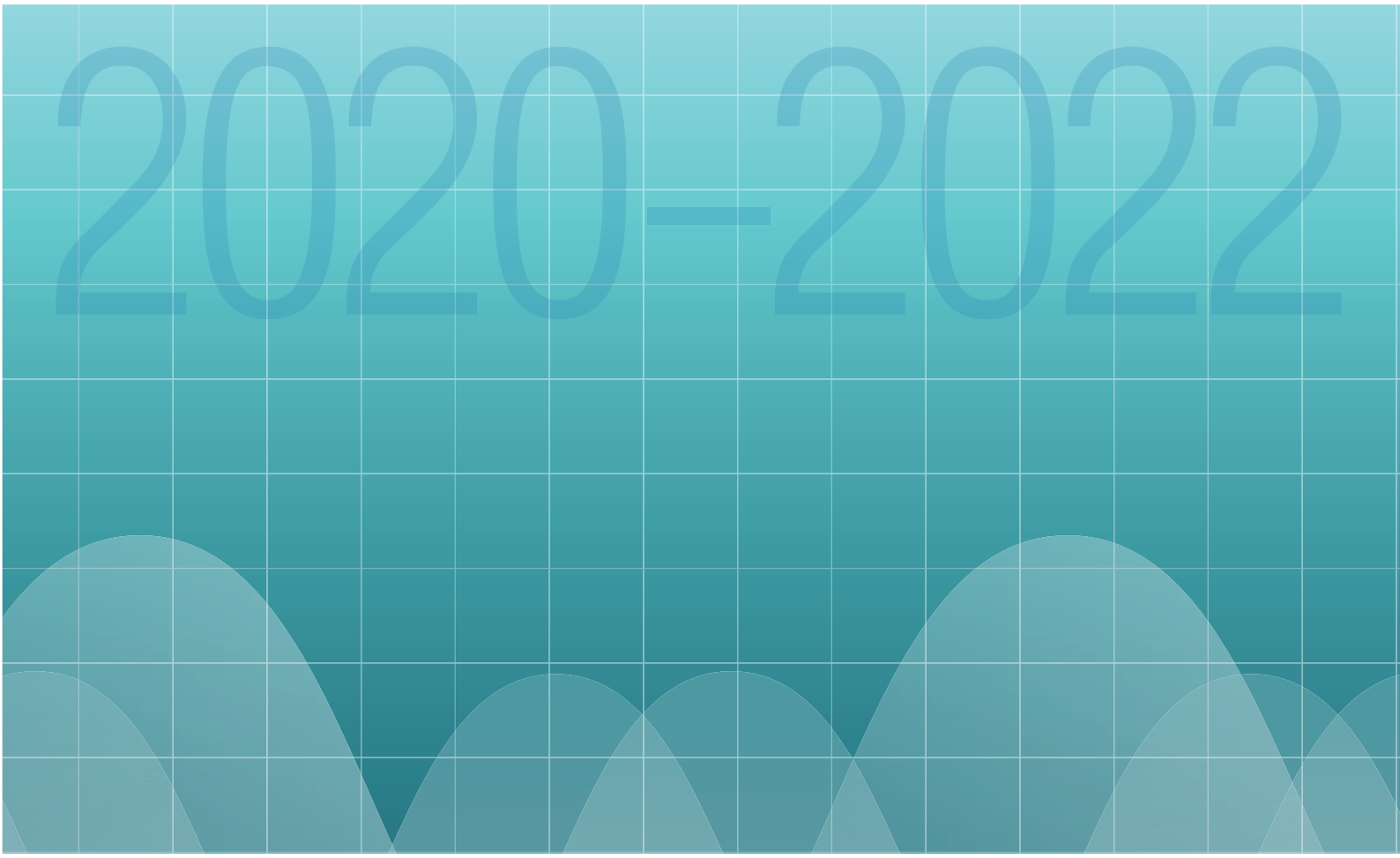
Report of the HSE National Children's Screening Programmes 2020–2022

National Newborn Bloodspot Screening Programme

National Universal Newborn Hearing Screening Programme



The National Healthy Childhood Programme

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Sláinte Leanaí Éireann
CHI
Children's Health Ireland

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 – Newborn Bloodspot Screening Programme and the Universal Newborn Hearing Screening Programme, for the years 2020, 2021 and 2022.

This report is available electronically from the National Newborn Screening Programme website: https://www.hse.ie/eng/health/child/newbornscreening or upon request from the project manager (contact child.screening@hse.ie)

Report of the HSE National Children's Screening Programmes 2020–22

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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

Dr Colm Henry
Chief Clinical Officer, HSE

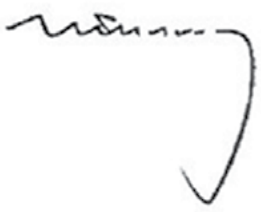
On behalf of the Health Service Executive and the National Healthy Childhood Programme I am pleased to present to you this three year report of the HSE's National Children's Screening Programmes for 2020 to 2022. In Ireland, the HSE National Children's Screening Services delivers two population level 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 are 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 Severe Combined Immunodeficiency (SCID) and we will progress implementation for this condition. We will continue to liaise with 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. This was very evident during the recent pandemic and cyber attack when bloodspot and hearing screening continued as usual. Their commitment to the programme is testament to their commitment to childrens' 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

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List of Abbreviations

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
NICU	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
NTGCHS	National Technical Group for Childhood Hearing Screening
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
UNHSP	Universal Newborn Hearing Screening Programme
UPI	Unique Perinatal Identifier

Introduction

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 if required. Screening is a pathway; it is not a diagnostic test.

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

- 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.

Shortly after birth, newborn babies, through their parent(s)/guardians, are offered the opportunity to be screened for rare but serious medical conditions through the national newborn bloodspot screening programme and for possible hearing loss through the national universal newborn hearing screening programme.

This report covers the HSE's Children's Screening service activity for three complete years; 2020, 2021 and 2022.

The report is in two sections with data provided for both screening programmes on case numbers, KPIs and programme performance.

Section 1: National Newborn Bloodspot Screening Programme

Section 2: National Universal Newborn Hearing Screening Programme

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. The NNBSP is available for all eligible babies within Ireland.

Each year, approximately 120 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'), 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 CoA 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 (2017–2022)	Worldwide Incidence*
Phenylketonuria (PKU)	1966	1:4,236	1:12,000
Homocystinuria (HCU)	1971	1:59,302	1:120,000
Classical Galactosaemia (CGAL)	1972	1:11,478	1:45,000
Maple Syrup Urine Disease (MSUD)	1972	1:355,813	1:225,000
Congenital Hypothyroidism (CHT)	1979	1:954	1:3,500
Cystic Fibrosis (CF)	2011	1:2,224	1:3,500
Glutaric Aciduria Type 1 (GA1)¥	2018	1:116,184	1:100,000
Medium Chain Acyl Co-A Dehydrogenase Deficiency (MCADD)¥	2018	1:14,523	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 four years of screening (2019-2022)
$Screening for ADA-SCID commenced in May 2022
**Burns et al (2021) Severe Combined Immunodeficiency (SCID) – The Irish Experience[1]

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

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

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 the years 2020, 2021 and 2022.

Covid 19 pandemic

In March 2020, the first lockdown of the Covid-19 pandemic occurred. The newborn bloodspot screening service was prioritised throughout this time due to its clinical urgency and the need to identify and treat babies who may have one of the rare conditions screened for. The clinical risk was considered to be too significant to pause or withdraw this screening service. The NNBSP is extremely grateful to colleagues across the 19 maternity hospitals/units and public health nursing services who, throughout the pandemic, continued to visit new parents/guardians, predominantly in their homes, support and counsel new families and take the newborn bloodspot screening samples. The pandemic also resulted in the redeployment of public health staff from the National Healthy Childhood Programme (NHCP) which reduced their capacity to support the NNBSP. The NNBSP is equally grateful to our colleagues within our National Newborn Bloodspot Screening Laboratory at Children's Health Ireland (CHI) at Temple Street who continued to provide the analysis, reporting and follow up of screening samples during the pandemic and re-oriented their laboratory service to ensure continuity of service provision.

HSE cyber-attack

On the 14th of May 2021, health services in Ireland suffered a major ransomware cyber-attack that caused health service Information Technology (IT) systems nationally to be shutdown. The shutdown of all HSE IT systems had a major impact across the health system and the NNBSL in CHI Temple Street was severely impacted. Access to the IT systems for running analytical laboratory processes and issuing results was unavailable. To safeguard the screening service, the NNBSL had to develop and implement manual processes immediately ensuring all samples received were logged, analysed and abnormal results reported. Screen positive results and repeat requests were phoned through to maternity hospitals/units or Public Health Nursing offices immediately and staff in these locations promptly actioned requests. Full electronic testing was reinstated on the 24th June 2021. The backlog of electronic reporting from 14th of May to 24th of June was completed by October 2021. The serious clinical risk that the cyber- attack posed to at risk newborns cannot be underestimated. The diligence and dedication of the staff of the NNBSL, and across the wider NNBSP must be commended for the actions taken to mitigate this serious clinical risk.

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: https://www2.hse.ie/conditions/heel-prick-screening/

HSE National Newborn Bloodspot Screening Programme Delivery

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

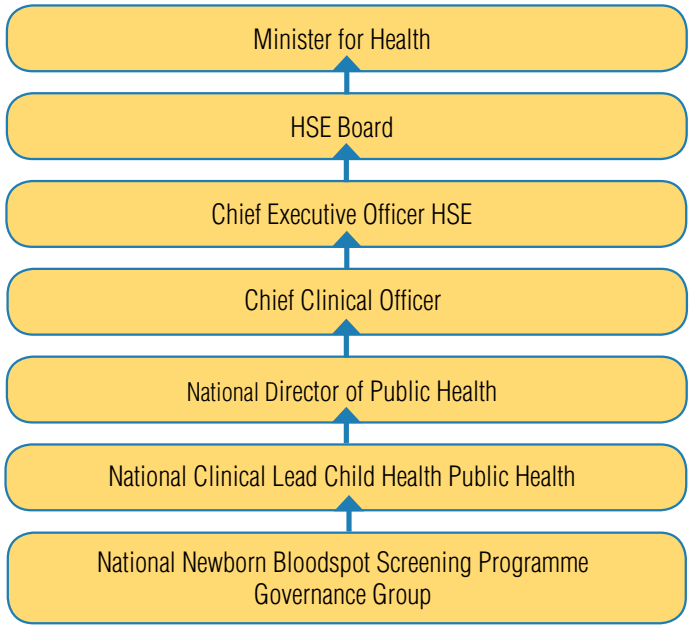
The operational and clinical governance of the NNBSP within the NHCP sits within the National Public Health function of the Office of the Chief Clinical Officer of the HSE.

Operationally, the collection of the 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



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 2020–2022

Metric	2020	2021	2022	Programme Target
Number of babies screened	57,016	60,985	54,784	
Number of samples analysed~	66,122	70,420	62,519	
% samples taken between 72-120 hours after birth	96.5%	96.0%	96.1%	95%
% samples received by NNBSL within 3 working days	99.2%	98.6%*	98.8%	100%
% of screening cards with Unique Perinatal Identifier (UPI) recorded	99.4%	99.5%	99.5%	99%
% 'screen positive' clinical referrals performed within 10 days of sample receipt (all conditions except Cystic Fibrosis)	100%	100%	100%	100%
% referrals sent to Cystic Fibrosis Centres by 4th week of life	93.8%$	91.9%¥	97.7%	95%

~ Babies can have more than one sample taken for clinical reasons; 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

$ 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

178 parent(s)/guardian(s) are known to have opted out of the newborn bloodspot screening programme in 2020, 2021 and 2022 (37, 60 and 81, respectively). This demonstrates that approximately 99.9% of parents opt to participate in the HSE NNBSP. Of note, 25% of those who opted out of the NNBSP in 2022 were parent(s)/guardian(s) of babies born outside of Ireland

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 2020–2022

	2020	2021	2022	2020–2022
Repeat Samples due to quality issues	2,172 (3.8%)	2,349 (3.85%)	2,755 (5.0%)	7,276 (4.2%)

Outcomes

The number of babies screened by the NNBSL in 2020 (57,016), 2021 (60,985) and 2022 (54,784) was 172,785.

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

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

Condition	Number of Confirmed Positive Patients (2020–2022 inclusive)	Rate per 100,000 (2020–2022 inclusive)
Congenital Hypothyroidism (CHT)	200	116
Cystic Fibrosis (CF)	77	45
Phenylketonuria (PKU)	42	24
Classical Galactosaemia (CGAL)	15	9
Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)	12	7
Homocystinuria (HCU)	4	2
Glutaric Aciduria Type 1 (GA1)	1	0.6
Maple Syrup Urine Disease (MSUD)	0	0
Total	**351**	**203**

Sensitivity and Specificity

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

Table 5: NNBSP Sensitivity and Specificity, 2020–2022¥

	Sensitivity (%)			Specificity (%)			PPV (%)
	2020	2021	2022	2020	2021	2022	2017–2022$
PKU	100	100	100	99.99	99.99	99.99	87.5
MSUD	*	*	*	99.99	99.99	99.99	5.0
HCU	100	100	100	99.94	99.94	99.99	3.02
CGAL	100	100	100	99.93	99.99	99.90	52.31
CHT	100	100	100	99.94	99.95	99.97	58.58
CF	100	100	100	99.87	99.9	99.88	30.08
MCADD	100	100	100	99.97	100	99.96	
GA1	*	*	100	99.97	99.99	99.99	

*Unable to calculate sensitivity, no true positive cases detected in these years

$ PPV data reviewed from 2017-2022 due to small numbers of cases detected in the years 2020–2022. PPV omitted for MCADD and GA1 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 has 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

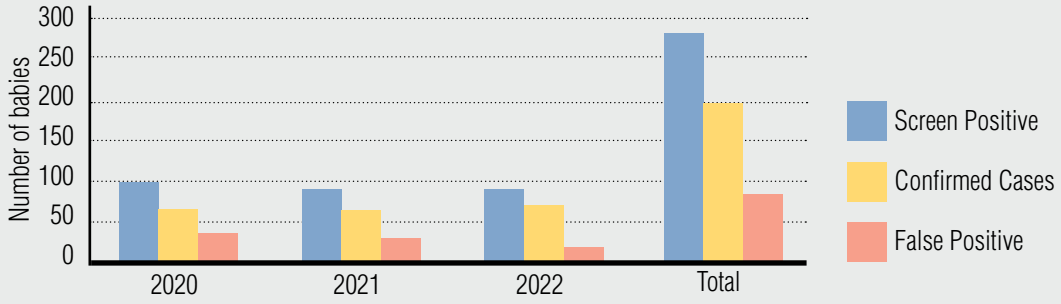
Congenital Hypothyroidism (CHT)

Congenital Hypothyroidism (CHT) is the most commonly diagnosed condition that is screened for by the NNBSP. Between 2020 and 2022 there were 200 confirmed cases of CHT. There were 84 babies who were ultimately determined to be false positive cases. In 2020 the NNBSP added the disorder type 'transient hypothyroidism' to more accurately diagnose 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 2020–2022

Congenital Hypothyroidism	2020	2021	2022	Total
Screen positive cases	100	91	94	285
Confirmed cases	64	62	75	201
Dysgenesis	16	8	6	30
Dyshormonogenesis	15	26	33	74
Unclassified	32	24	25	81
Transient Hypothyroidism	1	4	11	16
False positive cases	36	29	19	84

Figure 2: CHT data 2020–2022



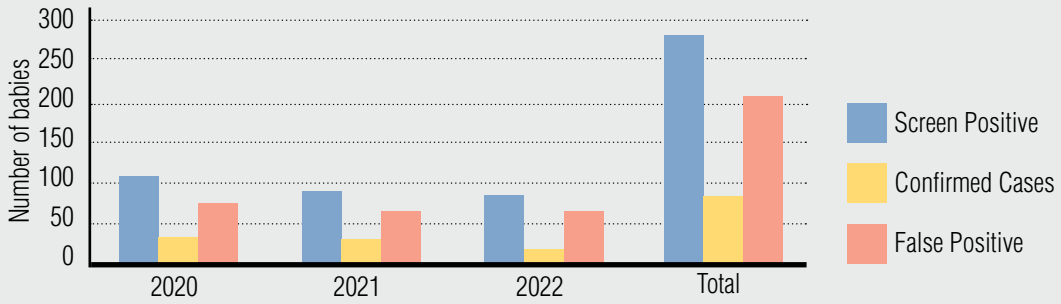
Cystic Fibrosis (CF)

Between 2020 and 2022, 2,174 samples were sent to the associated referral laboratory for CF genetic mutation analysis. This represented 1.26% of all initial newborn samples received between 2020 and 2022. Following mutation analysis, 276 babies were referred to a CF Centre for sweat testing; with 77 babies diagnosed with CF. There are 6 dedicated Paediatric CF centres across Ireland.

Table 7: Cystic Fibrosis 2020–2022

Cystic Fibrosis	2020	2021	2022	Total
No. referred for sweat test (screen positives)	102	89	85	276
No. with two CFTR mutations	22	24	20	66
No. with one CFTR mutation	80	65	65	210
Confirmed cases	29	27	21	77
False positive cases	73	62	64	199

Figure 3: CF data 2020–2022



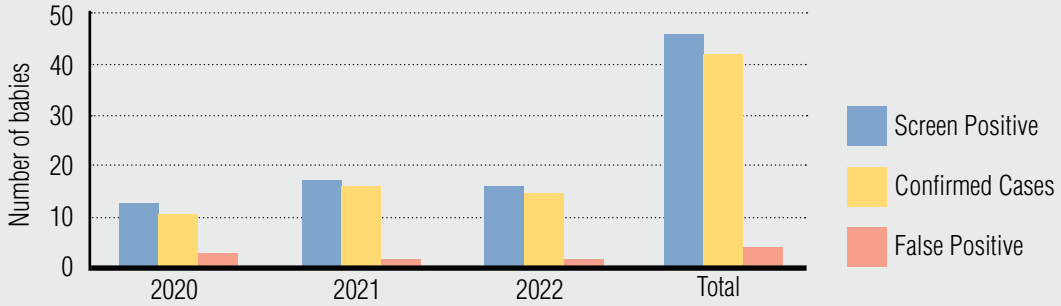
Phenylketonuria (PKU)

There were 42 babies diagnosed with PKU between 2020 and 2022. There were 4 babies who had false positive PKU screen.

Table 8: Phenylketonuria 2020–2022

Phenylketonuria	2020	2021	2022	Total
Screen positive cases	13	17	16	46
Confirmed cases	11	16	15	42
False positive cases	2	1	1	4

Figure 4: PKU cases 2020–2022



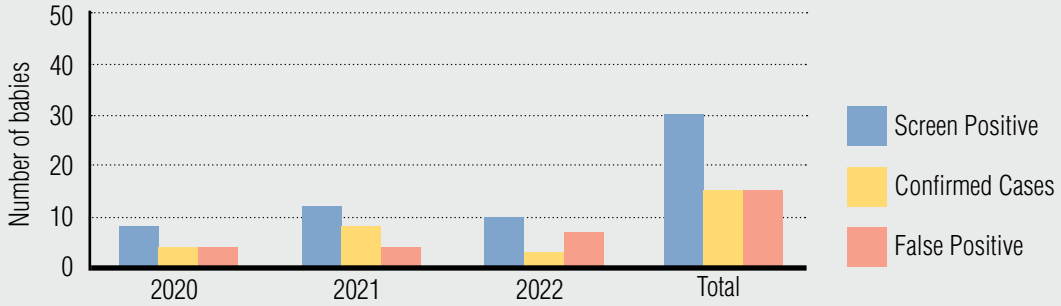
Classical Galactosaemia (CGAL)

There were 15 cases of CGAL diagnosed between 2020 and 2022. There were 15 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 2020–2022

Classical Galactosaemia	2020	2021	2022	Total
Screen positive cases	8	12	10	30
Confirmed cases	4	8	3	15
False positive cases	4	4	7	15

Figure 5: CGAL cases 2020–2022



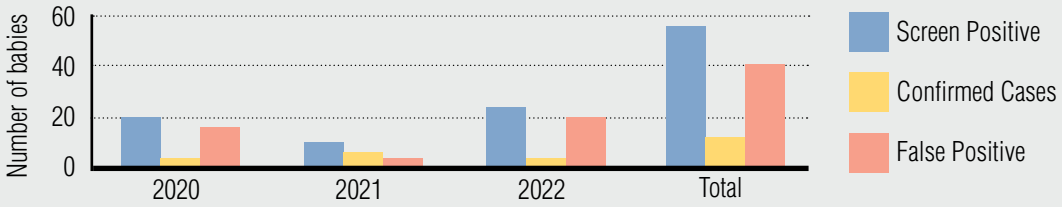
Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)

Screening for MCADD commenced in December 2018 and between 2020 and 2022 there were 12 confirmed cases of MCADD detected with 41 false positives.

Table 10: Medium Chain Acyl CoA Dehydrogenase Deficiency 2020–2022

Medium Chain Acyl CoA Dehydrogenase Deficiency	2020	2021	2022	Total
Screen Positive cases	19	10	24	53
Confirmed cases	2	7	3	12
False positive cases	17	3	21	41

Figure 6: MCADD cases 2020–2022



Howard et al (2023) published a paper on the first three years of screening for MCADD in Ireland which noted that the incidence of MCADD in Ireland is higher than previous estimates and that the biochemical and clinical phenotypes of the patients diagnosed with MCADD since the introduction of newborn screening were in keeping largely with expectation[1].

1. Howard et al (2023), *Medium Chain Acyl-CoA Dehydrogenase Deficiency: 3 years of newborn screening*. Ir Med J; March 2023; Vol 116; No 3; p743

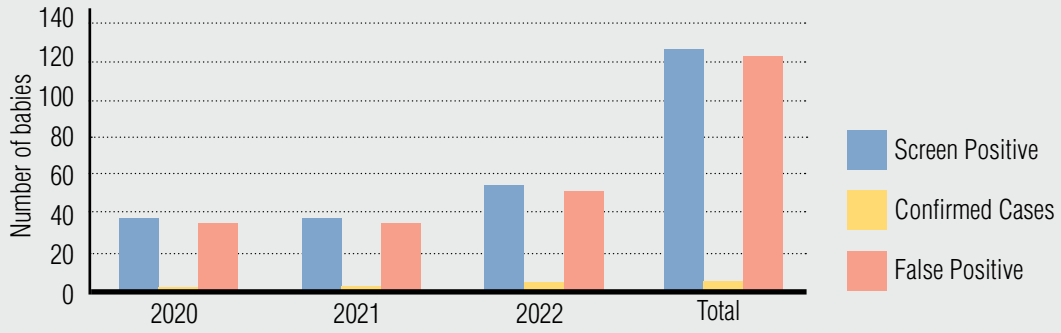
Homocystinuria (HCU)

There were 4 cases of HCU detected between 2020 and 2022 and 128 false positive cases. This is a rate of 74/100,000. 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 2020–2022

Homocystinuria	2020	2021	2022	Total
Screen positive cases	36	36	55	127
Confirmed cases	1	1	2	4
False positive cases	35	35	53	123

Figure 7: HCU cases 2020–2022



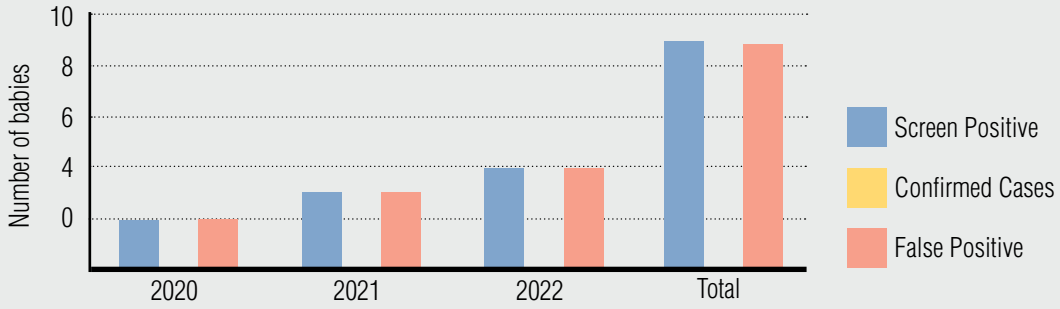
Maple Syrup Urine Disease (MSUD)

There were no cases of MSUD detected between 2020 and 2022 with 9 false positives.

Table 12: Maple Syrup Urine Disease 2020–2022

Maple Syrup Urine Disease	2020	2021	2022	Total
Screen positive cases	2	3	4	9
Confirmed cases	0	0	0	0
False positive cases	2	3	4	9

Figure 8: MSUD cases 2020–2022



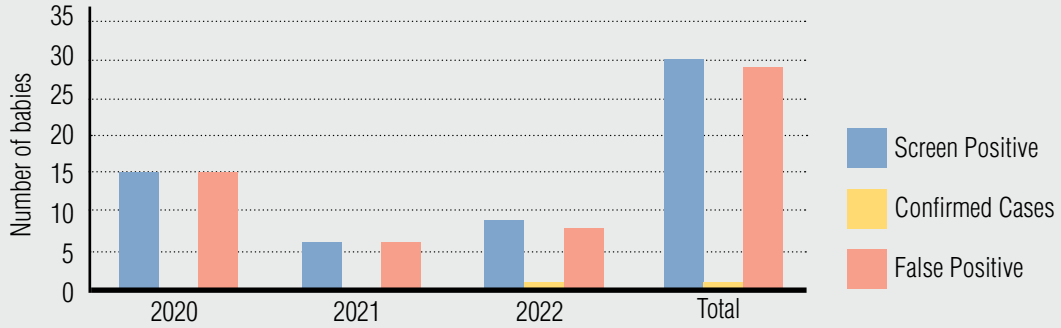
Glutaric Aciduria Type 1 (GA1)

There was 1 case of GA1 detected between 2020 and 2022 with 29 false positives. There was not as 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 and 8 in 2022.

Table 13: Glutaric Aciduria Type 1 (GA1) 2020–2022

Glutaric Aciduria Type 1	2020	2021	2022	Total
Screen positive cases	15	6	9	30
Confirmed cases	0	0	1	1
False positive cases	15	6	8	29

Figure 9: GA1 cases 2020–2022



ADA-SCID

Screening for ADA-SCID commenced in May 2022. There were no cases of ADA-SCID identified in 2022 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 are requested to immediately inform the NNBSP if a case (of a conditions screened for) is diagnosed, so a formal review can take place. The NNBSP has developed documentation to collect 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.

The NNBSP has not been informed of any false negative cases for babies screened between 2020 and 2022. 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 2020–2022, the NNBSP have completed investigations of three undetected cases for babies born outside of those years. In each of the cases, the investigation did not identify any error in the screening pathway as causative for 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.

State Claims Agency

The NNBSP requests reports from the State Claims Agency and the National Incident Management System (NIMS) to ensure awareness of incidents, and the nature of incidents, being reported by sample takers across the country as per HSE policy. This is used for programme quality assurance and improvement opportunities. It is also used to monitor potential risks of the programme.

Table 14: Number of bloodspot related incidents 2020–2022

Incident severity rating	2020	2021	2022
Negligible (category 3)	131	118	114
Minor (category 3)	1	1	0
Moderate (category 2)	3	3	1

Of note: Incidents are classified as below by the NIMS
Negligible: Near miss; no injury; injury not requiring first aid
Minor: injury or illness requiring first aid
Moderate: Injury requiring medical treatment

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 and 2022 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% and for 2022 was 96.1%. For this KPI to exceed the target during the Covid-19 pandemic is a testament to the staff working across the screening programme.

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% and in 2022 it was 98.8%. This is below the programme target of all samples to be in the lab within three working days and the NNBSP are actively reviewing this KPI.

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% 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% and in 2022 it was 99.5%, 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, ADA-SCID 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 programme across 2020, 2021 and 2022. 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. 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.

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 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 Governance Group met four times in both 2021 and 2022 with an additional specific meeting in 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.

A key project progressed by the NNBSP Governance Group in 2020 was the submission to the National Screening Advisory Committee (NSAC) of a proposal to add Adenosine Deaminase Deficiency Severe Combined Immunodeficiency (ADA-SCID) to the NNBSP. This was one of the first submissions the NSAC had to consider in their new role and therefore necessary work had been undertaken by the NNBSP rather than the NSAC as is the case for new conditions now. This submission was heard at their March 2020 meeting. The NSAC subsequently made a recommendation to add ADA-SCID to the screening programme and screening for ADA-SCID commenced in May 2022.

The NNBSP Governance Group has participated significantly in the work of the NSAC, and has been readily available to the NSAC across 2020–2022. The NNBSP Governance Group contributes to HIQA processes as they work to inform expansion of the NNBSP. The NNBSP Governance Group will continue to actively and keenly participate in these processes to support expansion to the NNBSP as recommended by NSAC and requested by the Minister for Health.

Section 2

National Universal Newborn Hearing Screening Programme

Introduction

The UNHSP is available for all eligible babies within Ireland and approximately 80–90 cases of permanent childhood hearing loss are diagnosed each year.

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 (UNHSP) 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. 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 HSE contracted NEC Software Solutions, previously called Northgate Public Services, to deliver the screening element of the UNHSP. 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 and 2022.

Covid-19 pandemic and HSE cyber-attack

In March 2020, the first lockdown of the Covid-19 pandemic occurred. Screening was suspended for 18 days but as the service was deemed critical it was re-commenced at this point. The standard screening period was extended from three to six months to manage the backlog of patients arising and community based screening locations were made available for outpatient appointments. The HSE Audiology diagnostic and habilitation services

continued with prioritisation of patients based upon risk factors and in line with Covid policies. Where parents did not wish to attend for the immediate diagnostic testing, they were offered a 9 month targeted follow up.

The impact of the cyber-attack on the newborn hearing screening programme was negligible as Smart 4 Hearing (S4H), the IT system used for the operational management of the screening programme, is housed external to HSE IT systems and as such was not adversely affected and the service continued as normal. Communication protocols were amended locally in order to obtain the necessary information required to manage screen referrals to Audiology. The diagnostic equipment used in Community Audiology was not impacted by the cyber attack as it could be used in a standalone mode without any connection to a HSE network. The uploading of the resulting backlog of required administrative data was managed as a local level.

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 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 (PCHL) are diagnosed each year through the UNHSP 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 (UNHSP). The UNHSP, 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 UNHSP. NEC Software Solutions provides the UNHSP IT management system, Smart 4 Hearing (S4H), which tracks and records all the data for the UNHSP. 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 National Technical Group for Children's Hearing Screening (NTGCHS) is responsible for the safety, effectiveness and quality of the UNHSP and its associated clinical pathways. The NTGCHS is the governance group that monitors 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 the National Clinical Lead for Audiology. The National Clinical Lead for Audiology then reports in to the National Clinical Lead for Child Health Public Health – see Figure 10 for the current governance structure.

Figure 10: Governance structure for the UNHSP

The national UNHSP 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 NTGCHS reviews the UNHSP performance against the 21 key performance indicators on a quarterly basis.

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 AccuScreen 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 UNHSP include babies born with congenital aural atresia and microtia. For the following babies, referral 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 suspected or confirmed bacterial meningitis prior to screening being offered
- 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 (2020–2022), 98.4% have a clear response with no follow up required. 1.2% of babies were referred to audiology as a result of having no clear response during screening – see Table 15.

Table 15: UNHSP Summary Data 2020–2022

2020–2022	No.	%
Clear Response – No Follow-up required	168,090	98.4
Clear Response – Targeted Follow-up required	673	0.4
Referred to Audiology (no clear response)	1,988	1.2
Total	**170,751**	**100**

Table 16: UNHSP Summary Statistics 2020–2022

Metric	2020	2021	2022	Programme Target
Number of babies registered	57,144	60,860	54,780	N/A
Number of babies eligible for screening	56,619	60,298	54,275	
Completion of Screening				
Number of eligible babies who started hearing screening	56,519 (99.95%)	60,218 (99.96%)	54,179 (99.93%)	>98%
Number of eligible babies completed screening by 4 weeks	53,766 (95.24%)	59,566 (99.01%)	53,760 (99.3%)	>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%)	≤30%
Number of eligible well babies with No Clear Response at Otoacoustic Emissions 2 (OAE2)	648 (1.27%)	757 (1.38%)	724 (1.46%)	≤6%
Number of eligible well baby referrals from AABR	514 (1.00%)	345 (0.63%)	343 (0.69%)	N/A
Number of eligible NICU baby referrals from OAE	572 (10.83%)	572 (11.12%)	556 (11.85%)	N/A
Number of eligible NICU baby referrals from AABR	265 (5.02%)	283 (5.5%)	240 (5.11%)	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%)	N/A
Number of eligible babies referred for immediate diagnostics	781 (1.38%)	630 (1.05%)	586 (1.08%)	≤3%
Number of eligible babies referred for targeted follow up	202 (0.36%)	238 (0.40%)	233 (0.43%)	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%)	≤80%

Across the period 2020–2022, 9.1% of babies (approximately 5,200 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, 5.8% (approximately 3,300 annually) both started and completed their screening as an outpatient – see Table 17.

Table 17: Location of screening 2020–2022

2020–2022	No.	%
Started and completed as Inpatient	145,339	85.1
Started as Inpatient but completed as Outpatient	15,563	9.1
Started and completed as Outpatient	9,832	5.8
Started as Outpatient but completed as Inpatient	17	0.01
Total	**170,751**	**100**

Data across 2020 to 2022 shows that for all babies screening is completed at a median of 2.5 days from date of birth to screen completion with an interquartile range of two days (2.4-4.4 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, IQR 2.5 days-16.5 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 12.5 days, IQR 4.6-36.5 days.

Yield, Specificity, Sensitivity and PPV

The yield from the UNHSP indicates that the number of cases of PCHL detected relative to the number of babies screened between 2020 and 2022 was 1 in 661.

The specificity of the UNHSP is 98.96%; with a positive predictive value of 12.7%.

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

Table 18: UNHSP Specificity 2020–2022

Year	Screen Completed	Screen Not Suspected	Screen Positive	PCHL	Prevalence	False Positives	PPV (%)	Specificity
2020	53,766	52,985	781	83	1.5	698	10.6	98.7
2021	59,566	58,936	630	82	1.4	548	13.0	99.08
2022	53,760	53,174	586	89	1.7	494	15.3	99.08
Total	**167,092**	**165,095**	**1,997**	**254**	**1.5**	**1,740**	**12.7**	**98.96**

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.

State Claims Agency

Reports by the State Claims Agency from the National Incident Management System (NIMS) identified that no UNHSP related incidents were created 2020–2022 inclusive.

Standards for the UNHSP

This section outlines in more detail the five main Key Performance Indicators (KPIs) and standards that are set by the UNHSP to monitor the performance of the screening programme. There are a suite of metrics that the UNHSP NTG 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%) and 2022 (99.3%). Even with the Covid pandemic impacting on service provision across 2020, 2021 and 2022 and the cyber-attack in 2021 the UNHSP consistently performed above expectations with regard to ensuring the maximum amount of babies possible are offered newborn hearing screening.

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 UNHSP 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% and in 2022 was 99.3%.

Referrals

The UNHSP 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% Number 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.

Audiology (immediate referrals and targeted follow up)

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

Standard 4: *≤3% of all eligible babies referred for immediate diagnostics*
In 2020, 1.38% (n=781) 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).

Outcomes

While numbers identified with a PCHL and average age at PCHL confirmation are all collected by the UNHSP, 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

Standard 5: *≥80% of babies identified with a Permanent Childhood Hearing Loss (PHCL) 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%).

The figure of 83 babies is a prevalence rate of 1.5 per 1,000 babies screened.

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

The prevalence rate in 2021 was 1.4 per 1,000 babies screened.

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

The prevalence rate in 2022 was 1.7 per 1,000 babies screened.

Impact of Covid-19

Following the period of suspension of UNHSP for 18 days the normal three month period for screening was extended to six months to enable the service to see those children who had not received screening and additional outpatient clinics were provided to complete the screening backlog.

Screening data was monitored on a weekly basis by site for babies born between the period 1st of February 2020 and the 14th of April 2020. During this period 11,020 babies were reported as having a clear response to screening and therefore no follow up was required. There were 151 babies that were recorded as having no clear response and were referred to audiology with a further 36 that had a clear response but it was deemed that targeted follow up was required.

The HSE Community Audiology services, in the context of Covid-19 pandemic, were classed as a non-essential service. Audiology staff were redeployed in relation to Covid-19, with minimal staff maintaining essential Community Audiology services. Guidance was provided for Audiology services during the pandemic, and face-to-face appointments continued for urgent cases which were risk assessed as essential since delay in diagnostic assessments could have negatively impacted on patient outcomes. Where parents did not wish to attend appointments for screening or audiology diagnostics they were placed on the wait list for targeted follow up. During this period there were 21 cases identified with hearing loss.

Table 19: Cases of hearing loss diagnosed for those born between 01/04/2020 and 14/04/2020

Protocol	NICU	Well Baby	Totals
Bilateral PHCL (including AN/AD)	4	9	13
Unilateral PCHL (including AN/AD)	3	3	6
Mild PHCL	0	2	2
Total	7	14	21

During the full period of the pandemic all services successfully managed challenges with Covid related staff absences at a local level.

Key Authors/Production Team

Paul Marsden, Project Manager, HSE Child Health Screening Programmes

Dr Abigail Collins, HSE National Clinical Lead Child Health Public Health

Gary Norman, HSE National Clinical Lead Audiology

Dr Mohamed Elsammak, Director, National Newborn Bloodspot Screening Laboratory, CHI Temple Street

Loretta O'Grady, Chief Medical Scientist National Newborn Bloodspot Screening Laboratory, CHI Temple Street

Catherine Harvey, Clinical Liaison Nurse National Newborn Bloodspot Screening Laboratory, CHI Temple Street

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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 CoA Dehydrogenase Deficiency (MCADD)
Medium Chain Acyl Co-A Dehydrogenase (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.

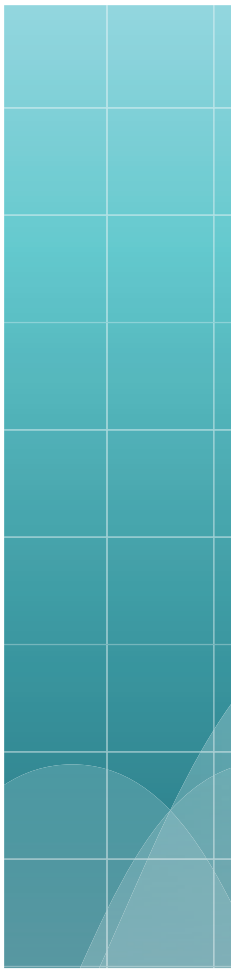
Glutaric Aciduria Type 1 (GA1)
Glutaric Aciduria Type 1 (GA1) is an inherited condition caused by a missing or non-functioning enzyme which is needed to break down protein in the diet. Without this enzyme harmful amino acids builds up in the body which can cause damage to the brain, movement difficulties, 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.

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

For more information and to provide any feedback about this report please contact: child.screening@hse.ie

Health Service Executive (2023).



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