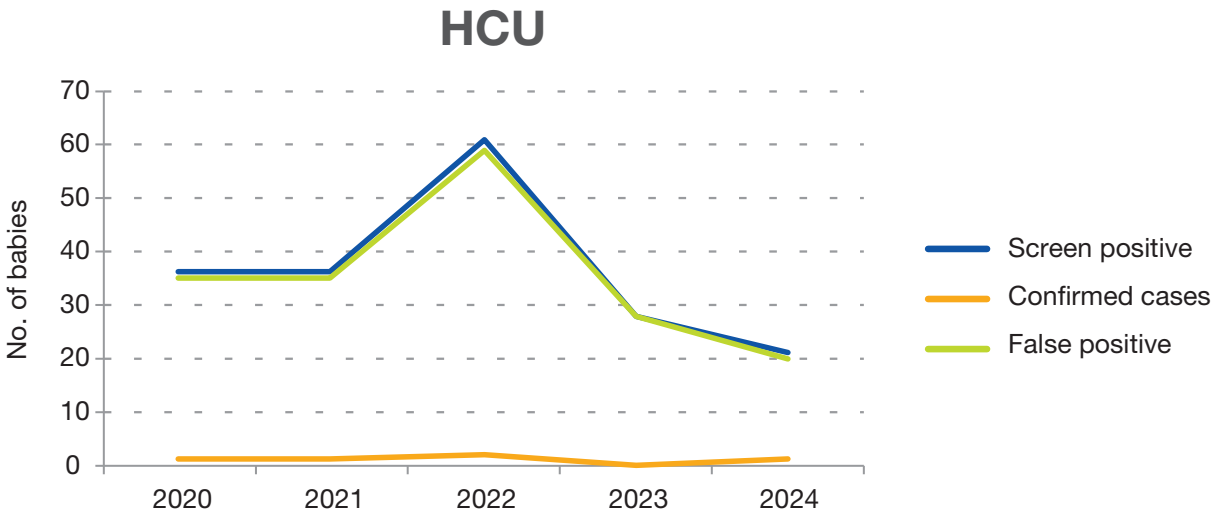
Table 10:

Homocystinuria 2020-2024

Homocystinuria	2020	2021	2022	2023	2024	Total (2020-2024)
Screen positive cases	36	36	61	28	21	182
Confirmed cases	1	1	2	0	1	5
False positive cases	35	35	59	28	20	177
False negative cases	0	0	0	0	0	0
Sensitivity (%)	100	100	100	-	100	100
Specificity (%)	99.94	99.94	99.89	99.95	99.96	99.93
PPV (%)	-	-	-	-	-	2.75

Figure 8:

HCU cases 2020-2024



Maple syrup urine disease (MSUD)

Definition: Maple syrup urine disease (MSUD) is an extremely rare but serious inherited condition where babies cannot break down certain amino acids that can build up in the blood and urine. This accumulation of amino acids can be harmful causing brain damage, developmental delay and can be life threatening. The disorder is so called because the urine may have an odour similar to that of maple syrup.

Screening test: The screening test for MSUD detects high levels of leucine in the bloodspot sample. If the levels of leucine are high, the baby will be referred for further diagnostic tests.

Yield, sensitivity, specificity, PPV: In 2024, 13 babies screened positive for MSUD, of which none were ultimately confirmed to have the condition ('true positive') and 13 were found to be false positive cases. The increase in false positive cases in 2024 relative to previous years (see Table 11) may be explained by changes to cut-off values for the screening test, implemented to increase the sensitivity of the screening test. Numbers of false positives may also have been impacted by numbers of babies receiving total parental nutrition (TPN) and/or ICU care. The number of MSUD false positives are being monitored closely by the NNBSP.

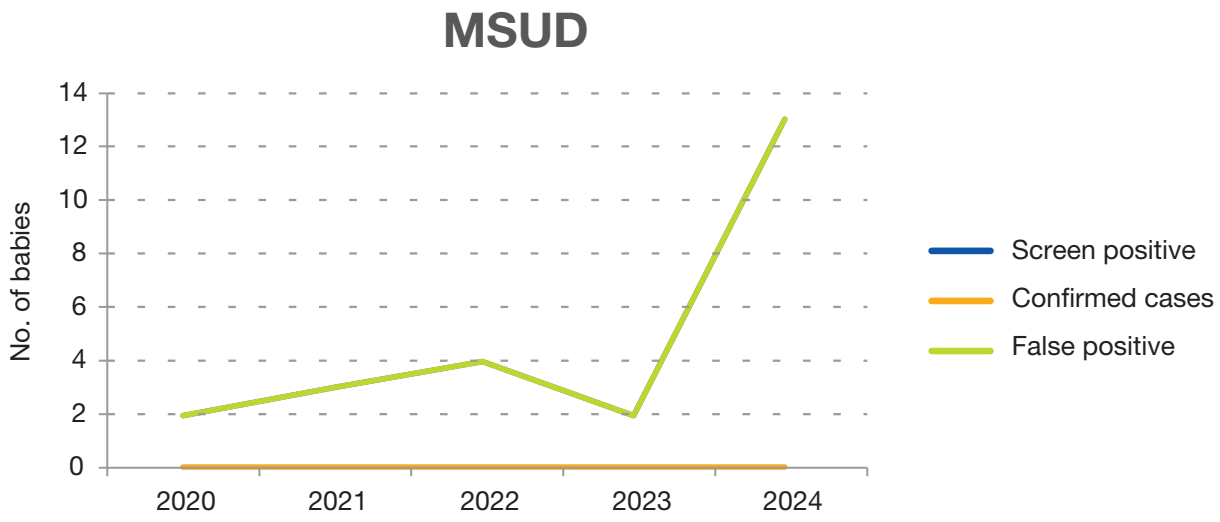
In the five-year period 2020-2024 inclusive, 24 babies screened positive for MSUD and all of these babies were ultimately determined to be false positive cases – refer to Table 11 and Figure 9.

The sensitivity of the screening programmes for MSUD was not possible to calculate as there were no positive cases. The specificity exceeded 99.9% each year. The overall PPV for the MSUD screening programme for 2020-2024 inclusive was not possible to calculate as there were no positive cases during that time period.

Table 11:
Maple syrup urine disease 2020-2024

Maple syrup urine disease	2020	2021	2022	2023	2024	Total (2020-2024)
Screen positive cases	2	3	4	2	13	24
Confirmed cases	0	0	0	0	0	0
False positive cases	2	3	4	2	13	24
False negative cases	0	0	0	0	0	0
Sensitivity (%)	-	-	-	-	-	-
Specificity (%)	99.99	99.99	99.99	100	100	99.99
PPV (%)	-	-	-	-	-	-

Figure 9: MSUD cases 2020-2024



Glutaric aciduria type 1 (GA1)

Definition: Glutaric aciduria type 1 (GA1) is an inherited condition caused by an absent or non-functioning enzyme needed to break down protein in the diet. Without this enzyme, harmful amino acids builds up in the body, which can cause damage to the brain, movement difficulties, seizures and difficulty swallowing and, if not detected and treated early, can be life threatening.

Screening test: The screening test for GA1 measures the levels of glutarylcarnitine (C5DC) in the blood spot sample. Glutarylcarnitine is produced during the breakdown of lysine and tryptophan and plays a role in transporting these amino acids and other acyl-groups into the mitochondria for energy production. If the levels of C5Dc are high, this is indicative of GA1 and urgent diagnostic tests are required.

Yield, sensitivity, specificity, PPV: In 2024, 8 babies screened positive for GA1, of whom 2 were ultimately confirmed to have the condition ('true positive') and 6 were found to be false positive cases.

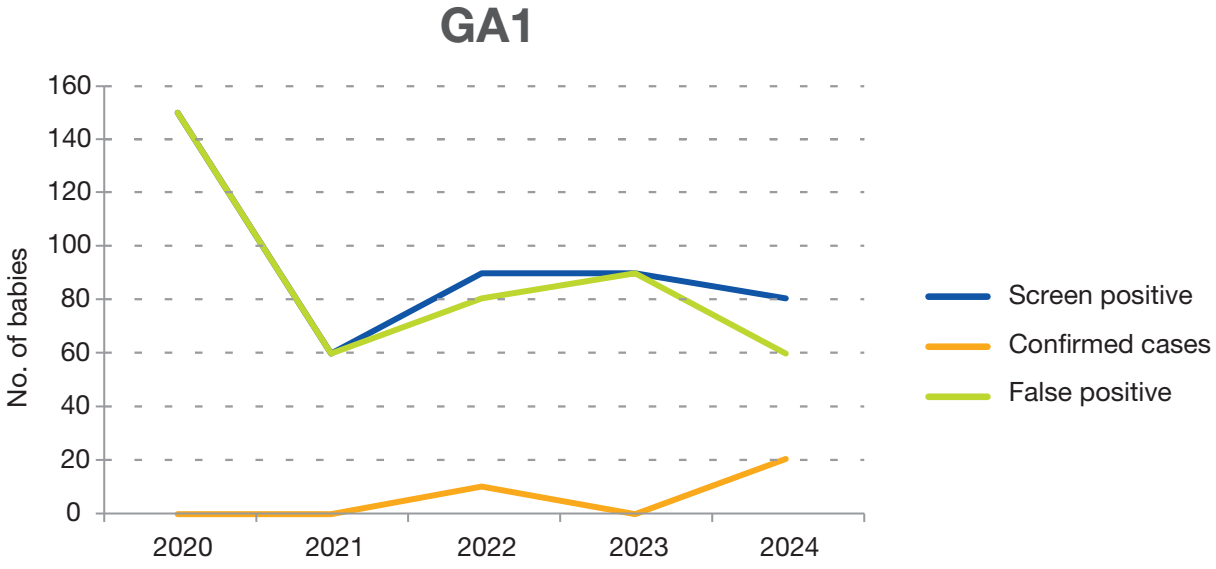
In the five year period 2020-2024 inclusive, 47 babies screened positive for GA1, of whom 3 were true positive/confirmed cases and 44 babies were ultimately determined to be false positive cases – refer to Table 12 and Figure 10.

The sensitivity of the screening programmes for GA1 was 100% in 2022 and 2024. It was not possible to calculate sensitivity for GA1 in 2020, 2021 or 2023 as there were no positive cases detected. The specificity exceeded 99.9% each year. The overall PPV for the GA1 screening programme for 2020-2024 inclusive was 6.38%.

Table 12:
Glutaric aciduria type 1 (GA1) 2020-2024

Glutaric aciduria type 1	2020	2021	2022	2023	2024	Total (2020-2024)
Screen positive cases	15	6	9	9	8	47
Confirmed cases	0	0	1	0	2	3
False positive cases	15	6	8	9	6	44
False negative cases	0	0	0	0	0	0
Sensitivity (%)	-	-	100	-	100	100
Specificity (%)	99.97	99.99	99.99	99.98	99.99	99.98
PPV (%)	-	-	-	-	-	6.38

Figure 10:
GA1 cases 2020-2024



Adenosine deaminase deficiency severe combined immunodeficiency (ADA-SCID)

Definition: Adenosine deaminase deficiency severe combined immunodeficiency (ADA-SCID) is a rare but serious inherited disease that is caused by the lack of an enzyme called adenosine deaminase (ADA). Babies with ADA-SCID have a weak immune system so they cannot fight infections and this can make common infections life threatening.

Screening test: The screening test for ADA-SCID measures the levels of deoxyadenosine (dADO) in the blood spot sample. Accumulation of dADO occurs in babies with ADA-SCID.

Yield, sensitivity, specificity, PPV: Screening for ADA-SCID commenced in May 2022. There were no cases of ADA-SCID identified in 2022 or 2023 and no false positives – refer to Table 13. There were no clinical presentations or diagnoses of any babies with ADA-SCID during this time period. In 2024, the first case of ADA-SCID was identified through screening, with no false positives identified, giving a sensitivity, specificity and PPV of 100%.

Table 13:
ADA-SCID cases 2022-2024

Adenosine deaminase deficiency severe combined immunodeficiency	2022	2023	2024	Total (2022-2024)
Screen positive cases	0	0	1	1
Confirmed cases	0	0	1	1
False positive cases	0	0	0	0
False negative cases	0	0	0	0
Sensitivity (%)	100	100	100	100
Specificity (%)	100	100	100	100
PPV (%)	-	-	-	100

Section 2

National Universal Newborn Hearing Screening Programme

Introduction - National Universal Newborn Hearing Screening Programme

Approximately 1 to 2 babies in every 1,000 are born with a hearing loss in one or both ears. Unaddressed hearing loss, including congenital hearing loss, has serious consequences for a child's development, education, and social integration. A key mitigating factor is the age at which intervention is initiated, with a significant body of research demonstrating improved outcomes in children whose hearing loss is detected early and who receive early intervention compared to those with later detection and treatment. The scientific literature demonstrates that children who are born deaf or who acquire hearing loss very early in life, and who are identified and receive appropriate intervention before six months of age, are on a par with their hearing peers in terms of language development by the time they reach five years of age.

Newborn hearing screening enables early identification and intervention for babies with hearing loss. When followed by timely access to definitive diagnostic testing and appropriate intervention, screening enables improved outcomes for these babies, including improved language skills, cognitive development, social integration and educational outcomes. Additionally, cost–effectiveness studies have demonstrated the financial benefits of universal newborn hearing screening for health and social care systems in high-income, lower-middle-income and middle-income countries.

Overview of the National Universal Newborn Hearing Screening programme

Universal newborn hearing screening commenced in Ireland in 2011, following publication of the report of the National Audiology Review Group[8], and has been implemented on a national basis since 2013. The National Universal Newborn Hearing Screening Programme (UNHSP) is a population-based screening programme for babies in Ireland, delivered as an integrated component of the HSE's National Healthy Childhood Programme (NHCP), which is the universal programme of clinical care offered to all children in Ireland to support them and their parents from birth.[9]

Aim of the UNHSP

The aim of the UNHSP is to identify babies who are likely to have congenital permanent childhood hearing loss (PCHL), defined as a moderate or greater hearing loss affecting one or both ears, and who should be offered further evaluation. Moderate or greater hearing loss is defined as a range of >/= 40dB, averaged over 0.5, 1, 2 and 4 kHz, (for the better ear in pure tone audiometry thresholds). The hearing loss may be sensorineural, conductive or mixed.

Babies who do not pass screening ('no clear response' in one or both ears) are referred for diagnostic testing, and those with PCHL are supported to achieve optimal outcomes though early intervention.

8 https://www.lenus.ie/handle/10147/128291?show=full
9 https://www.hse.ie/eng/about/who/healthwellbeing/our-priority-programmes/child-health-and-wellbeing/national-healthy-childhood-programmenew.html

Approximately 80-90 babies with PCHL are detected through the UNHSP each year in Ireland.

As with all screening programmes, not every case of PCHL will be detected through the UNHSP. It is also important to note that the UNHSP does not screen for mild hearing loss, and that children can develop or acquire a hearing loss later in life, so it is important that the child's hearing is checked as they grow. Parents are provided with hearing surveillance checklists after hearing screening is completed, and hearing checks are carried out at various time points, including as a component of the developmental checks performed by the PHN and in primary schools as part of the school health service.

Eligibility

All babies under 3 months of age, born or resident in Ireland, should be offered newborn hearing screening.

Newborn hearing screening is contraindicated in babies who are less than 34 weeks gestational age, or over 3 months (corrected) age. However, newborn hearing screening can be carried out up to 6 months corrected gestational age, e.g. in babies who were too unwell to undergo screening in the earlier neonatal period. Newborn hearing screening is also contraindicated in babies with the following conditions/ circumstances, all of whom should be referred for immediate audiological assessment:[10]

- Microtia or external ear canal atresia.
- Neonatal bacterial meningitis or meningococcal septicaemia.
- Confirmed congenital cytomegalovirus (cCMV).
- Suspected or confirmed Zika virus.
- Programmable ventriculo-peritoneal (PVP) shunts in place.
- Babies who have a prolonged period (> 6 months) in Special Care Baby Unit (SCBU)/ Paediatric Intensive Care Unit (PICU).

Babies receiving palliative care should not be automatically screened or referred for immediate or targeted hearing assessment. However, screening can take place at the request of the parent and/ or paediatrician.

Screening pathway/operational delivery of newborn hearing screening

The HSE has contracted NEC Software Solutions to deliver the screening element of the national UNHSP. Newborn hearing screening is undertaken by trained hearing screeners at each of the 19 maternity hospital/units. The vast majority of newborn hearing screening is completed in the maternity hospital/unit, but there are circumstances where delivery/ completion of screening is required in an outpatient setting.

Newborn babies who are eligible for screening are identified in each maternity hospital/unit from the local birth register.

10 Referrals should be made by the relevant paediatric service, and may be facilitated through the local screeners

Informed consent

Signed informed consent for screening is obtained by the hearing screener at the time of screening from the parent. A parent must sign the consent form to indicate their consent for their baby to undergo hearing screening. To facilitate informed consent, parents are provided with an information leaflet and the screening process is discussed with them by the hearing screener. Key information provided to parents includes:

- 1 to 2 babies in every 1,000 are born with a hearing loss in one or both ears.
- Newborn hearing screening can help identify if your baby has a hearing loss that could affect their speech and language development in the absence of early intervention and support.
- The national UNHSP aims to identify those babies with a permanent childhood hearing loss of a moderate or greater degree.
- Approximately 80-90 cases of permanent childhood hearing loss are diagnosed each year through the UNHSP.
- Newborn hearing screening is available to all eligible babies in the Republic of Ireland and is undertaken soon after birth.
- Screening is primarily carried out while the mother and baby are still in the maternity hospital/unit as inpatients.
- If the screening is not completed in the maternity hospital/unit it then takes place in an outpatient clinic within 2-3 weeks of birth.
- The screening process does not hurt or harm the baby in anyway.
- Parents receive information regarding newborn hearing screening via the leaflet 'Your Baby's Hearing Screening Test' and also verbally from the hearing screener who will answer any questions the parent(s)/guardian(s) may have.
- A range of translated information leaflets can be found at https://www2.hse.ie/conditions/newborn-hearing-screening/why-we-screen/

The screening tests

Newborn hearing screening involved the following tests:

1. **Automated Oto Acoustic Emission (AOAE) test**: This test checks the baby's inner ear (the 'cochlea'). When a healthy ear receives the sound, the outer hair cells of the inner ear produce an echo. Recording this echo indicates that the baby's hearing is likely to be satisfactory.

   If the baby does not have a clear response to the AOAE test this does not necessarily mean that they have a hearing loss. Other reasons why the first screening test may not show a clear response include:

   - the baby may have been unsettled at the time of screening
   - there may have been background noise during the screening test, for example on a noisy hospital ward, which affected the results
   - the baby may have fluid or a temporary blockage in their ear after the birth. This is very common and passes with time.

The AOAE test is repeated if the baby does not have a clear response in one or both ears. If there is still no clear response, the baby has a second type of hearing screening test – the Automated Auditory Brainstem Response (AABR) test.

2. **Automated Auditory Brainstem Response (AABR) test**: This test measures how the baby's hearing (auditory) nerve and brain respond to sounds. The auditory nerve carries information about sound from the inner ear to the brain.

   The AABR test is only done once. If the baby has no clear response to the AABR test they will then be referred to the community audiology service for diagnostic tests to determine whether they have a hearing loss or not.

Screening result and associated actions

Babies who have a clear response in both ears have completed screening and the parents are informed of the results, provided with a hearing surveillance checklist, and the baby is discharged from the screening programme.

If, on completion of screening, the baby has no clear response in one or both ears, the baby is referred for a diagnostic audiological assessment without delay ('immediate follow up'). This assessment should take place within four weeks of referral. Parents are provided with the information leaflet '*Your Baby's Visit to the Audiology Clinic*', to help prepare them for this appointment. Diagnostic assessments are carried out by HSE Audiologists. Babies identified with a hearing loss are fitted with hearing aids if appropriate and supported by the early years support services.

If certain risk factors for hearing loss are present, the baby is offered a paediatric audiology review in the HSE Community Audiology service at 9 months of age, regardless of whether they had a clear response at screening. These risk factors include:

- Congenital Syndromes associated with hearing loss, including Downs Syndrome, Waardenburg Syndrome, Treacher-Collins Syndrome, Goldenhaar Syndrome, Alport Syndrome, Usher Syndrome, etc.
- Craniofacial anomalies: Head and neck malformations, e.g. Cleft lip and palate, deformed pinnae (not isolated ear tags).
- Congenital Infection (confirmed or suspected): Toxoplasmosis / Rubella / Zika.
- NICU/SCBU care required for > 48 hours with no clear response on OAEs in BOTH ears despite clear responses on AABR and positive immediate family history of sensorineural hearing loss.
- Referred by screener for early audiological assessment but failed to attend.
- Parental or professional concern.

Governance of the Universal Newborn Hearing Screening Programme

The HSE is responsible for the delivery of the national UNHSP. The NHCP screening programmes are delivered as integrated components of the NHCP, which sits within the National Public Health function of the CCO.

Overall programme ownership, clinical governance and quality assurance for the UNHSP lies with the National Public Health Function of the Chief Clinical Officer (CCO) of the HSE.

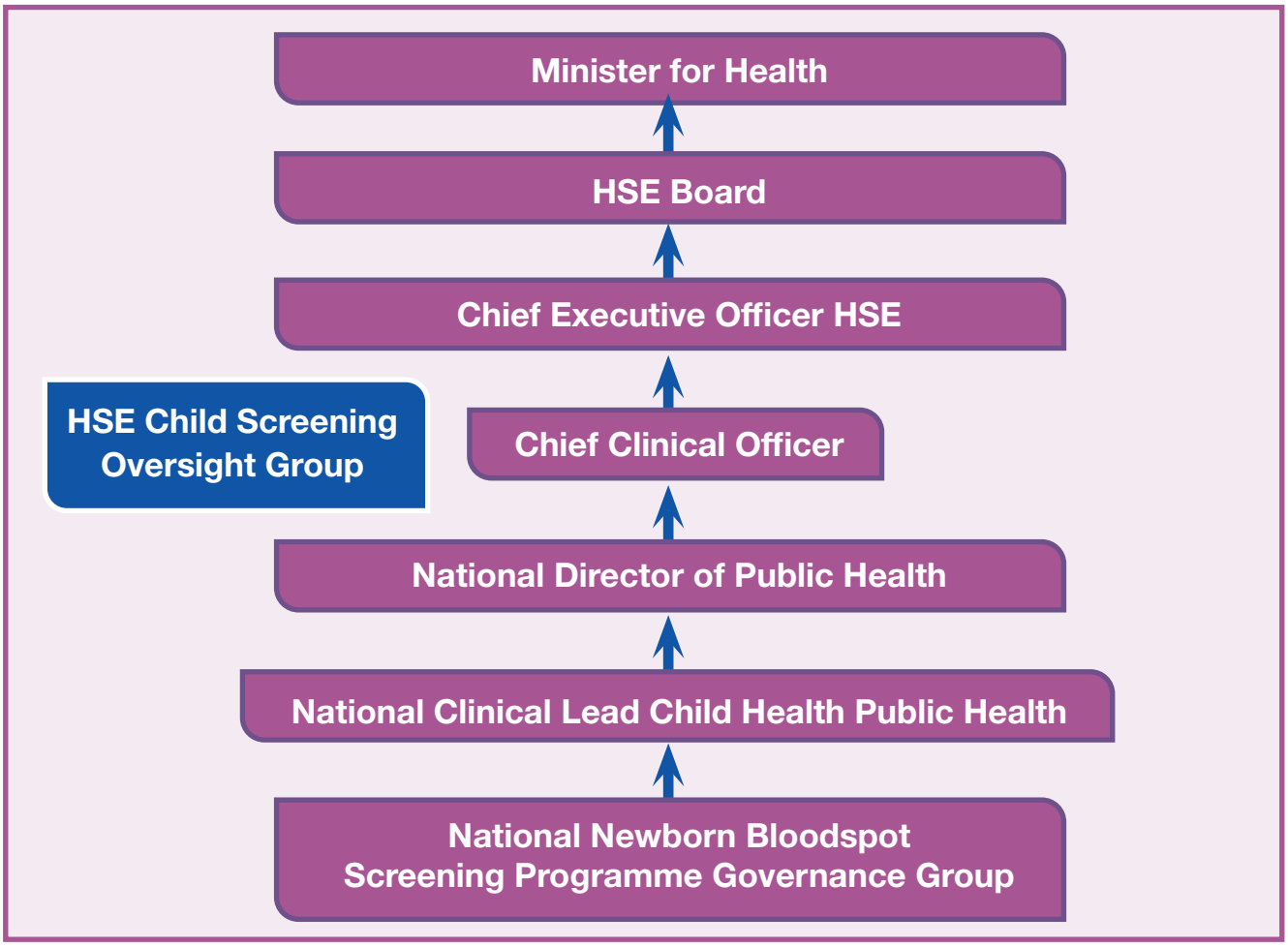
Clinical governance, including quality assurance, for the UNHSP is provided through the UNHSP Governance Group. This is a multidisciplinary group chaired by the National Clinical Lead Child Heath Public Health, with representation from key stakeholder groups involved in the identification and management of hearing loss in babies and includes paediatrics, public health nursing, hearing screeners, public health medicine and administrative staff.

The UNHSP Governance Group met quarterly during 2024, with quality assurance (including the metrics and programme evaluations/ reviews presented in this report), operational issues and programme updates discussed at each meeting and relevant actions agreed upon and progressed between meetings.

Further, the HSE has convened a Child Screening Oversight Group, chaired by Dr Orla Healy, National Clinical Director HSE Quality and Patient Safety. The purpose of the Oversight Group is to provide strategic oversight and direction to the UNHSP through review and monitoring the performance and governance of the UNHSP. The UNHSP provided updates to three meetings of the Oversight Group in 2024.

See Figure 11 for the governance structure for the UNHSP.

Figure 11:
Governance structure for the UNHSP



Quality assurance for the Universal Newborn Hearing Screening Programme

The Joint Committee on Infant Hearing (JCIH) defines three evidence based global benchmarks for early detection and intervention for PCHL, commonly referred to as the '1-3-6 benchmarks': screening completed by 1 month, audiologic diagnosis by 3 months, and enrolment in early intervention by 6 months. The World Health Organization (WHO) notes that most newborn hearing screening guidelines adopted internationally are based on the JCIH 1-3-6 benchmarks and acknowledges that these principles form a strong basis for universal newborn hearing screening.

The UNHSP collates and monitors a suite of 24 metrics (and additional sub-metrics), comprising 14 KPIs and 10 service demand/activity metrics. These metrics/ KPIs are grouped into 3 categories – screening, audiology and early intervention/ management – and are broadly aligned to the 1-3-6 JCIH benchmarks.

Universal Newborn Hearing Screening Programme activity and performance, 2024 - summary metrics

In 2024, 52,862 babies were screened though the UNHSP and a total of 68 babies with a PCHL were identified through the programme. Of the babies screened, 52,119 (98.6%) had a clear response with no follow up required. An additional 205 (0.4%) had a clear response but were referred for targeted follow up by way of a paediatric audiology review due to the presence of risk factors for hearing loss. A further 538 (1%) had a screening result of 'no clear response' in one or both ears and were referred to community audiology for further diagnostic assessment.

Table 14 outlines summary data for all babies screened in the time period 2020-2024 inclusive.

Table 14:
National UNHSP summary data 2020-2024

	2020	2021	2022	2023	2024	2020-2024
Clear response – no follow-up required	55,475 (98.3%)	59,302 (98.6%)	53,313 (98.5%)	53,360 (98.5%)	52,119 (98.6%)	273,569 (98.5%)
Clear response – targeted follow-up required	202 (0.4%)	238 (0.4%)	233 (0.4%)	209 (0.4%)	205 (0.4%)	1,087 (0.4%)
Referred to audiology (no clear response)	776 (1.3%)	629 (1.0%)	583 (1.1%)	612 (1.1%)	538 (1.0%)	3,138 (1.1%)
Total screened	**56,453 (100%)**	**60,169 (100%)**	**54,129 (100%)**	**54,181 (100%)**	**52,862 (100%)**	**277,794 (100%)**
Total identified with PCHL	89	84	92	79	68	412

Table 15 outlines key headline metrics (including KPIs) for the time period 2020-2024 inclusive. In 2024 there were 52,999 babies eligible for screening. Of these 52,914 (99.93%) started screening and 52,862 (99.9%) completed screening. In 2024, 52,611 babies (99.53) completed screening within four weeks, which exceeds the programme target of 95%.

With regards to referrals to audiology, in 2024 there were 743 babies referred to audiology, with 538 referred for immediate diagnostics and 205 for targeted follow up.

In 2024, 68 babies were identified with a PCHL through screening, 62 of whom (91.1%) were identified by 6 months of age, which exceeds the programme target of 80%.

Table 15:
National UNHSP summary metrics 2020-2024

Metric	2020	2021	2022	2023	2024	Programme Target (KPIs only)
Number of babies registered	57,144	60,860	54,780	54,766	53,458	
Number of babies eligible for screening	56,619	60,298	54,275	54,316	52,999	
Completion of Screening						**Programme Target (KPIs only)**
Number of eligible babies who started hearing screening (of those offered screening)	56,519 (99.95%)	60,218 (99.96%)	54,179 (99.93%)	54,242 (99.96%)	52,914 (99.93%)	>98%
Number of eligible babies who completed screening	56,453 (99.88%)	60,161 (99.91%)	54,118 (99.89%)	54,194 (99.91%)	52,862 (99.9%)	
Number of eligible babies who completed screening by 4 weeks of age	53,766 (95.24%)	59,566 (99.01%)	53,759 (99.34%)	53,973 (99.59%)	52,611 (99.53%)	>95%
Referrals						**Programme Target (KPIs only)**
Number of eligible **well** babies with no clear response at otoacoustic emissions 1 (OAE1)	2,176 (4.25%)	2,250 (4.09%)	2,064 (4.18%)	1,888 (3.84%)	1,844 (3.86%)	≤30%
Number of eligible **well** babies with no clear response at otoacoustic emissions 2 (OAE2)	648 (1.27%)	757 (1.38%)	724 (1.46%)	675 (1.37%)	624 (1.31%)	≤6%
Number of eligible **well** baby referrals from AABR	514 (1.00%)	345 (0.63%)	343 (0.69%)	287 (0.58%)	179 (0.37%)	
Number of eligible **NICU** baby referrals from OAE	572 (10.83%)	572 (11.12%)	556 (11.85%)	588 (11.7%)	560 (11.1%)	
Number of eligible **NICU** baby referrals from AABR	265 (5.02%)	283 (5.5%)	240 (5.11%)	327 (6.51%)	259 (5.13%)	

Audiology (immediate and targeted follow up)						Programme Target (KPIs only)
Number of eligible babies referred to Audiology (Immediate and targeted follow up)	983 (1.74%)	868 (1.44%)	819 (1.51%)	824 (1.52%)	743 (1.41%)	
Number of eligible babies referred for immediate diagnostics	781 (1.38%)	630 (1.05%)	586 (1.08%)	614 (1.13%)	538 (1.02%)	≤3%
Number of eligible babies referred for targeted follow up	202 (0.36%)	238 (0.40%)	233 (0.43%)	210 (0.39%)	205 (0.39%)	
Outcomes						**Programme Target (KPIs only)**
Number of babies identified with a hearing loss by 6 months of age	83 (93.26%)	82 (97.62%)	89 (96.74%)	76 (96.2%)	62 (91.18%)	≥80%

Key performance indicators for the national UNHSP

This section provides further detail on a subset of five important KPIs used by the Governance Group to monitor the performance of the UNHSP. These KPIs are reported to the national Governance Group on a quarterly basis and a summary of UNHSP performance against these KPIS for the time period 2020-2024 inclusive is provided in Table 16.

Table 16:
Summary of KPIs for the UNHSP 2020-2024

KPI	2020	2021	2022	2023	2024	Programme Target
1. Proportion of eligible babies offered screening	99.9%	99.9%	99.3%	99.9%	99.9%	**>99%**
2. Proportion of eligible babies who have completed screening by 4 weeks of age	95.24%	99.01%	99.3%	99.6%	99.5%	**>95%**
3. Proportion of eligible well babies with no clear response to OAE1	4.25%	4.09%	4.09%	3.84%	3.86%	**≤30%**
4. Proportion of eligible babies referred for immediate diagnostics	1.38%	1.05%	1.08%	1.13%	1.02%	**≤3%**
5. Proportion of babies with a permanent childhood hearing loss (PCHL) who have the PCHL identified by 6 months of age	93.26%	97.62%	96.74%	96.2%	91.18%	**≥80%**

KPI 1: Proportion of eligible babies offered screening

Target: *>99% of eligible babies are offered screening.*

Rationale: To maximise the number of eligible babies that are offered newborn hearing screening.

The target for this KPI was exceeded in 2024, at 99.9%, and a comparison with previous years is shown in Table 16.

KPI 2: Timely completion of screening

Target: *>95% of eligible babies to complete screening by 4 weeks.*

Rationale: The JCIH 1-3-6 benchmarks recommend that screening is completed by 1 month of age, to support timely identification and onward referral of babies with a suspected PCHL. This KPI aims to ensure that the Irish UNHSP meets this benchmark.

The target for this KPI was exceeded in 2024, at 99.5%, and a comparison with previous years is shown in Table 16.

KPI 3: Proportion of eligible well babies with no clear response to OAE1

Target: *≤30% of eligible well babies with no clear response on otoacoustic emissions 1 (OAE1).*

Rationale: This KPI monitors the performance of the OAE1 screening test, aiming to ensure that the specificity of OAE1 is appropriate and that the proportion of babies requiring OAE2 is not excessive**.**

The target for this KPI was achieved in 2024, at 3.86%. A comparison with previous years is shown in Table 16.

KPI 4: Proportion of eligible babies referred for immediate diagnostics

Target: *≤3% of all eligible babies referred for immediate diagnostics.*

Rationale: This KPI aims to minimise the occurrence of false positives and ensure that an appropriate proportion of babies are referred to the audiology services for immediate diagnostics.

The target for this KPI was achieved in 2024, at 1.02%. A comparison with previous years is shown in Table 16.

KPI 5: Proportion of babies with a PCHL who have the PCHL identified by 6 months of age

Target: *≥80% of babies identified with a PCHL have the PCHL identified by 6 months of age.*

Rationale: The JCIH 1-3-6 benchmarks recommend that, for babies with 'no clear response' at newborn hearing screening, audiologic diagnosis is completed by 3 months of age, and enrolment in early intervention is achieved by 6 months of age. This KPI aims to support the Irish UNHSP in meeting these benchmarks and ensuring early diagnosis and intervention for screen detected babies with PCHL.

The target for this KPI was achieved in 2024, with 91.2% (62/68) of babies that were identified as having a PCHL identified by six months of age. A comparison with previous years is shown in Table 16.

Yield, detection rate, specificity, positive predictive value

In 2024, 68 babies with a PCHL were detected through the UNHSP. This equates to a detection rate of 1 in 777 babies who completed screening. The specificity of the UNHSP in 2024 was 99.1%, with a PPV of 12.6.

Table 17 illustrates the specificity and PPV for the national UNHSP for the years 2020-2024 inclusive.

Table 17:
National UNHSP specificity 2020-2024

Year	Screen completed	Clear response	No clear response	PCHL	Prevalence (per 1,000)	False positives	PPV (%)	Specificity
2020	56,453	55,672	781	89	1.6	692	11.4	98.8
2021	60,161	59,531	630	84	1.4	546	13.3	99.1
2022	54,118	53,535	583	92	1.7	491	15.8	99.1
2023	54,194	53,580	614	79	1.5	535	12.9	99.0
2024	52,862	52,324	538	68	1.3	470	12.6	99.1
Total	**277,788**	**274,642**	**3,146**	**412**	**1.5**	**2,734**	**13.1**	**99.0**

Conclusion

This report demonstrates that the newborn bloodspot and newborn hearing screening programmes are highly acceptable to parents in Ireland, with 99.8% and 99.9% of eligible babies undergoing screening through each programme, respectively, in 2024. This compares extremely favourably with uptake rates for other population-based screening programmes.

Reassuringly, the data also attest to the high quality of the NNBSP and UNHSP, with KPI targets met or exceeded across both programmes in 2024. In 2024, 142 babies with a rare condition and 68 babies with a PCHL were detected through newborn screening. When followed by prompt diagnosis and treatment, early detection through screening allows for significantly improved health outcomes and quality of life for babies and families.

The National Healthy Childhood Programme would like to extend our sincere thanks to all those with a role in the delivery and quality assurance of these critical services in 2024, and to the parents whose trust and engagement with the services supports better outcomes for babies with rare conditions and PCHL.

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