National Children's Screening Services

Report of the HSE National Children's Screening Programmes 2023

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

Sláinte Leanaí Éireann
CHI
Children's Health Ireland

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Summary

This report is a detailed summary of the activity of, and information regarding, the Health Service Executive's (HSE) two National Newborn Screening Programmes – the Newborn Bloodspot Screening Programme and the Universal Newborn Hearing Screening Programme, for 2023 with comparisons made to the previous three years 2020-2022 also.

This report is available electronically from the National Newborn Screening Programme website or upon request from the project manager (contact child.screening@hse.ie).

For more information and to provide any feedback about this report please contact 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 I am pleased to present to you the report of the HSE's National Children's Screening Programmes for 2023. This builds on the publication last year of the three year report covering 2020 to 2022 and I am delighted to see the summary of the recent 10 year review of the newborn bloodspot screening programme for cystic fibrosis also included here. In Ireland, the HSE National Children's Screening Services delivers two population level screening programmes, both for newborn babies - the National Newborn Bloodspot Screening Programme and the National Universal Newborn Hearing Screening Programme.

These programmes are delivered as part of the National Healthy Childhood Programme (NHCP), the universal child health programme delivered to all children. The key focus of both screening programmes is the early identification and appropriate interventions to reduce mortality and/or morbidity in our population.

The HSE is committed to the delivery of the National Newborn Bloodspot Screening Programme and the work necessary to support expansion to include more conditions in the coming years. Minister Donnelly has approved the addition of two new conditions, severe combined immunodeficiency (SCID) and spinal muscular atrophy (SMA). We are progressing the work required for implementation of these conditions. We will continue to support the work of the National Screening Advisory Committee and HIQA with regard to proposals to add more conditions, so that we are more in line with other European countries.

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

Dr John Cuddihy

Dr Abigail Collins

On behalf of the HSE's National Public Health function we welcome this second report of the HSE's National Children's Screening Programmes. Screening is the process of identifying healthy people who may be at increased risk of having a disease or condition. Once identified, those at increased risk are offered information, further testing, clinical management, and treatment and/or intervention if required. Screening is a pathway; it is not a diagnostic test.

Screening and prevention are core aspects of public health and through newborn screening the HSE are detecting infants with very serious clinical conditions and hearing loss at the earliest possible opportunity and getting them started on treatment and interventions to improve morbidity and mortality.

In Ireland, the HSE Children's Screening Services delivers two population level screening programmes, both for newborn babies:

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

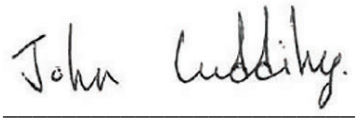
These programmes are delivered as part of the National Healthy Childhood Programme (NHCP), the universal child health programme delivered to all children. The NHCP sits within the National Public Health function of the Office of the Chief Clinical Officer.

The National Newborn Bloodspot Screening Programme detects approximately 120 babies a year whose lives are improved by the screening programme. There has been an increasing number of babies detected in the past few years and the HSE is committed to expanding the newborn

bloodspot screening programme to include other conditions that impact our population. We are also detecting permanent hearing loss in between 80 and 90 newborns a year and referring them into the appropriate audiology pathways to be assessed and fitted with hearing aids at the earliest opportunity so that they can develop their speech and language skills.

This report covers the HSE's Children's Screening Service activity for 2023 and also provides a comparison with the previous three years (2020-2022). The report is in two sections with data provided for both screening programmes on case numbers, KPIs and programme performance.

We express our gratitude to all our colleagues who do such great work as part of the two newborn screening programme and for their ongoing support 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
ADA-SCID	Adenosine Deaminase Deficiency Severe Combined Immunodeficiency
CGAL	Classical Galactosaemia
CCO	Chief Clinical Officer
CCMV	Congenital Cytomegalovirus
CF	Cystic Fibrosis
CHI	Children's Health Ireland
CHT	Congenital Hypothyroidism
GA1	Glutaric Aciduria Type 1
HCU	Homocystinuria
HIQA	Health Information Quality Authority
HSE	Health Service Executive
IT	Information Technology
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
NUNHSP	National Universal Newborn Hearing Screening Programme
UPI	Unique Perinatal Identifier

Section 1

National Newborn Bloodspot Screening Programme

Introduction

The overall aim of the HSE National Newborn Bloodspot Screening Programme (NNBSP) is to offer newborn babies screening for rare but clinically serious conditions that would benefit from early intervention to reduce mortality and/or morbidity. Each year, approximately 120 babies are diagnosed with a rare condition through the NNBSP. The NNBSP is available for all eligible babies within Ireland.

Newborn bloodspot screening involves taking a small sample of blood from a newborn baby's heel, (also referred to as the 'heel-prick'), between 72 and 120 hours after birth and placing the blood on a screening card. The screening card is a special card made of filter paper that absorbs the blood sample. The screening is undertaken as part of routine postnatal care for mothers and babies after delivery, by either a Midwife or a Public Health Nurse (PHN). The screening card is then sent to the National Newborn Bloodspot Laboratory (NNBSL) at Children's Health Ireland (CHI) Temple Street, where the samples are analysed and from where onward care is organised.

The NNBSP provides a quality assured programme for the following conditions which are listed based on when screening commenced in Ireland (see Appendix 1 for further details on conditions screened for):

- 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-coenzyme A dehydrogenase deficiency (MCADD) - 2018
- Glutaric aciduria type 1 (GA1) - 2018
- Adenosine deaminase deficiency severe combined immunodeficiency (ADA-SCID) - 2022

For more details on the conditions screened for please refer to 'A Practical Guide to Newborn Bloodspot Screening 9th Edition (2022) available at: https://www.hse.ie/eng/health/child/newbornscreening/newbornbloodspotscreening/information-for-professionals/a-practical-guide-to-newborn-bloodspot-screening-in-ireland.pdf

Conditions included in the NNBSP must fulfil criteria which have met the internationally accepted standards for newborn bloodspot screening. Ireland's National Screening Advisory Committee (NSAC) has been in place since November 2019 and the addition of new conditions to be screened for is considered and decided upon by the NSAC who then make recommendations to the Minister for Health. More details on the criteria and process can be found at: https://www.gov.ie/en/campaigns/nsac/

All nine conditions currently screened for as part of the NNBSP have a relatively high incidence in the Irish population as described in Table 1.

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

Condition	Date started	Irish incidence (2017-2023)	Worldwide incidence*
Phenylketonuria (PKU)	1966	1:4,065	1:12,000
Homocystinuria (HCU)	1971	1:68,433	1:120,000
Classical galactosaemia (CGAL)	1972	1:11,405	1:45,000
Maple syrup urine disease (MSUD)	1972	1:410,597	1:225,000
Congenital hypothyroidism (CHT)	1979	1:891	1:3,500
Cystic fibrosis (CF)	2011	1:2,150	1:3,500
Glutaric aciduria type 1 (GA1)¥	2018	1:143,598	1:100,000
Medium chain acyl-coenzyme A dehydrogenase deficiency (MCADD) ¥	2018	1:16,894	1:14,600
Adenosine deaminase deficiency severe combined immunodeficiency (ADA-SCID)$	2022	1:78,500**	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 five years of screening (2019-2023)

$Screening for ADA-SCID commenced in May 2022

** Burns et al (2021) Severe Combined Immunodeficiency (SCID) – The Irish Experience[1]

In January 2023, the Minister for Health, requested the HSE to add severe combined immunodeficiency (SCID) to the NNBSP after a recommendation from the NSAC following a detailed health technology assessment (HTA) carried out by HIQA.

In November 2023, the Minister for Health issued a further request to the HSE to add spinal muscular atrophy (SMA) to the conditions screened for as part of the NNBSP.

Following successful service plan bids for the required resources to enable this expansion, work is now progressing on the many complex processes necessary to commence screening for SCID and SMA.

This report provides information on the performance of the NNBSP. The report will outline the activity of the National Newborn Bloodspot Screening Laboratory (NNBSL) and the performance of the NNBSP against agreed set of key performance indicators (KPIs). The KPIs were developed and agreed by the NNBSP Governance Group in 2011 as part of the programme of work when adding cystic fibrosis to the NNBSP. The KPIs were selected based on an international review of other screening programmes, particularly the UK, and professional experience at the time. The regular analysis of NNBSP data is a vital tool in the quality assurance of the screening programme.

This report covers data for babies who had their first newborn bloodspot screening sample taken during 2023 and will reference previous years for comparison purposes.

1 Burns et al (2021) Severe Combined Immunodeficiency (SCID) – The Irish Experience. Journal of Clinical Immunology; 41:1950-1953

Key messages for parent(s)/guardian(s)

Parent(s)/guardian(s) are provided with information on newborn bloodspot screening through the Parent Information Leaflet. The Parent Information Leaflet has been translated from English into 14 different languages and has been approved by the National Adult Literacy Agency (NALA). It is provided at an antenatal visit and again at the time consent is being requested and the sample is taken. This is to ensure that when parent(s)/guardian(s) sign the newborn bloodspot screening card they have been fully informed when providing consent for their baby to be screened.

Core messages for parent(s)/guardian(s) are:

- the purpose of newborn bloodspot screening is to help identify babies that may be at risk of having one or more of the very rare 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 enormous with early diagnosis and treatment
- no screening programme is 100% accurate and some babies who are positive for screening will not be confirmed as having the condition (false positives), and, that very rarely some babies who do have the condition are not identified (false negatives/undetected cases)
- all parents for whom there is any concern from the screening are contacted as soon as possible
- the screening programme is recommended for all babies

Information is also available for parent(s)/guardian(s) and the general public on the HSE website.

HSE National Newborn Bloodspot Screening Programme delivery

The HSE is responsible for the delivery of the National Newborn Bloodspot Screening Programme (NNBSP).

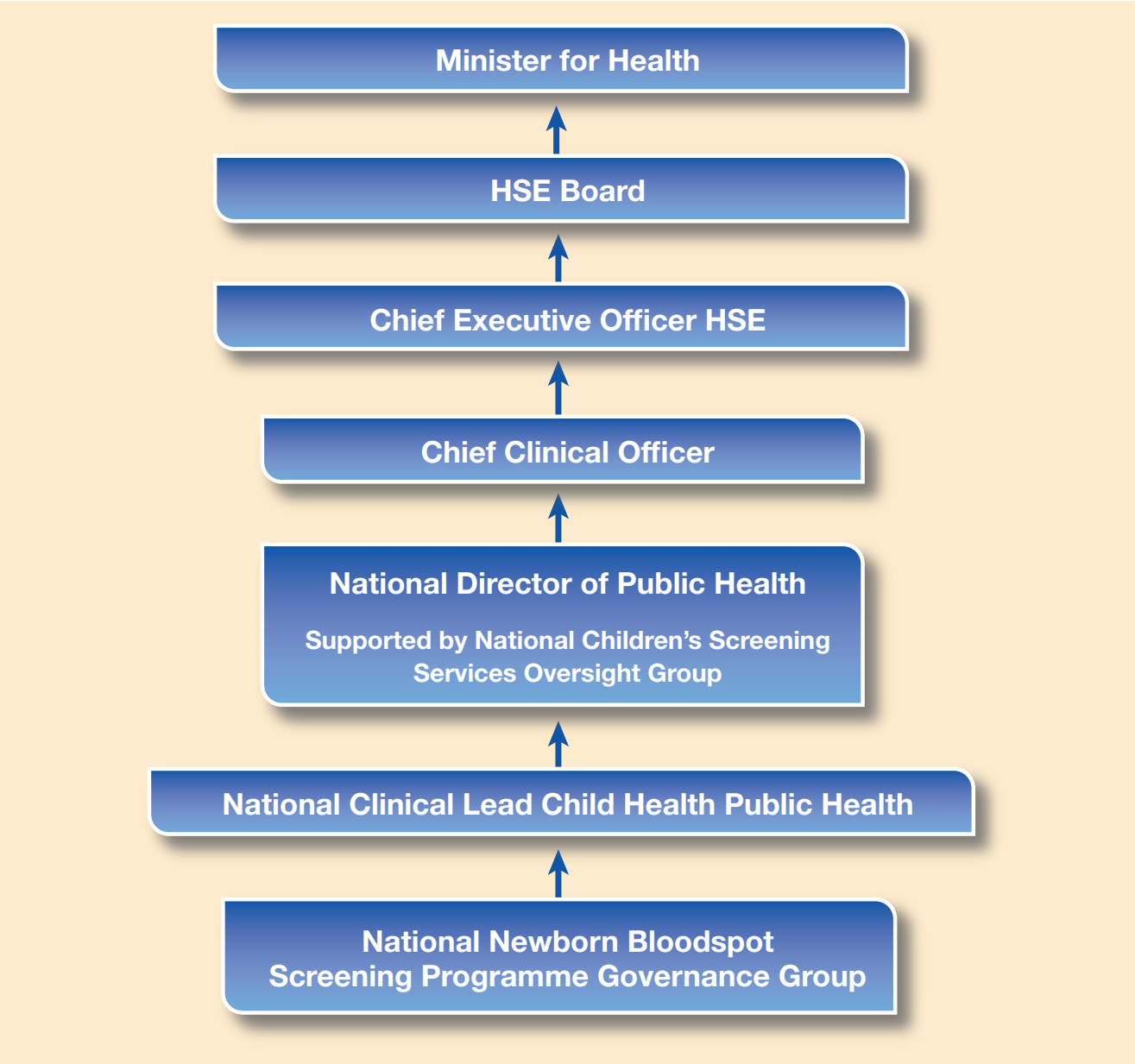
The operational and clinical governance of the NNBSP within the National Healthy Childhood Programme (NHCP) sits within the National Public Health function of the Office of the Chief Clinical Officer of the HSE.

Operationally, the collection of the bloodspot screening sample is carried out by healthcare professionals as part of the national child health service provided to all children - the National Healthy Childhood Programme (NHCP). Samples are taken in maternity hospital/units or by PHNs or Community Midwives if the baby has been discharged home.

The National Newborn Bloodspot Screening Laboratory (NNBSL) at CHI Temple Street provides the 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. The laboratory is accredited by the Irish National Accreditation Board (INAB) to ISO Standard 15189. The NNBSL is also responsible for the coordination of the onward clinical referral of babies who screen positive.

The HSE NNBSP Governance Group oversees the performance management and quality assurance of the NNBSP. It is chaired by the National Clinical Lead Child Health Public Health – see Figure 1 for the current governance structure.

Figure 1: Governance structure for the NNBSP



National newborn bloodspot screening pathway

Newborn bloodspot screening is available to all babies born in the Republic of Ireland. The screening is carried out when the baby is between 72 and 120 hours old (between day 3 and day 5 of life).

Newborn bloodspot screening is also available 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 unless clinically indicated.

When the newborn bloodspot screening sample is taken it is either sent by registered post or transferred by courier to the NNBSL at CHI Temple Street where they are logged and analysed.

Babies who are screen positive; i.e. suspected of having one or more of the conditions screened for, are contacted by their local maternity hospital and are requested to attend their maternity

hospital/unit for medical review, discussion and further testing. If it is required, they are referred by the NNBSL to the appropriate specialist clinical team according to the relevant clinical referral guidelines. At times a direct referral to a paediatric hospital with the specialist clinical team may be required.

Summary statistics

Table 2 below outlines some headline statistics.

Table 2:
NNBSP summary statistics 2020-2023

Metric	2020	2021	2022	2023	Programme target
Number of babies screened	57,016	60,985	54,775	54,820	
Number of samples analysed~	66,122	70,420	62,519	66,455	
% samples taken between 72-120 hours after birth	96.5%	96.0%	96.1%	96.8%	95%
% samples received by NNBSL within 3 working days	99.2%	98.6%*	98.8%	98.9%	100%**
% of screening cards with Unique Perinatal Identifier (UPI) recorded	99.4%	99.5%	99.5%	99.4%	99%
% 'screen positive' clinical referrals performed within 10 days of sample receipt (all conditions except cystic fibrosis)	100%	100%	100%	100%	100%
% referrals sent to cystic fibrosis Centres by 4th week of life	93.8%$	91.9%¥	97.7%	100%	95%

~ see section later on quality of newborn samples/avoidable repeats

* The HSE cyber-attack: the NNBSL ICT system was not available during Q2 2021. This figure is calculated using Q1, Q3 and Q4 only

** KPI target amended to 98.2% from Q1 2024

$ Q4 2020 Analytical problems in the genetics laboratory impacted the achievement of this KPI

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

Note: in-depth analysis of individual performance measures is contained later in this report

Programme acceptability

The acceptability rate of the NNBSP amongst parents is consistently high at over 99.7% between 2020 and 2023. 284 parent(s)/guardian(s) are known to have opted out of the newborn bloodspot screening programme in 2020, 2021, 2022 and 2023 (37, 60, 81 and 106, respectively), a number that has been increasing in recent years. Of note, 10% of those who opted out of the NNBSP in 2023 were parent(s)/guardian(s) of babies born outside of Ireland – this was 25% in 2022 and 10% in 2021.

Quality of newborn bloodspot samples/avoidable repeats

There are clinical reasons why repeat samples are requested, for example premature babies have more than one sample collected, and borderline results are often monitored through repeat sampling. It is very important to ensure that repeat samples required for quality issue; e.g. insufficient or contaminated samples are minimised. The collection of good quality bloodspot samples is important to limit the number of babies requiring repeat sampling, ensuring the efficient detection and referral of babies with suspected conditions. It is also important for minimising parental anxiety and maintaining the acceptability of the programme.

Table 3:
Avoidable repeat samples 2020-2023

	2020	2021	2022	2023	2020-2023
Repeat samples due to quality issues	2,172 (3.8%)	2,349 (3.85%)	2,755 (5.0%)	3,030 (5.5%)	10,306 (4.5%)

Outcomes

The number of babies screened by the NNBSL in 2020 (57,016), 2021 (60,985), 2022 (54,775) and 2023 (54,820) was 227,596 in total.

Table 4 outlines the number of babies confirmed as having one of the conditions that the NNBSP screens for, over the period 2020, 2021, 2022 and 2023 combined.

Table 4:
Confirmed positive cases 2020, 2021, 2022 and 2023 combined

Condition	Number of confirmed positive patients (2020-2023 inclusive)	Rate per 100,000 (2020-2023 inclusive)
Congenital hypothyroidism (CHT)	288	127
Cystic fibrosis (CF)	107	47
Phenylketonuria (PKU)	59	26
Classical galactosaemia (GAL)	20	9
Medium chain acyl-coenzyme A dehydrogenase deficiency (MCADD)	16	7
Homocystinuria (HCU)	4	2
Glutaric aciduria type 1 (GA1)	1	0.4
Maple syrup urine disease (MSUD)	0	0
Total	**495**	**218**

Sensitivity and specificity

Table 5 shows sensitivity and specificity for the conditions screened for by the NNBSP for 2020, 2021, 2022 and 2023.

Table 5:
NNBSP sensitivity and specificity, 2020-2023¥

	Sensitivity (%)				Specificity (%)				PPV (%)
	2020	2021	2022	2023	2020	2021	2022	2023	2017-2023$
PKU	100	100	100	100	99.99	99.99	99.99	99.99	88.6
MSUD	*	*	*	*	99.99	99.99	99.99	*	4.55
HCU	100	100	100	*	99.94	99.94	99.99	99.95	2.65
GAL	100	100	100	100	99.93	99.99	99.90	99.97	44.44
CHT	100	100	100	100	99.94	99.95	99.97	99.97	62.05
CF	100	100	100	100	99.87	99.9	99.88	99.89	30.56
MCADD	100	100	100	100	99.97	100	99.96	99.99	20.48
GA1	*	*	100	*	99.97	99.99	99.99	99.98	4.0

¥ Some conditions undetected by newborn screening may not be diagnosed for years after screening. Results presented are based on current data relating to 2020, 2021, 2022 and 2023. Data are updated if a clinical diagnosis is made at a later date

* Unable to calculate sensitivity, no true positive cases detected in these years (no false positive for MSUD 2023)

$PPV data reviewed from 2017-2023 due to small numbers of cases detected in the years 2020-2023. PPV for MCADD and GA1 included using 2019-2023 data as screening only commenced in December 2018

Sensitivity refers to a screening method's ability to designate an individual with a condition as positive. A highly sensitive method, e.g. 100%, minimises the possibility of false negatives – e.g. missing a case.

Specificity is the ability of a screening method to correctly identify an individual as not having a condition.

Positive predictive value (PPV) is the probability that after receiving a positive screening result an individual will definitely have the condition.

A **false positive** is a screening result that indicates that a person is at risk of having a specific disease or condition. However after further confirmatory tests, the person was negative or shown not to have this specific disease. A screening programme aims to have as low a number of false

positives as possible to minimise anxiety for parents who have to bring their baby for further tests to be eventually reassured that their baby is fine. A low false positive rate is also important to maximise parent acceptability to participate in the screening programme.

A **false negative** is a screening result that indicates that a person is not at risk of a specific disease or condition when the person actually does have the disease or condition (i.e. a patient condition was not detected during screening). A screening programme aims to have minimal amount of false negatives as these equate to undetected cases. It is recognised that screening is not 100% accurate and false negatives are expected as part of screening programmes.

Data on individual conditions

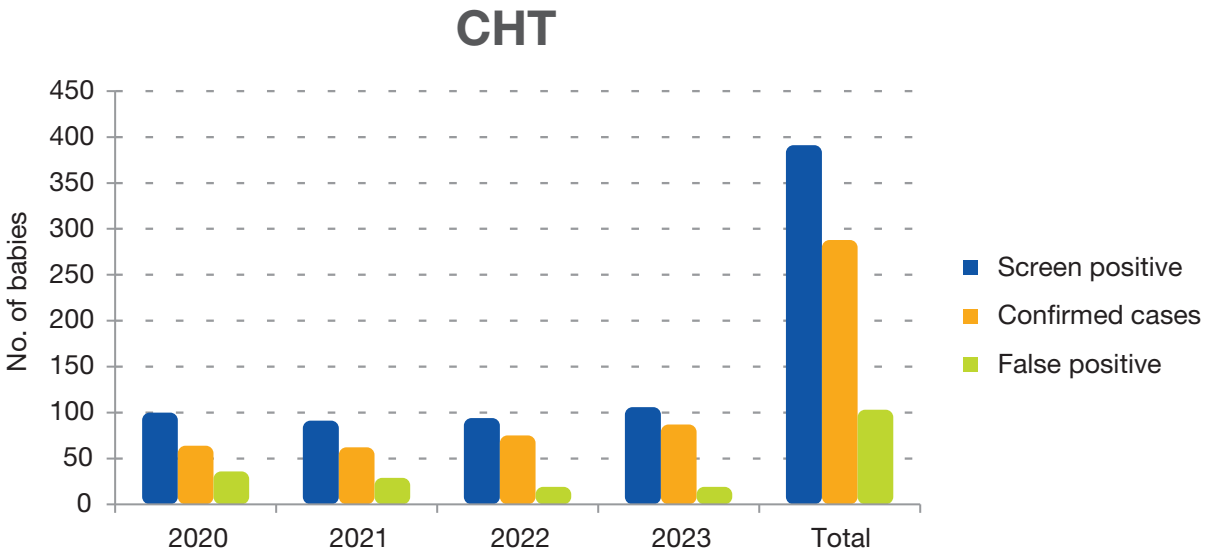
Congenital hypothyroidism (CHT)

Congenital hypothyroidism (CHT) is the most commonly diagnosed condition that is screened for by the NNBSP. Between 2020 and 2023 there were 288 confirmed cases of CHT. There were 103 babies who were ultimately determined to be false positive cases. In 2020 the NNBSP added the disorder type 'transient hypothyroidism' to more accurately describe and record a cohort of babies who would previously have been 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 'transient hypothyroidism' were recorded as 'false positive'. This had no impact on the management of the baby.

Table 6:
Congenital hypothyroidism 2020-2023

Congenital hypothyroidism	2020	2021	2022	2023	Total
Screen positive cases	100	91	94	106	391
Confirmed cases	64	62	75	87	288
Dysgenesis	*16*	*8*	*6*	*19*	*49*
Dyshormonogenesis	*15*	*26*	*33*	*24*	*98*
Unclassified	*32*	*24*	*24*	*27*	*107*
Transient hypothyroidism	*1*	*4*	*11*	*17*	*33*
False positive cases	36	29	19	19	103

Figure 2:
CHT data 2020-2023



Cystic fibrosis (CF)

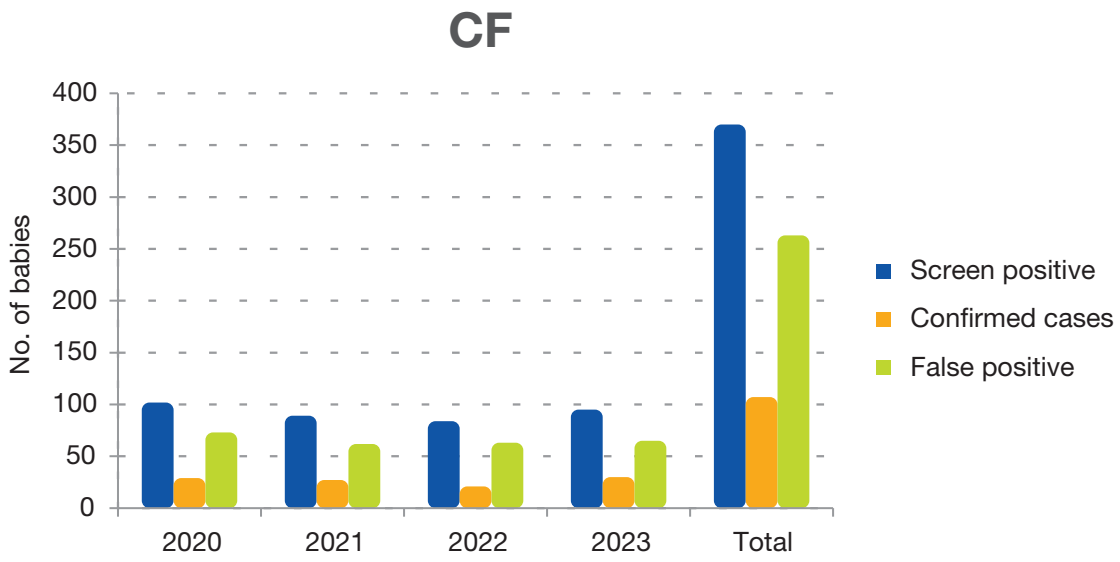
Between 2020 and 2023 3,056 samples were sent to the associated referral laboratory for genetic mutation analysis following a screen positive result on the bloodspot. This represented 1.3% of all initial newborn samples received between 2020 and 2023. Following mutation analysis, 370 babies were referred to a CF Centre for sweat testing; with 107 babies diagnosed with CF. There are six dedicated Paediatric CF centres across Ireland.

Please also refer to the later section that describes the recent ten year review of the CF newborn bloodspot screening programme.

Table 7:
Cystic fibrosis 2020-2023

Cystic fibrosis	2020	2021	2022	2023	Total
No. referred for sweat test (screen positives)	102	89	84	95	370
No. with two CFTR mutations	*22*	*24*	*20*	*20*	*86*
No. with one CFTR mutation	*80*	*65*	*64*	*71*	*280*
Confirmed cases	29	27	21	30	107
False positive cases	73	62	63	65	263

Figure 3:
CF data 2020-2023



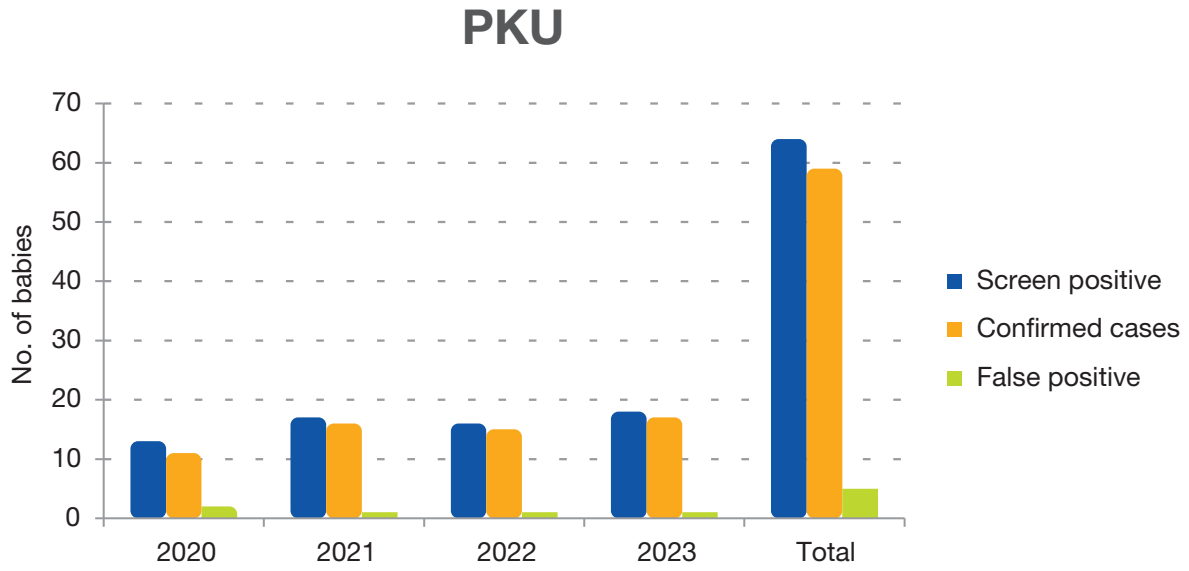
Phenylketonuria (PKU)

There were 59 babies diagnosed with PKU between 2020 and 2023. There were 5 babies who had false positive PKU screen.

Table 8:
Phenylketonuria 2020-2023

Phenylketonuria	2020	2021	2022	2023	Total
Screen positive cases	13	17	16	18	64
Confirmed cases	11	16	15	17	59
False positive cases	2	1	1	1	5

Figure 4:
PKU cases 2020-2023



Classical galactosaemia (CGAL)

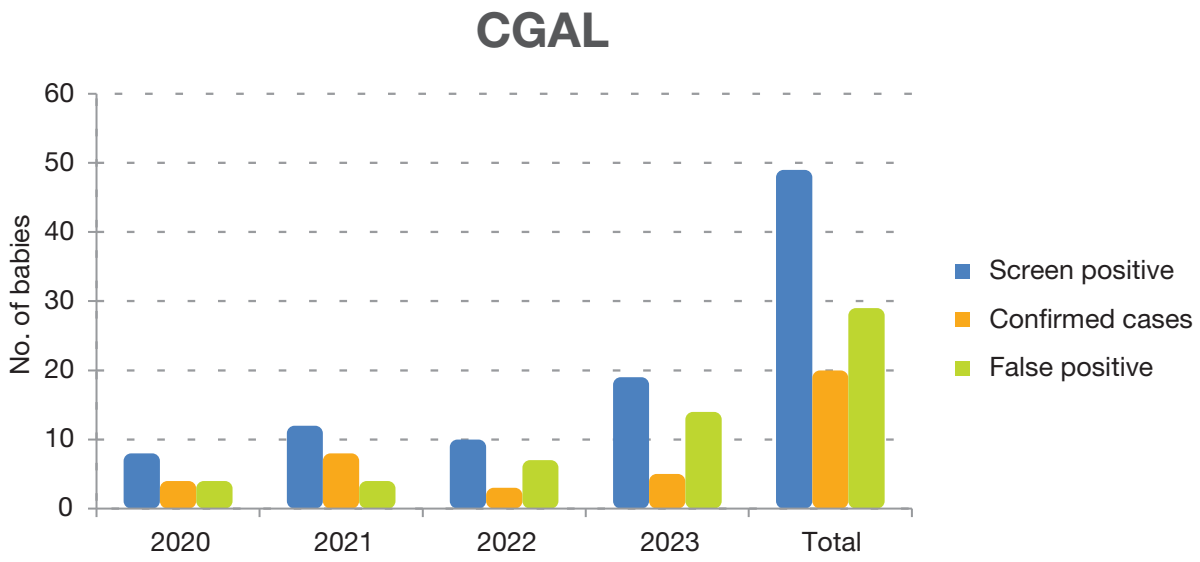
There were 20 cases of CGAL diagnosed between 2020 and 2023. There were 29 false positives.

The NNBSL also performs high risk screening for babies at increased risk of Classical galactosaemia (members of the Irish Traveller community and siblings of known cases of CGAL). The sample for this (Beutler) test can be collected on day 1 of life.

Table 9:
Classical galactosaemia 2020-2023

Classical galactosaemia	2020	2021	2022	2023	Total
Screen positive cases	8	12	10	19	49
Confirmed cases	4	8	3	5	20
False positive cases	4	4	7	14	29

Figure 5:
CGAL cases 2020-2023



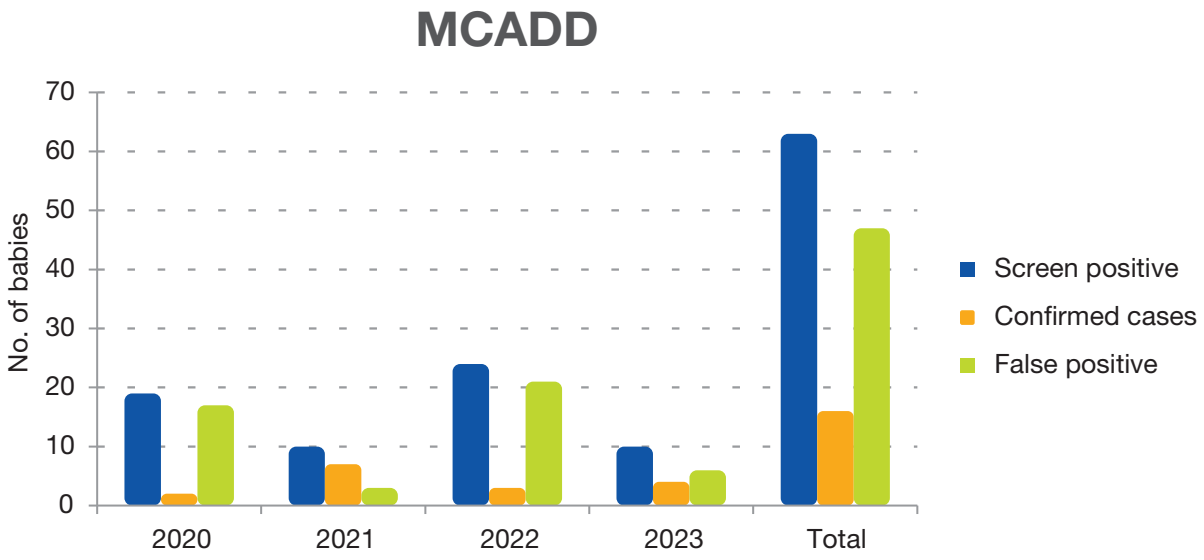
Medium chain acyl-coenzyme A dehydrogenase deficiency (MCADD)

Screening for MCADD commenced in December 2018 and between 2020 and 2023 there were 16 confirmed cases of MCADD detected with 47 false positives.

Table 10:
Medium chain acyl-coenzyme A dehydrogenase deficiency 2020-2023

Medium chain acyl-coenzyme A dehydrogenase deficiency	2020	2021	2022	2023	Total
Screen positive cases	19	10	24	10	63
Confirmed cases	2	7	3	4	16
False positive cases	17	3	21	6	47

Figure 6:
MCADD cases 2020-2023



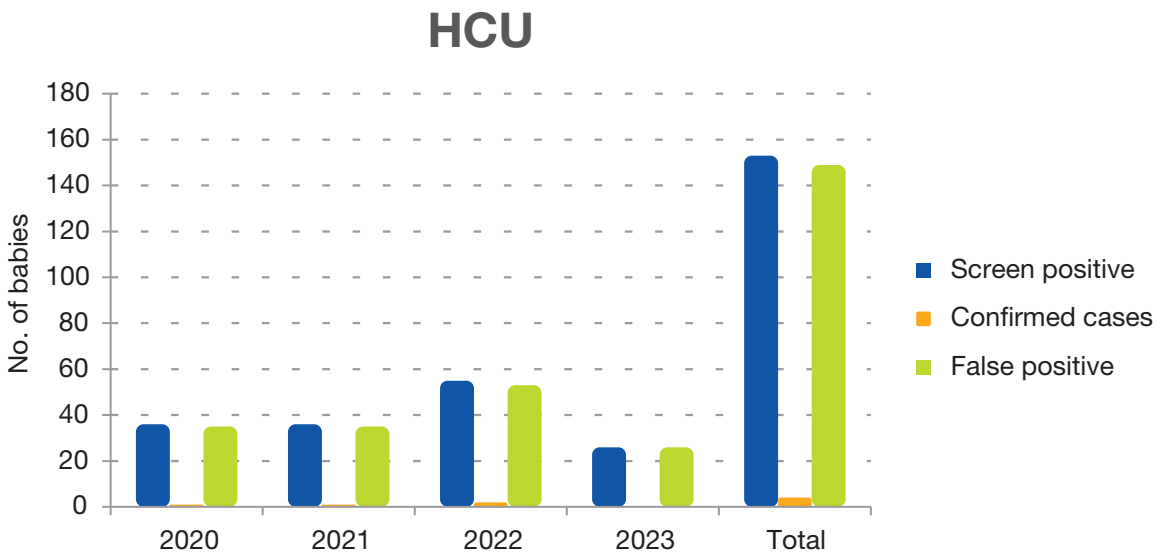
Homocystinuria (HCU)

There were 4 cases of HCU detected between 2020 and 2023 and 151 false positive cases. This is a rate of 68/100,000. A higher false positive rate for HCU is an expected outcome as it is considered a disorder that is difficult to detect through newborn screening. A lower analytical cut-off is in place to increase sensitivity but this can reduce specificity.

Table 11:
Homocystinuria 2020-2023

Homocystinuria	2020	2021	2022	2023	Total
Screen positive cases	36	36	55	28	155
Confirmed cases	1	1	2	0	4
False positive cases	35	35	53	28	151

Figure 7:
HCU cases 2020-2023



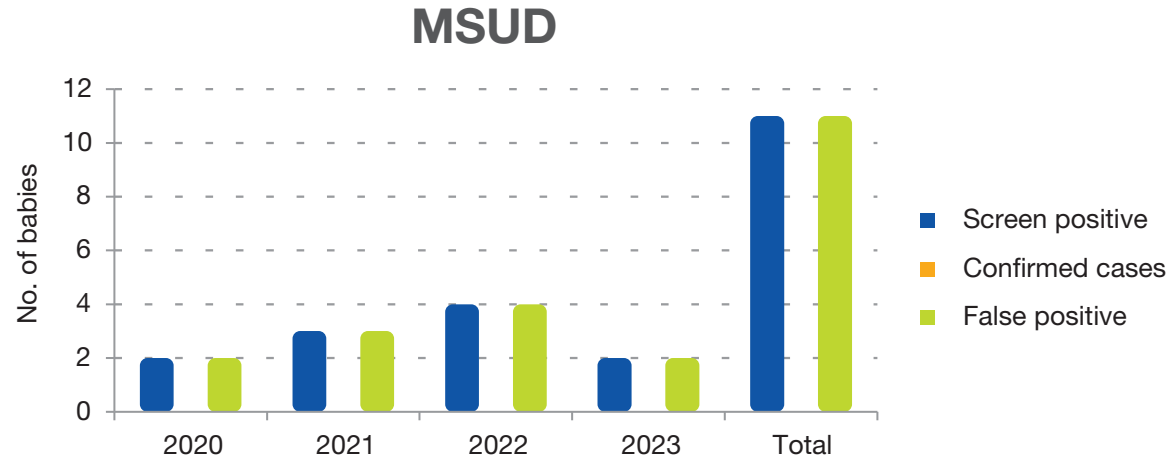
Maple syrup urine disease (MSUD)

There were no cases of MSUD detected between 2020 and 2023 with 11 false positives.

Table 12:
Maple syrup urine disease 2020-2023

Maple syrup urine disease	2020	2021	2022	2023	Total
Screen positive cases	2	3	4	2	11
Confirmed cases	0	0	0	0	0
False positive cases	2	3	4	2	11

Figure 8:
MSUD cases 2020-2023



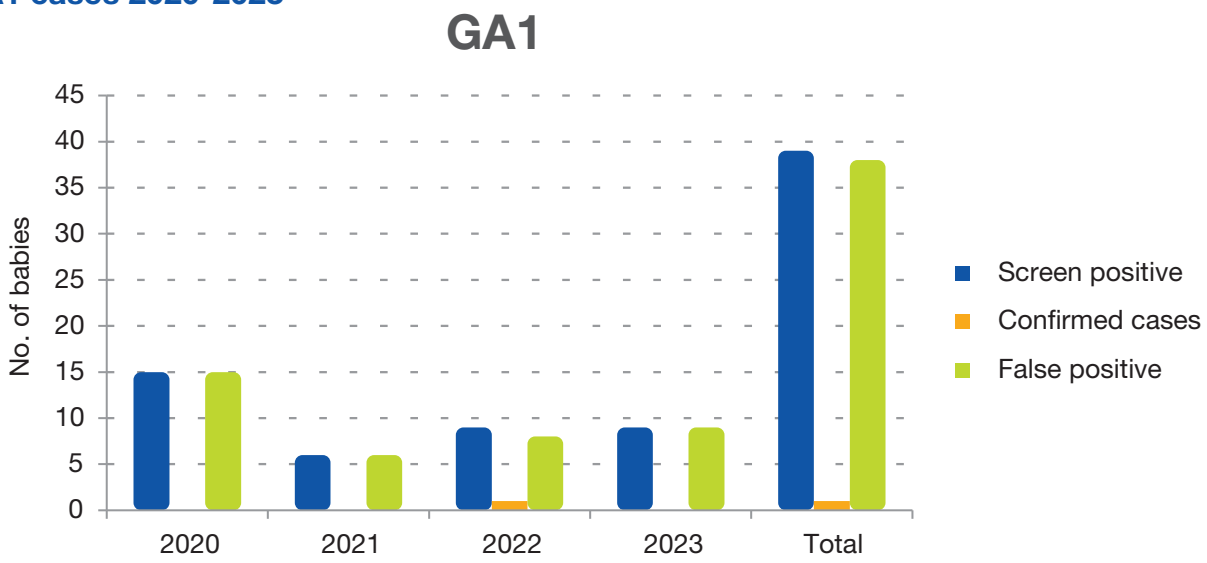
Glutaric aciduria type 1 (GA1)

There was 1 case of GA1 detected between 2020 and 2023 with 38 false positives. There was a relatively high false positive rate in 2020 (26/100,000) due to the use of a conservative cut-off at the introduction of screening. There were 6 false positives in 2021, 8 in 2022 and 9 in 2023.

Table 13:
Glutaric aciduria type 1 (GA1) 2020-2023

Glutaric aciduria type 1	2020	2021	2022	2023	Total
Screen positive cases	15	6	9	9	39
Confirmed cases	0	0	1	0	1
False positive cases	15	6	8	9	38

Figure 9:
GA1 cases 2020-2023



Adenosine deaminase deficiency severe combined immunodeficiency (ADA-SCID)

Screening for ADA-SCID commenced in May 2022. There were no cases of ADA-SCID identified in 2022 or 2023 and no false positives.

False negatives

The NNBSP takes a proactive approach to the investigation of clinically diagnosed cases, undetected through the screening programme i.e. potential false negatives. The identification and review of these undetected cases is considered by the NNBSP to be an integral part of the programme's quality assurance. Clinical colleagues for the metabolic conditions, ADA-SCID and CF are requested to immediately inform the NNBSP if a case (of a condition screened for) is diagnosed, so a formal review can take place. For these conditions, the NNBSP has developed documentation to collect the data required. An annual formal communication is sent, calling for any clinical cases that have presented which were not detected through the screening programme.

Any such cases the NNBSP is informed of are investigated with respect to the screening programme. As part of the review any identified possible corrective actions are considered, and based on these, quality improvements may be made to the programme. A formal report is finalised and shared with the appropriate governance line and treating clinician. Congenital hypothyroidism (CHT) will also be included in this process from 2025.

The NNBSP has not been informed of any false negative cases for babies screened between 2020 and 2023. If the NNBSP is informed of a case that is then determined to have been undetected following investigation, the data relating to the year of the baby's birth is updated to reflect this.

In the years 2020-2023, the NNBSP have completed investigations of five undetected cases for babies born outside of those years. In each of the cases, the investigation did not identify any non-compliance in the screening pathway as causative for the inability to detect the disorder. As with all screening programmes, unfortunately, not all cases will be detected. Following these reviews, any quality improvement measures which could be identified were implemented. Where learning has arisen, this is documented and will form part of reviews and, if relevant, will inform future discussions with the NSAC in relation to the particular programme.

Standards for the NNBSP

This section outlines the standards set 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 has five NNBSL based key performance indicators (KPIs) to performance manage the screening programme. These are collated on a quarterly basis and reported to the NNBSP Governance Group.

KPI 1: Timeliness of sample collection

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

This standard was met in 2020, 2021, 2022 and 2023 with 96.5% of all samples in 2020 taken between 72 and 120 hours after birth with the corresponding figure for 2021 at 95.8%, for 2022 was 96.1% and for 2023 it was 96.8%. The NNBSP proposed including this KPI in the suite of KPIs included in the HSE National Service Plan.

KPI 2: Timeliness of sample dispatch

Standard: 100% of samples are received by the NNBSL within 3 working days of the sample being taken

In 2020, 99.2% of all samples were received by the NNBSL within 3 working days of the sample being taken, in 2021 this figure was 98.6%, in 2022 it was 98.8% and for 2023 it was 98.9%. This has been consistently below the programme target of all samples to be in the lab within three working days and in 2023 the NNBSP carried out a detailed review of this KPI. After review by the NNBSP of all the quarterly data available since 2017, it was agreed to reduce the target for this KPI to 98.2% to ensure better visibility of any occasion/area where the screening card is not systematically received in an acceptably timely way and this can be investigated. The NNBSP Governance Group will continue to monitor this KPI against this revised target.

Note: In 2021 the cyber-attack on the HSE prevented the NNBSL from electronically processing the receipt of the screening cards into the laboratory information system for May and June; therefore the figure of 98.6% for 2021 is calculated on cards received in quarter 1, quarter 3 and quarter 4 only.

KPI 3: Enhanced tracking abilities

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

In 2020, 99.4% of all newborn bloodspot screening cards had the correct UPI recorded, in 2021 this was 99.5%, in 2022 it was 99.5% and in 2023 it was 99.4%, consistently in excess of the target of 99%.

In 2018 the target for this KPI was 95% and was increased to 99% to encourage adherence. This is a crucial identifier to track babies throughout the screening process in the absence of a national Individual Health Identifier.

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

Once a screen positive is identified in the NNBSL it is important follow-up actions are taken promptly.

a. **Standard: 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 for these referrals is 100% and that has been consistently met by the screening programme across 2020, 2021, 2022 and 2023. This ensures that all confirmed cases are referred to the relevant clinical team for timely access to clinical review and commencement of required treatment. Of note, at the point of clinical referral, appointments and admissions are organised and are predominantly for the same day or the following day.

b. **Standard: 95% of clinical referrals for CF to a CF specialist centre by 4th week of life.**
   In 2020 the performance of this KPI was 93.8% and 91.9% in 2021 which are below the recommended target of 95% but improved to 97.7% in 2022 and was 100% for 2023. In previous years this KPI consistently reports at 100%.

   In Q2 and Q4 2020 there were analytical issues in the referral laboratory for genetic analysis. This impacted in that laboratory's turnaround time and resulted in subsequent delays, up to a maximum of 2 week delay, in making clinical referrals for a number of screen positive cases. In Q2 2021 the cyber-attack on the HSE's IT systems impacted on the reporting of this KPI.

Ten year review of the Irish Newborn Bloodspot Screening Programme for cystic fibrosis

Cystic fibrosis (CF) is a multisystem autosomal recessive disorder caused by mutations in the cystic fibrosis transmembrane regulator (CFTR) gene.[2] CF is the most commonly-inherited disease of white populations, where the overall incidence is 1 in 2,500.[3] Ireland's incidence has always been considered one of the world's highest rated.[4,5] In the 1930s children with CF rarely survived infancy.[6] Today, the median predicted survival age for those born in Ireland is 52 years.[7] This is largely due to

2 Brown SD, White R, Tobin P. Keep them breathing: Cystic fibrosis pathophysiology, diagnosis, and treatment. JAAPA. 2017;30(5):23-7.
3 Davies JC, Alton EW, Bush A. Cystic fibrosis. BMJ. 2007;335(7632):1255-9
4 Farrell P, Joffe S, Foley L, Canny GJ, Mayne P, Rosenberg M. Diagnosis of cystic fibrosis in the Republic of Ireland: epidemiology and costs. Irish medical journal. 2007;100(8):557-9.
5 Sasaki E, Kostocenko M, Lang N, Clark T, Rogers M, Muldowney R, et al. National Newborn Screening for cystic fibrosis in the Republic of Ireland: genetic data from the first 6.5 years. Eur J Hum Genet. 2020;28(12):1669-74
6 Nazareth D, Walshaw M. Coming of age in cystic fibrosis - transition from paediatric to adult care. Clin Med (Lond). 2013;13(5):482-6
7 Cystic Fibrosis Ireland. Cystic Fibrosis Ireland - Annual Report for 2022 Dublin 2023 [Available from: https://www.cfireland.ie/images/uploads/resources/CFI_2022_Annual_Report_LR.pdf.]

earlier diagnosis, which has resulted in most having minimal lung disease at diagnosis. Mutation-specific therapies have also improved the prognosis of CF.[8,9]

Newborn screening for CF (NBSCF) is a key enabler of early diagnosis and prompt treatment, addressing pancreatic insufficiency, preventing malnutrition, and improving lung function.[10] NBSCF

is cost-effective.[11] However there are some disadvantages to NBSCF including the identification of carriers, false positive and inconclusive results, the psychological impact on parents of abnormal results, and potential inequities caused by using screening genetic panels centred around Caucasian populations.[12]

The Irish NBSCF programme commenced in July 2011.[13] Screening is performed on dried bloodspot samples taken when the neonate is 72-120 hours old, to measure concentrations of immunoreactive trypsinogen (IRT). High IRT concentrations, often seen in neonates with CF, prompt screening for CFTR gene mutations via a 38-mutation panel.[6,14]

The overall aim of this programme review was to evaluate the performance of the Irish NBSCF Programme from 2011 to 2021. This included describing the processes in the Irish NBSCF protocol, and comparing programme validity to European Cystic Fibrosis Society (ECFS) standards.[9] An analysis of two CF datasets collected by the National Newborn Bloodspot Screening Programme (NNBSP) was conducted. This dataset included neonates identified as cases of CF, CF carriers and Cystic Fibrosis Screen Positive Inconclusive Diagnosis (CFSPID) during the evaluation period.

Over the ten year evaluation period, 650,809 newborn bloodspot screens were performed, 290 neonates were diagnosed with CF, 533 identified as carriers and 21 classified as CFSPID. Of neonates with CF, 59 (20%) presented with meconium ileus (MI). NBSCF identified 284 (98%) of the children with CF. Six undetected cases presented clinically or following the CF diagnosis of a sibling. The observed incidence of CF over the evaluation period was 1 in 2,203. While lower than previously reported, Ireland's incidence remains Europe's highest.

The sensitivity of the NBSCF Programme was 97.93% (95% CI 96.29–99.57).The specificity was 99.91% (95% CI 99.91-99.92%). The positive predictive value was 0.34 (95% CI 0.31-0.37). These results exceeded recommended European Cystic Fibrosis Society (ECFS) Standards.

There were six undetected cases over the ten year period – false negative results. These were caused by IRT values below the cut-off value or CFTR mutations not included in the 38 mutation panel. Of the six undetected cases, four had CFTR mutations undetected by the 38-mutation panel, and two had screening sample IRT levels below the cut-off value. Upon evaluation of the cases where the mutations were undetected, three were of non-Caucasian ethnicity. With regard to the two cases where by the screening sample IRT level was below the cut-off, lowering the criterion of positivity for IRT levels significantly increases the risk of false positives. Alternatively, increasing the size of the genetic panel would result in increased detection of CFSPID or carriers. Therefore after full evaluation of the data for the programme and expert discussion, currently neither of these

8 Castellani C, Duff AJA, Bell SC, Heijerman HGM, Munck A, Ratjen F, et al. ECFS best practice guidelines: the 2018 revision. J Cyst Fibros. 2018;17(2):153-78

9 Quon BS, Rowe SM. New and emerging targeted therapies for cystic fibrosis. BMJ. 2016;352:i859.

10 Scotet V, Gutierrez H, Farrell PM. Newborn screening for CF across the globe—Where is it worthwhile? International journal of neonatal screening. 2020;6(1):18

11 Nshimyumukiza L, Bois A, Daigneault P, Lands L, Laberge AM, Fournier D, Duplantie J, Giguère Y, Gekas J, Gagné C, Rousseau F, Reinharz D. Cost effectiveness of newborn screening for cystic fibrosis: a simulation study. J Cyst Fibros. 2014 May;13(3):267-74. doi: 10.1016/j.jcf.2013.10.012. Epub 2013 Nov 12. PMID: 24238947.

12 Dankert-Roelse J, Vernooij-van Langen A. Newborn screening for cystic fibrosis: pros and cons. Breathe. 2011;8(1):24-30

13 Health Information and Quality Authority. Review of processes in use to inform the expansion of newborn bloodspot screening programmes Dublin, Ireland; 2021.

14 Marsden P. Procedure for all Community and Hospital Services Providing the National Newborn Bloodspot Screening Programme (NNBSP).

changes are recommended. Close attention and further interrogation of data will continue on these two areas.

In conclusion, the Irish NBSCF programme meets and compares favourably to ECFS standards. Ongoing validity monitoring and interrogation of data are vital and will continue through the governance structures, and published as per annual reports.

Review of Irish newborn screening programme for medium chain acyl-coenzyme A dehydrogenase deficiency (MCADD)

Ireland added newborn screening for MCADD to the NNBSP in December 2018. A recently published study by Howard et al[15] reviewed the initial three years of MCADD screening with the aim of reassessing the incidence of MCADD since screening commenced and to characterise detected cases biochemically, clinically and genetically. The review found that the incidence of MCADD in Ireland was higher than previous estimates but the biochemical, genetic and clinical profile of patients was in keeping with expectations.

There were 11 true positives identified giving an incidence of 1 in 16,164. Previous estimates of incidence of MCADD ranged from between 1 in 66,000 to 1 in 71,650.

All patients had at least one c.985A>G variant in the ACADM gene and screening C8/C10 ratio values were highest in homozygotes for c.985A>G and compound heterozygotes for c.799G>A/c.985A>G. One infant presented with hypoglycaemia prior to screening results being available. Two had hypoglycaemia related to comorbid diagnosis. No patients in the period of the review had decompensated since diagnosis. One additional case was detected through familial cascade screening.

Regarding false positives, there were 44 false positives detected during the period of the review. The vast majority of these, 84%, were attributed to the use of total parenteral nutrition (TPN) in preterm infants. The type of TPN used in most neonatal units in Ireland has high levels of medium chain triglycerides which has been reported to cause false positives in newborn screening for MCADD. The review showed that the C8 levels in false positives are lower than for true positive cases. There is potential to consider raising the cut off values to reduce the number of false positives but the review did not recommend this at this time, pending collection of further data.

Review of newborn screening programme for medium chain acyl -coenzyme A dehydrogenase deficiency (MCADD) and glutaric aciduria type 1 (GA1)

The NNBSP, as a result of the three year review of MCADD, will progress a detailed five year review of newborn screening for both MCADD and GA1 which were introduced at the same time in December 2018.

Table 14 outlines a summary of the case statistics for MCADD and GA1 since they were introduced and these will be included in the more detailed review to be carried out.

15 Howard et al (2023) Medium Chain Acyl Co-A Dehydrogenase Deficiency: 3 years of Newborn Screening Ir Med J; March 2023; Vol 116; No3; P743

Table 14:
MCADD and GA1 case statistics December 2018-2023

	2018	2019	2020	2021	2022	2023	Total
Babies screened	4,906*	59,591	57,016	60,985	54,775	54,820	292,093
MCADD cases	1	2	2	7	3	4	19
MCADD false positives	4	19¥	17	3	21$	6	70
GA1 cases	0	1	0	0	1	0	2
GA1 false positives	0	10¥	15	7	8$	9	49

*Screening for MCADD and GA1 commenced on 3/12/2018 – this is the number of babies screened between 3/12/2018 and 31/12/2018

¥ Initial lower cut-off values applied for first 6 months of screening increased the numbers of false positives, cut offs revised over time.

$May 2022 a new revised kit method introduced (Neobase 2) to allow introduction of ADA-SCID, cut-off values adjusted and monitored.

National Newborn Bloodspot Screening Programme Governance Group

The NNBSP is overseen by the NNBSP Governance Group that has multidisciplinary membership from paediatrics, public health nursing, midwifery, laboratory, public health medicine and administrative staff. The NNBSP Governance group met on three occasions during 2020. The planned April 2020 meeting was postponed due to the involvement of a large number of members in the Covid-19 pandemic response. The NNBSP Governance Group met four times in 2021, 2022 and 2023 with an additional specific meeting in June 2022 to review the first four weeks of ADA-SCID screening.

The NNBSP Governance Group assembles smaller subgroups to progress specific action points. Relevant members of the governance group will also convene urgent meetings with relevant members if they are informed of a potential undetected case to ensure a timely and appropriate review can commence.

The NNBSP Governance Group has, when requested, supported the work of the National Screening Advisory Committee (NSAC). The NNBSP Governance Group contributes to HIQA processes as they work to support the work of the NSAC to inform expansion of the NNBSP. Members of the NNBSP Governance Group contributed to two HIQA health technology assessments (HTA) on the addition of severe combined immunodeficiency (SCID) and spinal muscular atrophy (SMA) to the NNBSP.

In January 2023, the Minister for Health, requested the HSE to add SCID to the NNBSP after a recommendation from the NSAC following a detailed HTA carried out by HIQA.

In November 2023, the Minister for Health issued a further request to the HSE to add spinal muscular atrophy (SMA) to the conditions screened for as part of the NNBSP following a recommendation from the NSAC.

The resources required to include these two conditions were successfully service planned for and the complex processes required to add SCID and SMA are now commending and will continue in to 2025.

The NNBSP Governance Group will continue to actively and keenly participate in these processes to support expansion to the NNBSP as recommended by the NSAC and requested by the Minister for Health.

Section 2

National Universal Newborn Hearing Screening Programme

Introduction

Approximately 1 to 2 babies in every 1,000 are born with a hearing loss in one or both ears. The overall aim of the National Universal Newborn Hearing Screening Programme (NUNHSP) is to improve the health and well-being of children through high quality hearing assessments and early intervention. Early diagnosis and appropriate intervention for permanent childhood hearing loss (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 long-term costs associated with special education and support, as well as personal, family and societal costs resulting from lower educational achievement, poor employment prospects, and potential mental health problems. The NUNHSP is available for all eligible babies within Ireland and approximately 80-90 cases of permanent childhood hearing loss are diagnosed each year.

The HSE contracted NEC Software Solutions, previously called Northgate Public Services, to deliver the screening element of the NUNHSP. They aim to provide complete newborn hearing screening to all eligible babies in the Republic of Ireland by the time they are 12 weeks old.

Newborn hearing screening is undertaken by trained screening staff at each of the 19 maternity hospital/units using the Natus AccuScreen devices which are specifically designed for use with newborn babies.

This report covers data for babies who were born in 2020, 2021, 2022 and 2023.

Key messages for parent(s)/guardian(s)

- Newborn hearing screening identifies if your baby has a hearing loss that could affect their speech and language development without early intervention and support.
- Newborn hearing screening is available to all eligible babies in the Republic of Ireland and is undertaken soon after birth.
- 1 to 2 babies in every 1,000 are born with a hearing loss in one or both ears.
- 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.
- 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 process does not hurt or harm the baby in anyway.
- The NUNHSP 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 (PCHL) are diagnosed each year through the NUNHSP programme.

As with all screening programmes, not every case of hearing loss will be detected e.g. mild hearing loss or cookie bite hearing loss. It is also possible 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 older.

HSE National Universal Newborn Hearing Screening Programme delivery

The HSE is responsible for the delivery of the National Universal Newborn Hearing Screening Programme (NUNHSP). The NUNHSP, within the National Healthy Childhood Programme (NHCP), sits within the National Public Health function of the Office of the Chief Clinical Officer.

Operationally, the newborn hearing screening is carried out by hearing screeners as part of the child health service. The vast majority of newborn hearing screening takes place in the maternity hospital/unit but if not completed in the maternity hospital/unit, the baby can be called back to an outpatient clinic to complete the screening. The HSE has contracted NEC Software Solutions, previously called Northgate Public Services, to provide the newborn hearing screening element of the NUNHSP. NEC Software Solutions provides the NUNHSP IT management system, Smart 4 Hearing (S4H), which tracks and records all the data for the NUNHSP. The hearing screeners are responsible for the onward referral of babies who have no clear response from their screening. These babies are referred to the local audiology team for further assessment, work up and diagnosis.

The HSE's National Universal Newborn Hearing Screening Programme Governance Group is responsible for the safety, effectiveness and quality of the NUNHSP and its associated clinical pathways. The NUNHSP Governance Group consists of a range of professionals involved in the identification and management of infant hearing loss. They monitor the quality and performance of this nationally organised hearing screening programme, and works with expert groups to make sure the screening programme is based on the latest evidence and meets high standards. It is chaired by a Consultant in Public Health who reports to the National Clinical Lead for Child Health Public Health who reports into the National Director of Public Health and onto the Chief Clinical Officer for the HSE – see Figure 10 for the current governance structure.

Figure 10:
Governance structure for the NUNHSP

The NUNHSP collates a wide range of metrics with 21 assigned key performance indicators (KPIs) in order to monitor the programme, as well as providing operational data for day to day running of the service. The metrics are recorded in the UNHS national database, S4H. The NUNHSP Governance Group reviews the NUNHSP performance against the 21 key performance indicators on a quarterly basis.

National universal newborn hearing screening pathway

Newborn babies who are eligible for screening are identified in each maternity hospital/unit from the local birth register and added to the birth list for screening. The screener will carry out the initial screen using the approved device after having a conversation with the parent(s)/guardian(s) to ascertain family history, risk factors and to obtain consent.

Babies who have a clear response in both ears have completed screening and the parent(s)/guardian(s) are informed of the results and the baby discharged. If risk factors are present the baby is offered a nine months paediatric audiology review in the HSE Community Audiology service.

If the baby has no clear response in one or both ears they need to have a second screen carried out. If, after the second screen, there is still no clear response in one or both ears the baby is referred for a diagnostic audiological assessment. The appointment should be made within one working day and the parent(s)/guardian(s) informed that they should receive notice of this appointment in 2-3 working days and parent(s)/guardian(s) are provided with the information leaflet '*Your Baby's Visit to the Audiology Clinic*'.

Diagnostic assessments are carried out by HSE Audiologists within 28 days of referral from the corrected date of birth (or 28 days after screening completion whichever is greater). Babies identified with a hearing loss are fitted with hearing aids if appropriate and supported by the early years support professionals.

Babies not eligible for the NUNHSP include babies born with congenital aural atresia and microtia. For the following babies, referrals should be made by the paediatrician to the local audiology service for hearing assessment as appropriate. The diagnostic audiology appointments may be facilitated through the local screeners:

- babies who have a prolonged period (greater than 6 months) in Special Care Baby Unit (SCBU)/ Paediatric Intensive Care Unit (PICU)
- babies who have suspected or confirmed congenital cytomegalovirus (cCMV)
- babies who have suspected or confirmed Zika virus
- 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.

Summary statistics

Of all the babies screened (2020-2023), 98.5% have a clear response with no follow up required. 1.5% of babies were referred to audiology as a result of having no clear response during screening – see Table 16.

Table 15:
NUNHSP summary data 2020-2023

2020-2023	No.	%
Clear response – No follow-up required	221,450	98.5
Clear response – Targeted follow-up required	882	0.4
Referred to Audiology (no clear response)	2,600	1.1
Total	**224,932**	**100**

Table 16:
NUNHSP summary statistics 2020-2023

Metric	2020	2021	2022	2023	Programme target
Number of babies registered	57,144	60,860	54,780	54,766	N/A
Number of babies eligible for screening	56,619	60,298	54,275	54,316	N/A
Completion of screening					
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%)	>98%
Number of eligible babies completed screening	56,453 (99.88%)	60,161 (99.91%)	54,118 (99.91%)	54,194 (99.91%)	N/A
Number of eligible babies completed screening by 4 weeks	53,766 (95.24%)	59,566 (99.01%)	53,759 (99.3%)	53,973 (99.6%)	>95%
Referrals					
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%)	≤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%)	≤6%
Number of eligible **well** baby referrals from AABR	514 (1.00%)	345 (0.63%)	343 (0.69%)	287 (0.58%)	N/A
Number of eligible **NICU** baby referrals from OAE	572 (10.83%)	572 (11.12%)	556 (11.85%)	588 (11.7%)	N/A
Number of eligible **NICU** baby referrals from AABR	265 (5.02%)	283 (5.5%)	240 (5.11%)	327 (6.51%)	N/A
Audiology (immediate and targeted follow up)					
Number of eligible babies referred to Audiology (Immediate and targeted follow up)	983 (1.74%)	868 (1.44%)	819 (1.51%)	824 (1.52%)	N/A
Number of eligible babies referred for immediate diagnostics	781 (1.38%)	630 (1.05%)	586 (1.08%)	614 (1.13%)	≤3%
Number of eligible babies referred for targeted follow up	202 (0.36%)	238 (0.40%)	233 (0.43%)	210 (0.39%)	N/A
Outcomes					
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%)	≥80%

Across the period 2020-2023, the vast majority of babies, 85.7%, commence and complete their hearing screening as an inpatient in the Maternity hospital/unit. A further 9.1% of babies (approximately 5,100 annually), commenced their screening as an inpatient and completed it as an outpatient, indicating that they had no clear response at the first initial screen on the ward. Further, 5.2% (approximately 3,000 annually) both started and completed their screening as an outpatient – see Table 17.

Table 17:
Location of screening 2020-2023

2020-2023	Number	%
Started and completed as inpatient	192,793	85.7
Started as inpatient but completed as outpatient	20,363	9.1
Started and completed as outpatient	11,718	5.2
Started as outpatient but completed as inpatient	32	0.01
Started and completed as a home visit*	24	0.01
Started as a home visit but completed as an inpatient*	2	0.009
Total	**224,932**	**100**

*Home visit data only from 2023

Data across 2020 to 2023 shows that for all babies screening is completed at a median of 2.45 days from date of birth to screen completion with an interquartile range 2.0 to 4.41 days). The vast majority of these babies will have a 'clear response' finding on the post-natal ward, prior to hospital discharge. These babies are discharged from the hearing screening programme as having a low risk of hearing loss.

For babies whose screen result is 'no clear response'; this is usually identified on the post-natal ward initially. These babies return in the following week for a repeat of the initial screen. From this repeat screen approximately 1.2% of all screened babies, about 700-800 per annum, ultimately remain as having 'no clear response'. The median time for screen completion for 'well' babies ultimately determined from the screening programme as having 'no clear response' is 9.5 days, interquartile range of 2 days to 16 days.

The time for screen completion for babies in neonatal intensive care units (NICU) is slightly longer. Over the time period of this report, the median time for screen completion for NICU babies with 'no clear response' was a median of 11.4 days, interquartile range of 4.4 to 33.5 days.

Yield, specificity, sensitivity and positive predictive value

The yield from the NUNHSP indicates that the number of cases of PCHL detected relative to the number of babies screened between 2020 and 2023 was 1 in 654.

The specificity of the NUNHSP using data from 2020 to 2023 is 98.99%; with a positive predictive value of 13.2% (PPV).

Table 18 shows specificity for the NUNHSP for 2020, 2021, 2022 and 2023.

Table 18:
NUNHSP specificity 2020-2023

Year	Screen completed	Screen not suspected	Screen positive	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	99.1
Total	**224,926**	**222,318**	**2,608**	**344**	**1.5**	**2,264**	**13.2**	**98.99**

Yield refers to the number of cases detected by the screening programme out of all babies that were screened.

Sensitivity refers to a screening method's ability to designate an individual with a condition as positive. A highly sensitive method, e.g. 100%, minimises the possibility of false negatives – e.g. missing a case. Sensitivity for newborn hearing screening cannot be reported as there are no large scale studies that have performed diagnostic testing on all newborns to identify the number of false negative instances; i.e. newborns that passed their hearing screening but were later in life found to have deafness.

Specificity is the ability of a screening method to correctly identify an individual as not having a condition.

Positive predictive value (PPV) is the probability that after receiving a positive screening result an individual will definitely have the condition – in this case hearing loss.

A **false positive** is a screening result that indicates that a person has a specific disease or condition, in this case hearing loss, when the person actually does not have hearing loss. A screening programme aims to have as low a number of false positives as possible to minimise anxiety for parents who have to bring their baby for further tests to be eventually reassured that their baby is fine. A low false positive rate is also important to maximise parent acceptability to participate in the screening programme.

A **false negative** is a screening result that indicates that a person is not at risk of a specific disease or condition, hearing loss in this case, when the person actually does have a hearing loss. A screening programme aims to have minimal amount of false negatives as these equate to undetected cases. It is recognised that screening is not 100% accurate but the true sensitivity of newborn hearing screening is not known, as per above.

Standards for the NUNHSP

This section outlines in more detail the five main key performance indicators (KPIs) and standards that are set by the NUNHSP to monitor the performance of the screening programme. There are a suite of metrics that the NUNHSP Governance Group review on a regular basis but these five are the main KPIs used to quality assure the programme.

Offered screening

Standard 1: ***>99% of eligible babies are offered screening***

This standard was met for all babies in 2020 (99.9%), 2021 (99.9%), 2022 (99.3%) and 2023 (99.9%). Even with the Covid pandemic impacting on service provision across 2020, 2021 and 2022 and the cyber-attack in 2021 the NUNHSP consistently performed above expectations with regard to ensuring the maximum amount of babies possible are offered newborn hearing screening.

	2020 (%)	2021 (%)	2022 (%)	2023 (%)
>99% of eligible babies are offered screening	99.9	99.9	99.3	99.9

Completion of screening

A core aspect of any screening programme is to ensure that the full screening pathway is completed in a timely manner so that any issues that arise can be referred onwards to the relevant clinical teams. To asses this the NUNHSP has a standard that >95% of all eligible babies are to have completed screening by 4 weeks.

Standard 2: *>95% of eligible babies to complete screening by 4 weeks*

In 2020 this metric reported at 95.24%. While it did meet the target, it is noted that the Covid-19 pandemic had an impact. In 2021 this metric was 99.01%, in 2022 it was 99.3% and for 2023 it was 99.6%.

	2020 (%)	2021 (%)	2022 (%)	2023 (%)
>95% of eligible babies to complete screening by 4 weeks	99.9	99.9	99.3	99.9

Referrals

The NUNHSP aims to identify babies that are at high risk of having some element of hearing loss. These babies are then referred onwards for appropriate assessment and diagnosis. The vast majority of newborn babies have no hearing issues but for those that do early intervention is vital.

The screening programme pathway for well babies allows for two otoacoustic emissions (OAE) steps as after birth the baby's ears may have some fluid and other issues that may make it difficult to obtain an accurate OAE result so a second one is offered for any babies that do not pass OAE1 (NICU babies only have one OAE).

Standard 3: ***≤30% of eligible well babies with No Clear Response on otoacoustic emissions 1 (OAE1)***

In 2020 there were 4.25% of eligible well babies with no clear response to OAE1. In 2021 and 2022 the performance was similar with 4.09% and 4.18% respectively having no clear response to OAE1.

The low rate is due to the good processes implemented by the screening programme and the screeners.

	2020 (%)	2021 (%)	2022 (%)	2023 (%)
≤30% of eligible well babies with No clear response on otoacoustic emissions 1 (OAE1)	4.25	4.09	4.18	3.84

Audiology (immediate referrals and targeted follow up)

Referrals to audiology for assessment and onward diagnostics, if required, are key elements of the NUNHSP pathway.

Standard 4: ***≤3% of all eligible babies referred for immediate diagnostics***

In 2020, 1.38% (n=583) of eligible babies were referred for immediate diagnostics. In 2021 the rate was 1.05% (n=630) and in 2022 it was 1.08% (n=583).

	2020 (%)	2021 (%)	2022 (%)	2023 (%)
≤3% of all eligible babies are referred for immediate diagnostics	1.38	1.05	1.08	1.13

Outcomes

While numbers identified with a PCHL and average age at PCHL confirmation are all collected by the NUNHSP, there are no programme performance targets associated with that data. The outcome data that have performance targets assigned are related to diagnosis by 6 months of age.

Confirmation of hearing loss

<u>Standard 5</u>: *≥80% of babies identified with a permanent childhood hearing loss (PCHL) by 6 months of age (Note: hearing loss >40dB either unilateral or bilateral, excludes mild hearing loss)*

In 2020, there were 83 babies that were identified as having a hearing loss by six months of age (93.26%).

In 2021 there were 82 babies that were identified as having a hearing loss by six months of age (97.62%).

In 2022 there were 89 babies that were identified as having a hearing loss by six months of age (96.74%).

In 2023 there were 76 babies that were identified as having a hearing loss by six months of age (96.2%).

	2020 (%)	2021 (%)	2022 (%)	2023 (%)
≥80% of babies identified with a permanent childhood hearing loss (PCHL) by 6 months of age	93.26	97.62	96.74	96.2

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Appendix 1:

Overview of the conditions currently screened for as part of the National Newborn Bloodspot Screening Programme

Phenylketonuria (PKU)

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. Around 1 in 4,200 babies born in Ireland has PKU.

Homocystinuria (HCU)

Homocystinuria (HCU) is an inherited condition caused by an altered gene that can cause a build up of amino acids that can lead to eye problems, impaired brain development and bone disorders. Around 1 in 59,300 babies are born in Ireland with HCU.

Classical galactosaemia (CGAL)

Classical galactosaemia (CGAL) is caused by the body not having enough of the enzyme that breaks down galactose, a sugar found in milk, and it can be like-threatening for babies, or lead to liver damage or sepsis. There are around 1 in 11,500 babies born with CGAL but CGAL is found to be more common in babies born to Irish Traveller parents.

Maple syrup urine disease (MSUD)

Maple syrup urine disease (MSUD) is an extremely rare but serious inherited disorder where babies cannot break down certain amino acids that can build up in the blood and urine which can be harmful causing brain damage, developmental delay and can be life threatening. Around 1 in 355,800 babies are born with MSUD in Ireland.

Congenital hypothyroidism (CHT)

Congenital hypothyroidism (CHT) is the most commonly diagnosed condition that is screened for by the NNBSP. Babies born with CHT do not make enough of a hormone called thyroxine. Thyroxine is made in the thyroid gland and babies who do not make enough thyroxine can have growth problems or disabilities. In Ireland around 1 in every 1,000 babies born has CHT. Most cases of CHT happen randomly and only a very small number of cases are inherited.

Cystic fibrosis (CF)

Cystic fibrosis (CF) is probably the most well-known of the conditions that are screened for. It 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. Around 1 in 2,200 babies born in Ireland has CF and about 1 in every 19 people has one copy of the altered CF gene and 1 copy of the unaltered gene. If both parents carry the altered gene they have a 1 in 4 chance of having a baby with CF.

Medium chain acyl-coenzyme A dehydrogenase deficiency (MCADD)

Medium chain acyl-coenzyme A dehydrogenase deficiency (MCADD) is an inherited condition where people 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. There are around 1 in 14,500 babies born in Ireland with MCADD.

Glutaric aciduria type 1 (GA1)

Glutaric aciduria type 1 (GA1) is an inherited condition caused by a missing or non-functioning enzyme which is 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. Around 1 in 232,400 babies born in Ireland have GA1.

Adenosine deaminase deficiency severe combined immunodeficiency (ADA-SCID)

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. Around 1 in 78,500 babies are born with ADA-SCID in Ireland.

For more details on the conditions screened for please refer to 'A Practical Guide to Newborn Bloodspot Screening 9th Edition (2022) available at: https://www.hse.ie/eng/health/child/newbornscreening/newbornbloodspotscreening/information-for-professionals/a-practical-guide-to-newborn-bloodspot-screening-in-ireland.pdf

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