



A Practical Guide to **Newborn Bloodspot Screening** **in Ireland**

National Newborn Bloodspot Screening Laboratory
Temple Street Children's University Hospital
Temple Street, Dublin D01 YC67

6th Edition – May 2016

Table of Contents

INTRODUCTION	5
Significant changes from 5 th Edition	6
Section 4 Responsibility for ensuring all babies are offered Newborn Screening.....	6
Section 6 Sample Collection.....	6
Section 7 Recording of Information and use of Eircodes.....	6
Section 8 Procedure for taking the routine blood sample	6
Section 9 Transport/Drying Boxes and pre-printed plastic envelopes	6
Section 10 Procedure for Reporting results.....	6
1 Contact details and NNBS Laboratory Opening Times	7
Telephone Numbers.....	7
Laboratory Opening Times.....	7
2 Abbreviations	8
3 Conditions Tested	9
3.1 Mode of Inheritance	9
3.2 Phenylketonuria (PKU).....	10
3.3 Maple Syrup Urine Disease (MSUD).....	10
3.4 Homocystinuria (HCU)	11
3.5 Classical Galactosaemia (CGal)	11
3.6 Cystic Fibrosis (CF).....	12
3.7 Congenital Hypothyroidism (CHT).....	13
4 Responsibility for ensuring all Babies are offered Newborn Screening	15
4.1 Responsibility of the Health Service Executive	15
4.2 Responsibility and role of the NNBS Screening Programme Steering Committee	16
4.3 Responsibility of Community Healthcare Organisation (CHO) Child Health Leads	16
4.4 Responsibility of Maternity Units and Maternity Hospitals	16
4.5 Responsibility of the Public Health Nursing Service in the CHO/LHOs	16
4.6 Responsibility of GPs, Self Employed Community Midwives and Private Midwifery Groups	17
4.7 Responsibility of Parents/Guardians.....	17
4.8 Responsibility of the National Newborn Bloodspot Screening Laboratory	17
5 Consent for Newborn Screening.....	18

5.1	Parental Consent.....	18
5.2	Right to Opt-out of Newborn Screening	19
5.3	Retention of the Newborn Bloodspot Screening Card (NBSC).....	19
5.4	Data Protection Legislation and Newborn Bloodspot Screening	20
5.4.1	Access to the Database	20
5.4.2	Return or disposal of Newborn Bloodspot Screening Card	20
6	Sample Collection	21
6.1	Baby's Feed at time of sample collection	21
6.1.1	Breast Fed Babies.....	21
6.1.2	Babies on Total Parenteral Nutrition (TPN) or IV fluids	21
6.2	Babies receiving Blood Transfusion	21
6.3	Premature Babies.....	21
6.4	High Risk Screening.....	22
6.4.1	Siblings of known cases.....	22
6.4.2	Baby born to Parent diagnosed with disorder on newborn screening panel	23
6.4.3	Family history of a metabolic disorder in extended family	23
6.4.4	Cousin Marriages within the Irish Traveller Community	23
6.5	Maternal Phenylketonuria	23
6.6	Babies born to Immigrant Parents or Refugees.....	23
6.6.1	Arriving <i>before</i> newborn bloodspot screening has been performed	23
6.6.2	Arriving <i>after</i> newborn bloodspot screening has been performed	24
6.7	Babies presenting with Meconium Ileus at birth.....	24
7	Recording of Information and use of Eircodes	25
7.1	Recording of Results of NBS Test	26
7.2	Early Discharges from Hospital	26
7.2.1	Newborn Bloodspot Screening to be performed by PHN.....	26
7.2.2	Newborn Bloodspot Screening to be performed by GP or independent midwife	27
7.3	Transfer from Maternity Unit/Hospital to Tertiary Referral Paediatric Hospital.....	27
8	Procedure for Taking the Routine Blood Sample.....	28
8.1	Equipment required	28
8.2	Completion of the Newborn Bloodspot Screening Card.....	28
8.3	Technique for Sample Collection	28
8.4	Sample collection from central line in sick babies.....	29

8.5	Packing and dispatching the Newborn Bloodspot Screening Cards	29
8.6	General Tips on Blood Collection	29
8.7	Quality of the Bloodspot Sample	30
8.8	Why Repeat Blood Samples may be requested	31
8.9	Problems with Sample Collection	31
8.10	Completion of Newborn Bloodspot Screening Card at Time of Discharge	31
9	Transport / Drying Boxes and pre-printed plastic envelopes	32
9.1	Procedure for Transporting Samples	32
9.2	Pre-printed registered Envelopes	32
9.3	Responsibility of Sender	33
10	Procedure for Reporting Results	34
10.1	Setting up Authorised Users for eReports	34
10.2	Benefits of eReporting	34
10.3	Requests for Repeat Sampling and responsibility for checking eReports	34
11	Procedures for Contacting Parent(s) with a suspected Positive Result	35
11.1	General procedure of contacting Parent(s)/legal Guardians with Query Positive Results	35
11.2	Query Positive Cases for PKU	35
11.3	Query Positive Cases for MSUD	36
11.4	Query Positive Cases for HCU	36
11.5	Query Positive Cases for Classical Galactosaemia	37
11.6	Query Positive Cases for Cystic Fibrosis	37
11.7	Query Positive Cases for Congenital Hypothyroidism	38
	Appendices	39
Appendix No.1	Opt-Out Form	40
Appendix No.2	Newborn Screening Card	41
Appendix No.3	Specimen Gate eReports™	42
Appendix No.4	Guide for Sample Takers	45
Appendix No.5	Community Healthcare Organisations and Local Health Offices	47

INTRODUCTION

The Newborn Screening Programme for phenylketonuria was started in Ireland on behalf of the Department of Health by Drs Seamus Cahalane and Doreen Murphy at the Children's University Hospital, Temple Street in February 1966; approximately four years after the first babies were screened in the States of Massachusetts and New York (USA). The Irish programme was one of the first national programmes in the world. A number of other conditions have since been added. Some of these have been discontinued and others will be added in the future. All babies born on or after 1st July 2011 have now been offered screening for Cystic Fibrosis.

The screening programme involves many healthcare professionals, each with their own expertise and responsibilities. The Programme is co-ordinated through the National Newborn Bloodspot Screening Laboratory at Temple Street Children's University Hospital on behalf of the Health Service Executive and the Directorate of Health and Wellbeing – Public Health and Child Health. The Screening laboratory is fully integrated into the Hospital's Department of Paediatric Laboratory Medicine thus ensuring rapid confirmatory testing of abnormal test results and the biochemical monitoring of those diagnosed. The NNBSL is accredited by the Irish National Accreditation Board (INAB).

The worldwide screening community is small and we in Ireland are very cognisant of the differences between regional and national programmes and the Irish Programme. The Irish programme has been tailored for the local population and the incidence of disorders occurring within the Irish population. The Newborn Bloodspot Screening Programme will continue to develop as new information and treatments become available for these rare inherited disorders. This expansion, whenever agreed, will be announced well in advance of implementation.

We would like to acknowledge the dedication and commitment that so many different healthcare professionals put into this programme for the benefit of the few babies born each year who are affected by one of these conditions.

This Practical Guide should be read in conjunction with the *Standard Operating Procedure for Maternity Hospital /Unit and Primary Care Services delivering National Newborn Bloodspot Screening Programme*. We would welcome any comments and feedback that individuals may have on this Edition. More information about the programme may be obtained at www.newbornscreening.ie and www.hse.ie/go/newbornscreening

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Significant changes from 5th Edition

Some of the more significant changes which have been incorporated into the 6th Edition are highlighted below. However, users are advised to read the entire section in order to familiarise themselves with the information or practice, thus ensuring that they are adhering to the correct procedures.

Section 4 Responsibility for ensuring all babies are offered Newborn Screening

HSE Governance Structure for newborn bloodspot screening

Responsibility for Community Healthcare Organisation (CHO) Child Health Leads

Section 6 Sample Collection

- 6.2 Intrauterine blood transfusion must be recorded on the NBSC.
- 6.3 Premature babies should have their NBS sample repeated at weekly intervals for a maximum of 4 weeks or until established on full feeds. For very premature infants born before 30 weeks gestation the sample should be repeated at discharge or at 36 weeks post conception age.
- 6.6 All infants of immigrant Parents/legal Guardians, up to one year of age, who have not been screened before arrival, should be screened for PKU, HCU, MSUD, Classical Galactosaemia and Congenital Hypothyroidism. Screening for CF by measuring blood IRT is not reliable for infants over six weeks of age.
- 6.7 Babies presenting with Meconium Ileus should have CF excluded with CFTR mutational analysis as the Bloodspot IRT may give a false negative result in this clinical setting.

Section 7 Recording of Information and use of Eircodes

The Eircode should be recorded in the appropriate boxes on the NBSC, as this will eventually be linked to the LHO to verify that the LHO recorded is correct.

Section 8 Procedure for taking the routine blood sample

- 8.3 Do not press or squeeze the bloodspot to 'force' it through the NBSC;
Ensure that the NBSC is air-dried before putting it into the envelope.

Section 9 Transport/Drying Boxes and pre-printed plastic envelopes

Pre-printed plastic envelopes may be purchased directly from An Post.

Section 10 Procedure for Reporting results

This section outlines the reporting of results by eReporting via the www and the mechanism for setting up authorised users.

1 Contact details and NNBS Laboratory Opening Times

Telephone Numbers

Temple Street Children's University Hospital	01 878 4200
National Newborn Bloodspot Screening Laboratory	
Enquiries:	01 878 4577
Email:	info@newbornscreening.ie
FAX	01 878 4596
Director (Prof Philip D Mayne)	01 878 4266
Chief Medical Scientist (Ms Loretta O'Grady)	01 878 4277
Clinical Liaison Officer (Ms Olivia Walsh)	01 892 1804
National Centre for Inherited Metabolic Disorders	
Enquiries	01 878 4317

Laboratory Opening Times

Monday to Friday	09.00 to 17.00
Analysis including Beutler tests and reporting of results	
Saturday Morning	09.00 to 12.00
Reporting of results and Beutler Assay – samples must be in the laboratory before 10.00	
Christmas, Easter and Bank holidays	Opening hours will be circulated well in advance and/or posted on the website.

2. Abbreviations

CHO	Community Healthcare Organisation
CHT	Congenital Hypothyroidism
CF	Cystic Fibrosis
CFTR	Cystic Fibrosis Transmembrane Conductance Regulator
CGal	Classical Galactosaemia
DNA	Deoxyribonucleic acid
GP	General Practitioner
HCRN	Health Care Record Number
HCU	Homocystinuria
HSE	Health Service Executive
IHI	Individual Health Identifier
IV	Intravenous (fluids)
INAB	Irish National Accreditation Board
KPI	Key Performance Indicators
IRT	Immunoreactive Trypsinogen
LHO	Local Health Office
MSUD	Maple Syrup Urine Disease
NBS	Newborn Screening
NBSC	Newborn Bloodspot Screening Card
NNBSL	National Newborn Bloodspot Screening Laboratory
NNBSP	National Newborn Bloodspot Screening Programme
OLCHC	Our Lady's Children's Hospital, Crumlin
PHN	Public Health Nurse
PKU	Phenylketonuria
RBC	Red Blood Cell
SECM	Self-employed community midwives
TPN	Total Parenteral Nutrition
TSCUH	Temple Street Children's University Hospital
TSH	Thyroid Stimulating Hormone
URL	Uniform Resource Locator

3 Conditions Tested

All conditions which form part of the Newborn Bloodspot Screening Programme, have been selected because they all have a relatively high incidence within the Irish population and they fulfil, in part or in full, the criteria which have been set out internationally for newborn screening. These include:

- the conditions screened are treatable;
- there is a test available which is easily applied to large population groups;
- there are few false positive and false negative results i.e. the test is reliable;
- the incidence of the conditions in the community is sufficiently high to warrant screening;
- the cost of screening makes the process cost-effective.

For all of the conditions, early diagnosis and treatment significantly improves the clinical outcome. Some of the conditions, for example, Classical Galactosaemia and Maple Syrup Urine Disease, benefit from the earliest detection possible. Unfortunately, as with all screening programmes, not all individuals with a condition will be detected. This is particularly true for Homocystinuria where about one in five babies may not be detected and for Cystic Fibrosis where milder variants of the condition may not be detected.

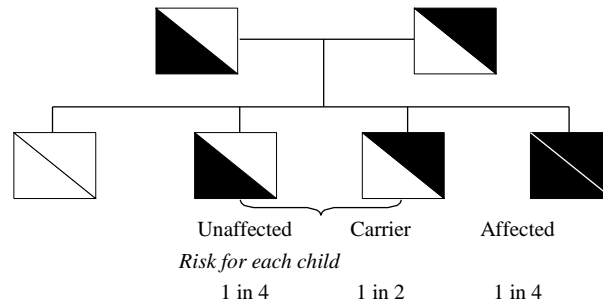
Table 1: Conditions included in the Irish Newborn Bloodspot Screening Programme

Condition	Date started	Irish Incidence	Worldwide Incidence
Phenylketonuria (PKU)	1966	1 : 4,500	1 : 12,000
Homocystinuria (HCU)	1971	1 : 69,400	1 : 120,000
Classical Galactosaemia (CGal)	1972	1 : 16,200	1 : 45,000
Maple Syrup Urine Disease (MSUD)	1972	1 : 155,200	1 : 225,000
Congenital Hypothyroidism (CHT)	1979	1 : 2,300	1 : 3,500
Cystic Fibrosis (CF)	2011	1 : 2,300	1 : 3,500

3.1 Mode of Inheritance

The majority of the conditions involve a defect in a metabolic process or pathway and are inherited as ‘autosomal’ conditions, not being dependent on the gender or sex of the individual. Each step in a metabolic pathway is governed by an enzyme, a protein produced by a set (two) of genes on a pair of chromosomes. Each parent transfers to their off-spring one set of genes so that the off-spring has a set from each parent. If one of these genes is defective (mutated), the metabolic pathway continues, albeit at a reduced rate. These individuals, known as carriers, do not have symptoms of the condition but carry the defective gene. If both parents are carriers, their offspring have a one in four chance of having the condition.

For autosomal recessively inherited conditions, both parents must be carriers of a defective gene and each of their off-spring has a one in four chance of having the condition and a one in two chance of being a carrier.



The clinical presentation and severity of the condition may vary between unrelated families. Different mutations may affect the same metabolic process differently; some individuals may present with a severe form of the condition and others with a very mild form.

Congenital hypothyroidism is slightly different in that for 90% of cases the condition just occurs, for reasons which are not fully understood; it is twice as common in girls as in boys. However, for one in 10 babies (10%) the condition is inherited as an autosomal recessive condition, as described above.

3.2 Phenylketonuria (PKU)

Phenylketonuria is an autosomal recessive condition involving the breakdown of the amino acid phenylalanine. Approximately one in every 4,500 babies born in Ireland have PKU or a milder form called hyperphenylalaninaemia. When diagnosed within the newborn period and started on treatment, these babies grow up healthy and well. However, without treatment this condition may cause intellectual or physical disability.

In the majority of cases, the condition is caused by a lack of the enzyme phenylalanine hydroxylase, which normally converts phenylalanine, one of the building blocks in protein, into tyrosine. In the absence of the enzyme, phenylalanine accumulates and high levels have a direct toxic effect on the brain.

Early treatment is very beneficial; it aims at giving a reduced intake of phenylalanine but a normal intake of all the other amino acids. This diet has to be continued for life. The screening test depends on detecting a high level of phenylalanine in the blood. If the test is carried out before about 72 hours after birth, there is a possibility that the level of phenylalanine in blood may not be sufficiently elevated for the condition to be detected.

The treatment for PKU has been one of the major successes in medicine since it was first introduced in the early 1950s. There is substantial evidence to show that the earlier treatment is started and the better the biochemical control throughout life, the better the outcome.

3.3 Maple Syrup Urine Disease (MSUD)

Maple Syrup Urine Disease is a life threatening condition if it is not detected and treated early. It too is an autosomal recessive condition caused by a defect in the metabolism of three amino acids, known as the branched chain amino acids because of their similar biochemical structure. Approximately one in every 155,200 babies born in Ireland may have this condition or about one baby born every two to three years. The disorder is so called because the urine may have an odour similar to that of maple syrup.

Screening was originally justified on the basis that chronic handicap and even premature death had occurred in a number of families where this condition had gone undetected. The branched-chain amino acids accumulate in blood following the establishment of feeding during the first few days of life, and may cause brain damage. Some babies may present with signs similar to that of infection.

A diet similar to that for PKU but with low levels of the branched-chain amino acids is started as soon as the diagnosis is made. Normal brain development and good health result from early treatment; life-long adherence to the diet is essential. Urgent medical intervention may be required during illness, which may be precipitated by infection or by 'stress'.

Some variant forms of Maple Syrup Urine Disease may not be detectable in the newborn period and may present clinically later in life.

3.4 Homocystinuria (HCU)

Homocystinuria results from the accumulation in blood of the amino acid methionine and one of its metabolic products homocysteine. Homocysteine accumulates due to a deficiency of the enzyme cystathionine β -synthase. Homocysteine is toxic to the lining of blood vessels and predisposes the individual to thrombus formation and blood clots and a number of other complications including osteoporosis (thinning of the bones) and dislocation of the lens of the eye. Again the treatment is similar to that for PKU. For those individuals who adhere to the diet, the risk of developing any of the complications is greatly reduced. Approximately one in every 69,400 babies born in Ireland may have the condition or one baby every year.

The screening programme detects high blood levels of methionine. This is one of the more difficult conditions to detect, as the blood methionine level may not be raised initially. The methionine concentration is low in many baby foods, particularly in breast milk. The screening programme may not detect approximately one in every five babies born with this condition. There are a variety of reasons why this may occur. These include:

- breast fed babies as there may be an inadequate intake of methionine in the feed to enable the blood methionine level to rise above the level for diagnosis;
- a milder vitamin B₆ responsive form of the condition.

Consequently, all babies or children who present clinically in later life with signs and symptoms suggestive of Homocystinuria, such as dislocation of lenses, osteoporosis or inappropriate tall stature should have the disorder excluded formally by measuring plasma levels of methionine and total homocysteine.

3.5 Classical Galactosaemia (CGal)

Classical Galactosaemia is an autosomal recessive condition caused by a deficiency of an enzyme galactose-1-phosphate uridyl transferase. This enzyme is important for the breakdown of galactose, one of the two sugars that make up lactose in human and cow's milk. Approximately one in every 16,200 babies born in Ireland each year may have this condition. However, it is particularly common among babies born to Irish Traveller parents in whom the incidence is approximately 1 in 450 births. Consequently, in the non-traveller Irish community the incidence

is about one in every 36,000 births.

If not detected and treated during infancy, the disorder may cause damage to the liver or there may be an increased risk of infection, which may occasionally be life threatening. As a result of the condition, galactose and its metabolite galactose-1-phosphate accumulate in blood; galactose-1-phosphate is extremely toxic. The baby may present with jaundice. There may be a bleeding disorder with a tendency to bleed spontaneously. The affected baby may also develop an *E coli* infection of the blood or septicaemia or present with cataracts in the eyes. Early detection and treatment with a lactose or galactose-free diet will prevent the early clinical complications of the disorder; some of the longer term complications, such as dyspraxia, ataxia or reduced fertility in women, may still occur in older children and adults despite dietary treatment.

Because the condition is more common in babies born to Traveller parents and to siblings of known cases, a special screening test, the Beutler test, is offered to all these babies at birth (Day 1 of life). They are advised to go on a galactose free feed (Soya-based) until the result of the test is available. This protects the baby should he/she have the condition. For those mothers wishing to breast feed, they should discuss this with their midwife – they can express their milk until the result of the Beutler test is available.

Clinicians should never depend upon the general population screening for the diagnosis of Classical Galactosaemia, but should query this condition in any baby who presents early with jaundice and other symptoms suggestive of Galactosaemia e.g. vomiting or floppiness, hypoglycaemia, conjugated hyperbilirubinaemia or abnormal clotting of unknown cause. There are a number of mild variants of the condition which the screening programme tries to avoid detecting as such individuals will never present with any symptoms.

3.6 Cystic Fibrosis (CF)

Ireland has one of the highest incidences of Cystic Fibrosis (CF) in the world with approximately one in every 2,300 babies being affected. CF is also an autosomal recessive condition with both parents carrying an abnormal CFTR gene. As a consequence of the condition, thick mucus secretions are produced by a number of organs including the lungs and the pancreas; it is this thick, mucous secretion which causes the problems. The thick secretions in the lungs may become infected, ultimately causing damage to the lungs; if the pancreas is involved this may cause, diabetes mellitus, digestive problems and malabsorption of important vitamins. Consequently babies with CF may present with failure to thrive and frequent chest infections.

Newborn screening means that babies with CF are identified earlier; they can be treated with a high energy diet to improve weight gain, and medicines and physiotherapy to improve lung function. Although a child with CF may still become ill, early treatment does improve their quality of life, significantly reducing the time that they have to spend in hospital; they live healthier and longer lives. Specific treatment is now available for a significant number of persons with CF depending on the specific mutation affecting the CFTR gene

The screening programme measures the blood level of immunoreactive trypsinogen (IRT). IRT is normally excreted by the pancreas into the intestinal tract but in individuals with CF, it is 'regurgitated' back into the blood due to the thick mucus secretions which block the pancreatic ducts. Levels of IRT may remain high in blood for about the first six weeks of life. If the blood

IRT level is high the sample will be referred for CFTR mutational analysis. This DNA test screens for the presence of 38 mutations on the original bloodspot.

- If two mutations are detected then the baby probably has CF and this will be confirmed by a Sweat Test.
- If one mutation is identified then a Sweat Test will be performed to determine whether the baby is a carrier of CF or has the condition. If the Sweat Test is positive, further DNA analysis will be undertaken to identify the second mutation.

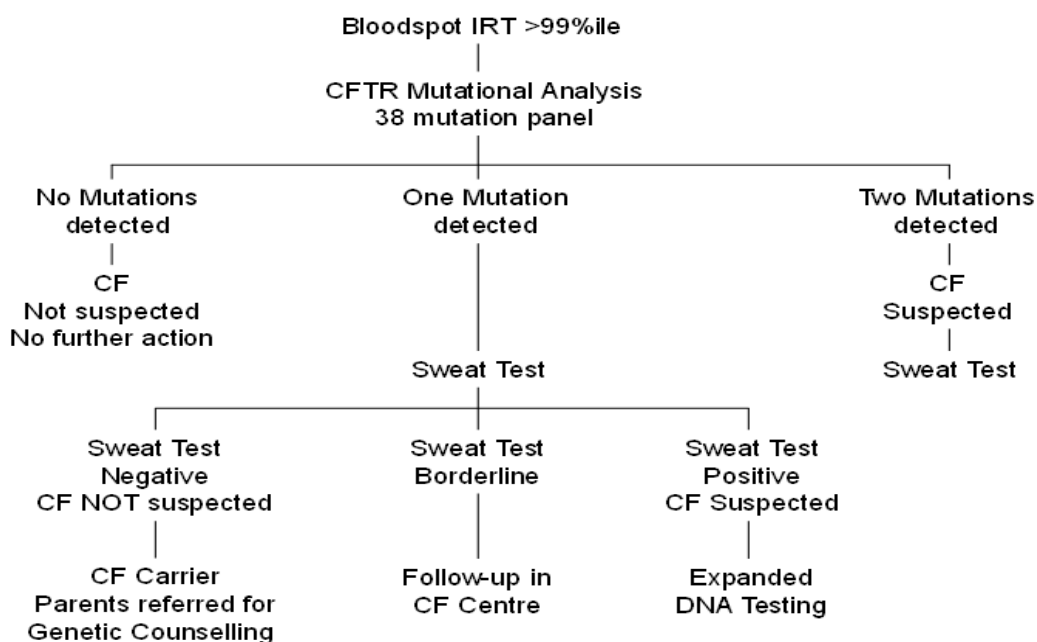


Figure 1. Algorithm for newborn screening for Cystic Fibrosis

Sweat Tests, which measure the chloride concentration in sweat, are usually performed before the fourth week of life in one of six designated HSE paediatric CF centres, based on the baby's address. The Sweat Test is considered the 'gold' standard for confirming the diagnosis of CF. As with some of the other conditions included in the screening programme, not all babies with CF will be detected by the newborn bloodspot screening programme. Milder variants of the condition may not be detected; some of these individuals may have a very benign clinical course which may not require treatment.

3.7 Congenital Hypothyroidism (CHT)

Unlike the other conditions, CHT is a congenital rather than an inherited condition, in the majority of cases. This is an endocrine condition, which results from failure of the thyroid gland to produce the hormone thyroxine. There are a number of different forms of the condition. Some babies may have a very small thyroid gland or no gland at all while others may not be able to make thyroxine. It is important to identify the cause; this can be done by performing a thyroid scan soon after the diagnosis has been made and usually before treatment has been started. Approximately one in every 3,500 babies born in Ireland may have the condition; early detection

allows for early treatment and the prevention of symptoms.

The diagnosis is made by measuring blood thyroid stimulating hormone (TSH), high levels of which are suggestive of the condition. However, TSH rises in blood immediately after birth and then falls to normal by about the second day of life. This is one of the reasons why the heel-prick sample should not be taken before 72 hours after birth, otherwise a false-positive result for congenital hypothyroidism could be produced.

The majority of babies with congenital hypothyroidism require thyroid hormone replacement. Some babies will be reviewed between two and three years of age at which time a small number may be able to discontinue treatment under medical supervision. Otherwise treatment is for life and the dose of thyroxine adjusted as the baby grows.

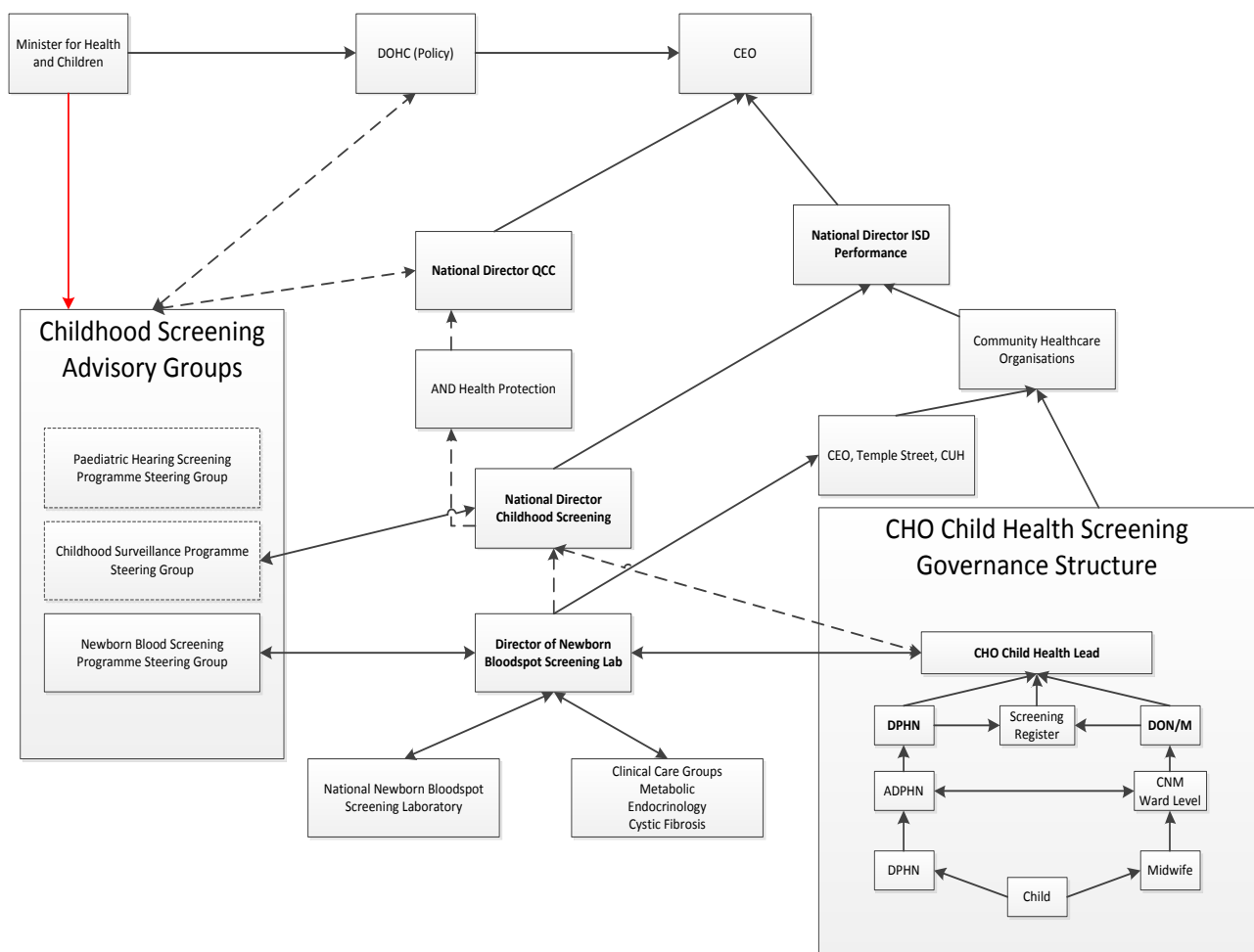
Compared to some of the other conditions, the frequency of false positive results for CHT is relatively high. Consequently the number of requests for repeat blood samples is also high. Possible reasons include:

- a transiently raised plasma TSH concentration which returns to normal in time. These babies may require a number of repeat samples to be collected. As hypothyroidism is more common in babies and children with Down syndrome, a disproportionate number of repeat samples may be requested from these babies as they too may have a transiently elevated plasma TSH level during the newborn period before developing hypothyroidism later;
- babies who may have had surgery before having the screening sample taken, may have a transiently elevated plasma TSH level. This may occur as some antiseptic skin preparations contain iodine which may be absorbed through the skin and cause transient hypothyroidism. This occurs more commonly in premature babies.

4 Responsibility for ensuring all Babies are offered Newborn Screening

Newborn bloodspot screening is an integral part of the health service provided to newborn babies. Screening involves the co-operation of many agencies involved in sample collection and transport, sample analysis and recording of results to referral and management of those babies diagnosed with one of the conditions. It is a complex process ensuring that all babies born in hospital or in the community are offered screening and followed-up until the screening process has been completed.

In 2011, the HSE put in place a Governance structure to manage the newborn bloodspot screening programme. This is outlined in the Figure 2 below.



4.1 Responsibility of the Health Service Executive

The ultimate responsibility for ensuring that all babies are offered screening in accordance with agreed protocols and procedures (*Standard Operating Procedure for Maternity Hospital/Units and Primary Care Services delivering the National Newborn Bloodspot Screening Programme 2016*) rests with the Health Service Executive (HSE) and the Assistant National Director of

Health and Wellbeing – Public Health and Child Health, who chairs the NNBSP Steering Committee. The Director of the NNBSP is responsible for the day to day coordination and management of the programme.

4.2 Responsibility and role of the NNBSP Screening Programme Steering Committee

The NNBSP Steering Committee is responsible for providing multidisciplinary advice to the HSE regarding technical and operational aspects of the programme and to the Director of Childhood Screening regarding strategic direction, policy and quality standards, funding objectives, legislation reviews and documentation relating to the programme, such as referral guidelines, manuals for practitioners, guidelines for storage, retention and use of residual bloodspots etc.. The Steering Committee is responsible for coordinating a quality assurance programme in partnership with health professionals and parents and monitors and facilitates improvements in the quality of screening processes and their outcomes for parents and babies. The Steering Committee is also responsible for supporting the development of information and training resources for health professionals and parents.

4.3 Responsibility of Community Healthcare Organisation (CHO) Child Health Leads

The CHO Child Health Leads are responsible for ensuring that all babies born, and all newborns residing in their CHO are offered screening and that structures for the timely checking and recording of the test results are in place. The CHO Child Health Leads are also responsible for reporting newborn screening uptake and coverage to the NNBSP Steering Committee on a quarterly basis.

4.4 Responsibility of Maternity Units and Maternity Hospitals

Directors of Nursing/Midwifery of maternity units/hospitals are responsible for ensuring that all babies born in hospital are offered screening. If the test is not performed in the maternity unit/hospital before discharge, hospital staff are responsible for ensuring that the baby is screened either by returning to the maternity unit/hospital or in the community, by informing public health nurses (PHNs). The maternity unit/hospital is responsible for informing the PHN Nursing Service of the discharge and the requirement for newborn bloodspot screening. The maternity unit/hospital must also inform the PHN Nursing Service if the newborn screening sample is not exempt from the packaging transport regulations due to infection either in the mother or baby.

4.5 Responsibility of the Public Health Nursing Service in the CHO/LHOs

Directors of Public Health Nursing are responsible for ensuring that the test is carried out in the Local Health Area (LHO) following notification from the maternity unit/hospital and that all babies residing in their designated area have been offered screening. The Directors of PH Nursing are also responsible for the timely recording of results in the NBS Register including details of those babies who require follow-up and further investigation.

4.6 Responsibility of GPs, Self Employed Community Midwives and Private Midwifery Groups

General Practitioners (GP), self-employed community midwives (SECMs) and Private Midwifery groups are responsible for performing the NNBS test in accordance to agreed HSE protocols and procedures and for dispatching the newborn bloodspot screening card (NBSC) to the NNBSL as soon as possible after collection in accordance with packaging transport regulations (See Section 9.0).

N.B. *The dispatching of the NBSC is the responsibility of the sample taker and should not be delegated to the parents.*

4.7 Responsibility of Parents/Guardians

Parent(s)/legal Guardians are responsible for their baby and their participation in the NNBS. In the case of Parent(s)/legal Guardians opting out of the NNBS and having been informed by healthcare practitioners of the potential consequences to their baby in so doing, the responsibility for any possible adverse consequences of their decision lies with the Parent(s)/legal Guardian. They must be requested to signify their decision to opt-out in writing by completing the Opt-out Form (Appendix No.1). A copy of the completed form must be retained by the Parent(s)/legal Guardians and a copy forwarded to the Directors of Midwifery/Nursing and Public Health Nursing, the NNBSL and the baby's GP. If Parent(s)/legal Guardians change their mind in the future, it is their responsibility to bring this 'change of mind' to the attention of the PHN or their GP.

In the case of babies born outside the jurisdiction to Parent(s)/legal Guardians who reside in the jurisdiction, the Parent(s)/legal Guardians are responsible for ensuring that the baby is screened. If they notify the PHN of the birth on their return to this jurisdiction, the PHN is responsible for ensuring that the newborn bloodspot screening test is carried out.

4.8 Responsibility of the National Newborn Bloodspot Screening Laboratory

The NNBSL is responsible for managing the programme on a day to day basis within the parameters and standards laid down by the Director of Childhood Screening and the NNBS Steering Committee. The NNBSL is responsible for overseeing the quality management system and reporting activities of the screening programme and ensuring that all screen positive cases are referred to the appropriate clinical care team within agreed standards and procedures and that a definitive diagnosis is ultimately made. The NNBSL liaises with the CHO Child Health Leads to ensure optimisation of the agreed screening standards and protocols.

The NNBSL is responsible for providing on-going training and education of all staff involved with the programme within agreed standards as required. The NNBSL also liaises with the individual clinical care groups to ensure that appropriate clinical audits are performed, thus optimising screening outcomes.

5 Consent for Newborn Screening

The newborn bloodspot screening or ‘heel-prick’ test should be offered to Parent(s)/legal Guardians of all babies who are born in Ireland or who enter the country before the test would have been performed in their country of origin. Parent(s)/legal Guardians must be given the appropriate information and adequate time to make a decision. HSE Parent Information leaflets are available and should be given to parents during the third trimester of pregnancy and again at the time of sample collection. Parent(s)/legal Guardians should also be given the top page of the Newborn Bloodspot Screening Card (NBSC) which contains some information about the programme. Parent(s)/legal Guardians do have the right to opt-out of newborn screening on behalf of their baby (See Section. 4.7 and 5.2).

5.1 Parental Consent

As Parent(s)/legal Guardians have a right to opt out, it is essential that they are informed fully of the benefits of the NNBSP. They should be told:

- about the nature of the conditions included in the screening programme;
- if these conditions are detected soon after birth, treatment can be started early and significant handicap and possibly premature death can be prevented. Treatment is essentially by modification of the diet under careful medical supervision or by drugs, as for congenital hypothyroidism or treatment for the prevention of complications as for CF.
- that a further sample may be required, either to check the first result or because of an equivocal result or a technical problem in the first analysis, such as a borderline result, an insufficient quantity of blood collected, a contaminated sample or assay interference;
- a further sample may be requested if the identity of the baby is unclear; this can occur if the NBSC is not filled in accurately – this is particularly important for twins, triplets etc. It is important to record on the NBSC the rank of the baby, twin 2 of 2, recorded as 2/2;
- that Parent(s)/legal Guardians of babies with a positive result will be contacted directly, usually by the maternity unit/hospital; clear instructions on how to proceed will be given.

Depending on the condition, Parent(s)/legal Guardians may be asked to bring their baby immediately to hospital, usually to Temple Street Children’s University Hospital, where the baby may be admitted for a period of time while treatment is commenced. If the baby is suspected of having CF, they will be contacted by a CF Nurse Specialist from one of the six HSE designated paediatric CF Centres and asked to bring their baby in the following day for further tests.

N.B. *Parent(s)/legal Guardians must be reassured that, with early detection and treatment of most of the conditions, their baby will develop essentially normally providing that they and then the child/adolescent adheres to the medical advice and treatment given.*

If Parent(s)/legal Guardians consent to their baby being screened, one parent must sign the NBS card, preferably the mother as under the *Guardianship of Baby’s Act, 1964* she has automatic parental responsibility for the newborn. They sign the NBS card to;

- verify that the details for her baby are correct;
- verify that she has received and read the Parent Information Leaflet;
- consent to her baby being screened.

Parent(s)/legal Guardians must be informed that the NBSC will be retained at a secure site for at least 10 years for the benefit of their child; thereafter, the NBSC will be disposed of.

The appropriate NBSC leaflet should be given to Parent(s)/legal Guardians and the Nurse Copy filed in the baby's clinical notes as a record that consent for newborn bloodspot screening has been given.

If the Parent(s)/legal Guardian has literacy difficulties they can be asked to make a mark on the form – this mark must be witnessed by an adult other than the sample taker.

5.2 Right to Opt-out of Newborn Screening

Parent(s)/legal Guardians do have the right to opt-out from the programme on behalf of their baby. However, they should be actively discouraged from so doing in the best interest of their baby's well-being; opting-out does conflict with the provisions of the Child Care Act (1991; 2001). In such cases, PHNs and midwives should consult with the GP or the consultant paediatrician. If they do decide to opt-out, it is essential that they are fully informed of the potential clinical consequences to their baby. The benefits of screening are explained in the Parent Information Leaflet and the consequences of not detecting a case summarized in the Opt-out Form.

Parent(s)/legal Guardians must sign the HSE Opt-out Form; this must be witnessed by an adult other than the potential sample taker and signed by all parties. A copy of the Opt-out Form must be given to the Parent(s)/legal Guardians and copies sent to the Directors of Public Health Nursing and Midwifery/Nursing and to the NNBSL. A copy should be sent to the baby's GP. Copies of the Opt-out Form are available at www.newbornscreening.ie and in Appendix No. 1.

Parent(s)/legal Guardians should be informed that they may change their mind in the future. However, it is their responsibility to make their change of mind known either to the PHN or to their GP.

5.3 Retention of the Newborn Bloodspot Screening Card (NBSC)

After screening, the NBSC is stored as part of the baby's health record for at least 10 years, after which it may be disposed of. After screening the sample may be used to:-

- check the results of the screening test or to perform other investigations recommended by the child's doctor for which Parent(s)/legal Guardians must give consent. In the event of sudden unexpected death, consent may be obtained from the Coroner;
- for quality control purposes and to help improve the screening programme as approved by the HSE. In such circumstances, all samples will be completely anonymised and it will not be possible to trace any result back to an individual baby.

Parent(s)/legal Guardians have the right to request that the NBSC be returned to them or to be disposed of earlier than ten years. However, it is the policy to retain all NBSCs for a minimum of six months.

5.4 Data Protection Legislation and Newborn Bloodspot Screening

Under a Service Level Agreement between the NNBSL Steering Committee and Temple Street Children's University Hospital (TSCUH) on behalf of the NNBSL, the NNBSL manages the programme on behalf of the Steering Committee on a day to day basis. In addition, TSCUH acts as the Data Controller under the terms of the Data Protection legislation on behalf of the NNBSL Steering Committee, retaining baby demographic details and a copy of the results as part of the baby's record and retains the NBSC for a minimum of ten years.

5.4.1 Access to the Database

Access to the database is password protected and restricted to scientific and secretarial staff working in the NNBSL and to a third party who supplied and maintains the software. Maternity Units/Hospitals and LHOs have limited password protected access to data and results on babies born in their Maternity Unit/Hospital or residing within their LHO, through an electronic report handling system (eReports™).

5.4.2 Return or disposal of Newborn Bloodspot Screening Card

Parent(s)/legal Guardians may request that the blood portion of the NBSC be returned or disposed of after six months. Such requests must be made in writing to the Director of the NNBSL, Temple Street Children's University Hospital, Dublin D01 YC67. They must provide proof of identity, to include a copy of their passport or driving license and a recent utility bill and a copy of the baby's birth certificate.

6 Sample Collection

Samples on all babies should be collected by heel-prick **after 72 Hours and before 120 Hours** from Birth and despatched immediately to the NNBSL, do not store and batch samples for despatch. If the sample is collected outside this time window it may adversely affect the result:

- *Before 72 hours* – it is essential that all babies should receive an adequate protein and galactose intake before the sample is taken, otherwise a false negative result may occur. Therefore, the sample is taken after 72 hours. Blood TSH levels, the test used to screen for congenital hypothyroidism, may be transiently elevated immediately after birth; consequently if the sample is taken too early, some babies may have a false positive test result for congenital hypothyroidism.
- *After 120 hours* – because the programme includes screening for Classical Galactosaemia and MSUD, it is essential that samples are not collected too long after birth, otherwise some babies may present clinically before the results of the screening test are available.

6.1 Baby's Feed at time of sample collection

Babies should be on oral or parenteral feeds for at least 24 hours before the heel-prick sample is collected.

6.1.1 Breast Fed Babies

Babies who are being solely breast fed should have the sample taken towards the end of the 72 to 120 hour time window. If protein intake is deemed to be suboptimal, a further sample should be taken on or about Day 10 of life. This is to ensure that protein intake has been adequate to reveal a positive test; breast milk contains less protein than formula feeds. This is particularly important when screening for Homocystinuria or some of the milder variants of the other conditions.

6.1.2 Babies on Total Parenteral Nutrition (TPN) or IV fluids

This should be clearly indicated on the NBSC. Because these babies may not be on any galactose/lactose containing feed and the test could give a false negative result for Classical Galactosaemia. A Beutler test will be performed to out rule CGal.

6.2 Babies receiving Blood Transfusion

If a Rbc transfusion has to be given before the 'routine' NBSC can be taken, a bloodspot sample should be collected for a Beutler Test; Rbc transfusion invalidates the test. The 'routine' sample should be collected between 72 and 120 hours after birth, and/or repeated 72 hours after the transfusion, depending on the time interval after the last transfusion.

If the foetus received an intrauterine transfusion, this should be clearly stated on the NBSC, as this may invalidate the result of the Beutler test for CGal.

6.3 Premature Babies

All premature babies, defined as less than 36 weeks gestational age, should have their NBS sample taken after 72 hours and before 120 hours from birth and repeated at weekly intervals for a maximum of 4 weeks or until the infant has been established on full feeds. For very premature infants born before 30 weeks gestation the sample should be repeated at discharge or at 36 weeks post conception age.

6.4 High Risk Screening

Details of any family history, relevant to the conditions included in the NNBSP must be stated clearly on the NBSC and the condition indicated.

6.4.1 Siblings of known cases

- *Siblings of known cases of Phenylketonuria (PKU)*

A newborn screening sample AND a lithium heparin blood sample should be taken between 72 and 120 hours following birth AND another NBS sample on Day 10 if the initial sample was reported as <PKU not suspected>. If the 72 hours following birth is due to fall on a Saturday, then we recommend that the liquid sample is taken and sent to the Metabolic Laboratory in Temple St on the Friday (in order to avoid parental anxiety over the weekend)

- *Siblings of known cases of Homocystinuria (HCU)*

A newborn screening sample should be taken between 72 and 120 hours following birth AND a lithium heparin blood sample taken at the same time for plasma methionine, total homocysteine and free homocysteine and be immediately deproteinised. A further lithium heparin blood sample should be taken on Day 10 of life and again deproteinised immediately.

N.B. For advice on sample deproteinisation, please contact the Metabolic Laboratory in Temple St.

- *Siblings of known cases of Maple Syrup Urine Disease (MSUD)*

A lithium heparin blood sample should be taken on Day 1 after the second feed and then daily until established on full feeds. Urine should be tested daily for ketones. A routine NBS sample should be taken between 72 and 120 hours to test for the other conditions. Staff at the NNBSL and the National Centre for Inherited Metabolic Disorders, Temple Street must be informed, prior to the delivery of the baby.

- *Siblings of known cases of Cystic Fibrosis (CF)*

Soon after birth, an EDTA blood sample should be sent directly by the Maternity Unit/hospital if parents request/consent, to the Department of Medical Genetics in OLCH, Crumlin, clearly stating the name and DOB of the affected sibling.

N.B. Blood IRT measurement is unsuitable to screen for CF in infants older than 6 weeks of age.

- *Siblings of known cases of Congenital Hypothyroidism (CHT)*

Thyroid function tests should be performed on Day 3 of life on a lithium heparin blood sample. The risks are dependent on the type of CHT. For those with dysmorphogenesis the risk is one in four, while those with a family history of thyroid agenesis or dysgenesis the risk is about one in 2000, depending on the gender of the baby; thyroid agenesis/dysgenesis CHT is more common in girls than in boys.

- *Siblings of known cases of Classical Galactosaemia and all babies born to Irish Traveller and Settled Traveller parents*

Both these groups are at particularly high risk of having Classical Galactosaemia. The incidence of Classical Galactosaemia among Irish Travellers is high at one in 450 births.

A NBS sample should be taken immediately after birth and *before any blood transfusion has been given*. A cord blood sample is not recommended. The sample should be sent to the NNBSL for a 'Beutler Test'. This test measures the enzyme activity in red blood cells and is NOT dependent of feeds. The NBSC should be clearly marked "FOR BEUTLER TEST".

The Beutler assay is not performed as an emergency on-call investigation. However, they are performed on Saturday mornings providing that the NBSC is received in the NNBSL by 10.00am. Special provision is made for Bank Holidays and long weekends; this information is circulated in advance. Two completed circles are required.

All *at-risk* babies should be fed with a lactose/galactose-free feeds (e.g. wysoy) until the result of the Beutler test is known. The normal NBS sample should be taken between 72 and 120 hours following birth to test for the other conditions.

6.4.2 Baby born to Parent diagnosed with disorder on newborn screening panel

Babies born to a parent with a condition included on the newborn screening panel are at high risk of having the condition, the risk being approximately twice the carrier incidence for the disorder within the Irish population. Irrespective of the condition, these babies should be screened as outlined above as if they were a sibling of a known case for the specific condition.

6.4.3 Family history of a metabolic disorder in extended family

If there is a history of a metabolic disorder within the extended family other than a sibling, this should be clearly stated on the NBSC to include the name of the disorder. It may not be necessary to take any additional samples; in such circumstances please contact the NNBSL for advice.

6.4.4 Cousin Marriages within the Irish Traveller Community

Some members of the Traveller community might wish to seek genetic counselling advice; this can be arranged through the Department of Medical Genetics, OLCH, Crumlin.

6.5 Maternal Phenylketonuria

Phenylalanine is actively transported across the placenta; the blood level in the foetus is about twice that of the mother. Therefore, women who themselves have PKU, should plan conception so that their condition is under optimal control at the time of conception. Regular and frequent monitoring of blood levels of phenylalanine and tyrosine are required throughout pregnancy in order to safe-guard the well-being of their foetus. Following the birth of the baby a liquid sample should be taken from the baby at the same time as the newborn screening sample and a repeat NSC sample on Day 10 of life.

6.6 Babies born to Immigrant Parents or Refugees

6.6.1 Arriving *before* newborn bloodspot screening has been performed

All babies and infants of immigrant Parents/Guardians, up to one year of age, who arrive in Ireland before any newborn screening test has been performed, should be screened for PKU, HCU, MSUD, Classical Galactosaemia and Congenital Hypothyroidism. Screening for CF by measuring blood IRT is not reliable for infants over six weeks of age.

6.6.2 Arriving *after* newborn bloodspot screening has been performed

The sample taker should be aware that the UK (including NI) newborn screening programme does not include screening for Classical Galactosaemia. If there is clinical concern, then the Beutler test should be requested on a dried bloodspot.

The decision to screen older children is a local decision, made by local clinicians and GPs and dependent on the family history and the country of origin of the parents. In such circumstances, please contact the NNBSL in advance on sample type.

6.7 Babies presenting with Meconium Ileus at birth

Meconium Ileus is a common complication in babies with CF, occurring in about 18% of all CF babies born in Ireland. These babies may have been transferred to a paediatric hospital before the newborn bloodspot screening test has been performed. Not all such infants present with a raised blood IRT from the NBSC. Therefore, CF should be considered in all babies who present with meconium ileus within the first days of life. An ETDA sample should be collected and sent directly by the Maternity unit/hospital to the Department of Medical Genetics, OLCH, Crumlin for CF mutation analysis, giving full clinical information.

7 Recording of Information and use of Eircodes

Until the HSE's Individual Health Identifier (IHI) is introduced nationally, the Unique Perinatal Identifier (UPI), issued by the Maternity Units/Hospitals must be used to track each baby through the NBS process. The UPI consists of the three digit HIPE code of the Maternity Unit/Hospital of the birth followed by the Healthcare Record Number (HCRN) of the baby. Babies born either at home or in a Maternity hospital outside Ireland will be issued with a UPI by the Director of Public Health Nursing in the area in which the birth is registered following notification of the birth.

The same UPI must be used on all samples sent to the NNBSL, even when the baby is transferred to a paediatric hospital. The HCRN of the paediatric hospital should also be included in the appropriate space on the NBSC.

N.B. *Eircode: the Eircode should now be recorded in the appropriate boxes on the NBSC. This will be linked to the LHO to verify that the LHO on the NBSC is correct.*

Table 2. HIPE Code Numbers for Maternity Units/Hospitals

HIPE Code	Maternity Hospital / Unit	HIPE Code	Maternity Hospital / Unit
201	Midland Regional Hospital, Portlaoise	724	Cork University Maternity Hospital incl Cork University Hospital
202	Midland Regional Hospital, Mullingar	726	Kerry General Hospital, Tralee
301	University Maternity Hospital, Limerick	800	Galway University Hospital, Galway
402	Cavan General Hospital	802	Mayo General Hospital
500	Letterkenny General Hospital	919	Portiuncula Hospital, Ballinasloe
501	Sligo General Hospital	922	Our Lady of Lourdes Hospital, Drogheda
600	University Hospital, Waterford	930	Coombe Women's Hospital, Dublin
601	St. Luke's General Hospital, Kilkenny	931	National Maternity Hospital, Holles St, Dublin
605	Wexford General Hospital	932	Rotunda Hospital, Dublin
607	South Tipperary General Hospital, Clonmel		

7.1 Recording of Results of NBS Test

The following steps must be adhered to, to ensure that adequate records are maintained in the hospital. This will help identify any baby who has not been screened before discharge:

- a) a person should be nominated by the hospital to take responsibility for checking records;
- b) details of each baby should be recorded in a single register;
- c) the register must indicate whether or not the test has been carried out;
- d) when the results of the test are returned from the NNBSL, the entry should be ticked in a different colour pen (if a manual procedure is used) or entered into the electronic register – therefore, any case where there is no report received from the NNBSL is noticeable at a glance and can be followed up;
- e) the person who checks the register to identify possible omissions should initial the entries to indicate that he/she has undertaken the exercise.

7.2 Early Discharges from Hospital

The nurse/midwife discharging a baby from hospital before the test has been carried out must ensure that the mother understands the following:

- the importance of the test;
- when the test should be done;
- that the test should be carried out either:
 - at the maternity unit/hospital, or
 - by a PHN, or
 - by a GP or independent midwife in the community.

If it is not appropriate to carry out the test before discharge from hospital, the nurse/midwife should ensure that the baby is screened either:

- by returning to the hospital
- or in the community. The nurse/midwife should notify the Director of Public Health Nursing that the baby has been discharged prior to the test being carried out to enable the PHN to give priority to these babies.

The Director of Public Health Nursing in the LHO in which the Parent(s)/legal Guardians live is responsible for ensuring that babies, discharged from hospital prior to having the test performed, have the test carried out.

7.2.1 Newborn Bloodspot Screening to be performed by PHN

If the test is to be performed by a PHN, the following procedures should be followed:

- (a) the ward sister should ensure that the appropriate Director of Public Health Nursing has been notified that the test is to be carried out. The Director should be provided with the full details of the baby, including the UPI. She should also be informed if either the baby or mother harbours an infection which could mean that the heel-prick sample was not exempt from the postal transport regulations (See Section. 9)
- (b) the Director of Public Health Nursing should:
 - keep a register of requests from hospital/s;
 - request the appropriate PHN to perform the test and to send the sample including the baby's UPI by registered post (and obtain a receipt of postage) to the NNBSL, Temple Street Children's University Hospital, Dublin D01 YC67.

- notify the maternity unit/hospital where the birth took place that the test has been carried out.
- (c) The NNBSL will send a copy of all results both to the Maternity Unit/Hospital and to the Director of Public Health Nursing.

7.2.2 Newborn Bloodspot Screening to be performed by GP or independent midwife

If the test is to be performed by a GP or an independent midwife, the following procedures should be followed:

- (a) the Director of Public Health Nursing must contact the GP or the independent midwife to ensure that the test will be performed and sample sent by registered post with the UPI (and obtain a receipt of postage) to the NNBS Laboratory;
 - it is the responsibility of the GP/independent midwife to arrange the transport of the sample; this should NOT be delegated to the parents.
- (b) the NNBSL will send a copy of results to the maternity unit/hospital, the Director of Public Health Nursing and to the GP / independent midwife;
- (c) the GP must contact the Director of Public Health Nursing in cases of non-attendance by the Parent(s)/legal Guardians.

In the case of Traveller babies, the parents may have moved out of the area before the test has been carried out. The Director of Public Health Nursing must be informed of the circumstances as early as possible so that alternative arrangements may be made.

7.3 Transfer from Maternity Unit/Hospital to Tertiary Referral Paediatric Hospital

The nurse/midwife responsible for the transfer of a baby from a maternity unit/hospital to a tertiary referral paediatric unit for continuing medical or surgical treatment before the NBS sample has been taken, MUST inform the receiving unit and give them the baby's UPI.

The paediatric unit must have written procedures for:

- performing the test between 72 and 120 hours after birth;
- sending the sample to the NNBSL;
- recording the results in the baby's medical records;
- informing the Maternity Unit/Hospital of the results of the tests.

8 Procedure for Taking the Routine Blood Sample

The blood sample should be taken between 72 hours and 120 hours after birth and when feeding has been established (See Section 6).

Ref.: Appendix No. 4: Guide for Sample Takers

8.1 Equipment required

- sterile lancet (metered tip no more than 2.5mm in depth);
- latex free gloves;
- cotton wool;
- newborn bloodspot screening card (NBSC);
- envelope (water resistant, tear-proof – Tyvek[®] or equivalent).

8.2 Completion of the Newborn Bloodspot Screening Card

Ensure that the NBSC has been completed in full and that all the information is correct, including the name of the baby; particular care should be taken to distinguish twin, triplets etc. The NBSC must be signed by a Parent(s)/legal Guardian before the sample is collected.

8.3 Technique for Sample Collection

- a) Ask the parents to keep the baby's feet warm prior to the visit by the PHN, by applying two sets of socks or placing a set of socks beneath the babygro.
- b) Read the instructions printed on the back of the NBSC carefully.
- c) Explain to the Parent(s)/legal Guardian the reason for the test and its importance to the baby's well-being.
- d) Obtain consent (See Section. 5) if not already obtained.
- e) Tear of the Top Information sheet and the <Parent Copy> from the NBSC and give them, along with the Parent Information leaflet, to the Parent(s)/legal Guardian; retain the <Nurse Copy> for filing in the child health record.
- f) Assemble the equipment and put on gloves. Do NOT touch the printed rings on the NBSC with gloves – Latex interferes with the Beutler Test and may cause a false positive result.
- g) Preferably take the sample from the baby while the mother cuddles the baby on her knee or on her shoulder; this not only assists you but also comforts the baby. It also allows the mother the opportunity ask questions about the test.
- h) Place a paper towel on the lap of the individual holding the baby.
- i) Ensure that the heel is warm; warm water, tested by the elbow of the sample taker, may be used.
- j) Rub the skin for 1-2 minutes to increase blood supply.
- k) Cleanse the heel thoroughly with warm (to touch) soapy water. Air dry the heel or wipe dry with cotton gauze. Avoid using Alcohol wipes to clean the skin as this may interfere with the formation of a blood drop.
- l) Encircle the heel with finger(s) and thumb and squeeze gently until the skin looks taut and suffused with blood.
- m) Press the lancet firmly against the side of the ball of the heel and trigger the sterile lancet.
- n) Hold the foot downwards and gently massage the heel to encourage blood flow.
- o) Touch the circle marked on the NBSC gently to the hanging drop of blood so that the blood soaks through from the back of the NBSC to the front:

- blood drops must soak through from the back to the front of the card, filling all circles completely.
 - check that the blood has soaked completely through the circle on the front as well as the rear of the card
 - do not press/squeeze the bloodspot to ‘force’ it through the NBSC
- p) Wipe away excess blood with cotton gauze. Press clean gauze firmly onto the wound until bleeding stops.
- It is not recommended that a plaster is used as this may be picked off by the baby and swallowed.
- q) Ensure that the NBSC is air-dried before putting it into the envelope. Do not use excessive heating as this may invalidate the test. Failure to fully dry the bloodspot before placing it in the envelope may result in serum rings, invalidation the sample and thus requiring a repeat sample to be taken.
- r) If a repeat sample is required for one test only, then two well-saturated bloodspots are sufficient.

8.4 Sample collection from central line in sick babies

On occasion the blood sample may be collected from a central line. Ensure that sufficient blood is discarded prior to placing drops of blood on the NBSC. Failure to do this may result in a dilute sample and give false negative results.

8.5 Packing and dispatching the Newborn Bloodspot Screening Cards

- a) Send the NBSC by registered post or by courier to the NNBSL using the yellow fluorescent address labels, to reach the laboratory as soon as possible after collection. Do NOT store or batch the completed NBSCs.
- b) If more than one NBSC is placed in a Tyvek[®] envelope, they should be placed at 180° to each other so that blood does not touch blood.
- c) The sender MUST indicate how many NBSCs have been placed in each envelope and preferably provide a separate list of the names of the babies whose specimens have been included in each envelope.
- d) Keep a record of all samples sent in each envelope; a sample checklist is available for downloading from the newbornscreening.ie website.
- e) Dispose of lancets as per local guidelines; never enclose the lancet in the envelope with the NBSC.

Please read Section. 9 on the *Transport of Pathological Specimens by Post* and make sure that the NBSC has been packed according to the current regulations.

8.6 General Tips on Blood Collection

- Alcohol wipes are not recommended for very premature babies – use soap and water.
- The use of a very thin smear of Vaseline over the puncture site might improve the quality of the blood drop by increasing the meniscus effect.
- The heel must be clean prior to skin puncture to prevent skin infection and avoid contamination of sample.
- Blood will flow more easily if the limb is lowered and warm.
- Alternative releasing of pressure for several seconds between squeezes should maintain blood flow.
- If blood flow stops, release pressure, lower the limb, wipe the puncture site firmly with

cotton wool, wait ten seconds and then squeeze gently again.

- Do not use plasters over the puncture site – babies may swallow them; if they are used the mother must be warned of the potential risk.

8.7 Quality of the Bloodspot Sample

Please ensure that there is an adequate amount of blood on the NBSC that completely fills each circle. An inadequate amount of blood or a poor quality dried bloodspot will require the procedure to be repeated causing potential trauma to the baby and inconvenience to the sample taker and may result in delay in making a diagnosis.

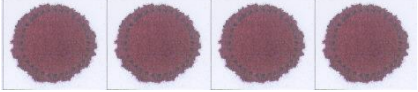



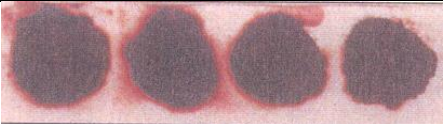

 <p>Valid sample</p>	<p>Sufficient amount of blood to soak through to completely fill all circles.</p>
Quality of Dried Bloodspot	Possible Causes/Reasons
 <p>Insufficient sample</p>	<ul style="list-style-type: none"> • Removing card before blood has completely filled circle or before blood has soaked through to second side • Touching filter paper before or after blood specimen collection with gloved or ungloved hands, hand lotion, etc.
 <p>Sample not dry before posting</p>	<ul style="list-style-type: none"> • Posting sample before drying for a minimum of two hours.
 <p>Sample appears supersaturated</p>	<ul style="list-style-type: none"> • Applying excess blood to card • Applying blood to both sides of card.
 <p>Sample appears diluted or contaminated</p>	<ul style="list-style-type: none"> • Squeezing of area surrounding the puncture site. • Allowing card to come in contact with gloved or ungloved hands or substances such as alcohol, formula, antiseptic solutions, water, milk, urine, hand lotion or powder, etc., either before or after blood specimen collection. • Exposing blood spots to direct heat.
 <p>Sample shows serum rings.</p>	<ul style="list-style-type: none"> • Not wiping alcohol from puncture site before making skin puncture. • Allowing card to come in contact with alcohol, hand lotion, water, milk, etc. • Squeezing area surrounding puncture site excessively. • Drying specimen incorrectly.

Figure 3: Quality of suitable and unsuitable Dried Bloodspots

8.8 Why Repeat Blood Samples may be requested

1. Insufficient blood on NBSC for all or some of the tests to be performed.
2. Unsatisfactory analysis:
 - test needs to be repeated (rechecked);
 - test result may be difficult to interpret because the specimen was contaminated or had deteriorated during transit.
3. Equivocal or borderline test result.
4. Baby too young when blood sample was collected. Sample was collected before 72 hours after birth.
5. Blood sample on the NBSC had not been dried properly before the NBSC was put into a plastic coated (Tyvek®) envelope, thus causing 'serum rings'.
6. There is a query about the identification of the baby or babies if multiple births.
7. The sample was taken on an expired card.
8. The card was delayed, greater than 14 days getting to the NNBSL and is too old for analysis.
9. The bloodspot portion of the NBSC and demographic portion were re-attached by sellotape but NBSC barcodes do not match up.
10. The name on the bloodspot portion does not match that on the demographic portion of the card; the identity of the baby may need clarification or a repeat sample.
11. Sample is unsuitable for analysis if taken within 72hrs of a RBC transfusion.

N.B. *It is the responsibility of the sample taker to tell the parents the correct reason why the repeat sample is required*

8.9 Problems with Sample Collection

1. Unlabelled or inadequately labelled specimens cannot be accepted for analysis.
2. Biohazards – babies whose mothers are known or suspected of being infected with HIV or Hepatitis B MUST have the screen performed. The NBSC MUST be identified as a Biohazard, but NOT the outer envelope and placed in a Biohazard envelope
3. Exchange transfusions may invalidate the screening test. Sample collection should be delayed for 72 hours.
 - All samples for the Beutler assay MUST be taken before a RBC transfusion is given; this must be clearly stated on the NBSC.

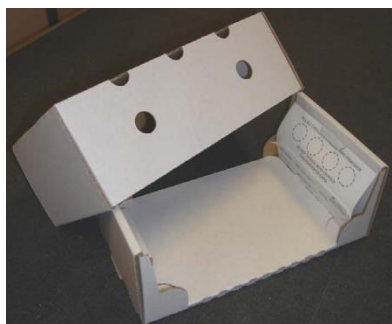
8.10 Completion of Newborn Bloodspot Screening Card at Time of Discharge

It is the practice of some Maternity Units/Hospitals to give the NBSC to the Parent(s)/legal Guardians either completed or blank at the time of discharge from hospital. It is the responsibility of the sample taker to check that ALL the details are correct, including the Day of Collection and Local Health Area. In addition, the name of the baby, both surname and first name, may have changed since discharge and these corrections MUST also be made.

9 Transport / Drying Boxes and pre-printed plastic envelopes

The NNBSL has designed drying/transport boxes in conjunction with Mega-Pak Ltd, to facilitate the transport of NBSCs from the baby's home to the PHN's car in a safe manner. Once the bloodspot has dried, the NBSC should be removed from the box and packaged according to the regulations. These boxes can contain two NBSCs.

The boxes are reusable. However, if they become contaminated with blood they should be disposed of either by incineration or through the accepted procedure for disposal of hazardous waste. These boxes can be ordered directly from Mega-Pak Ltd – the minimum order is 150 boxes, flat packed in batches of 50.



Mega-Pak Ltd (Irish Office)
16 Highfield Green
Swords, Co. Dublin

Tel: +353 (0) 1 840 2063
Fax: +353 (0) 1 840 2063
e-mail: megapakireland@eircom.net
Website: www.mega-pak.com

Figure 4: Transport/Drying Box

9.1 Procedure for Transporting Samples

The sender of NBSCs by registered post or by courier is responsible for ensuring that the packaging and transportation of the sample complies with current transport regulations regarding Health and Safety as laid down in the European Directive (ADR 2015) Packaging Regulations P650.

Dried bloodspots, collected by applying a drop of blood onto absorbent material are not subject to these Regulations but must be packaged appropriately. For this reason the NNBSL recommends that once the blood has dried, the NBSC should be inserted into a water-resistant, tear-proof Tyvek® envelope or equivalent. The yellow fluorescent address label should be fixed to the outer envelope.

9.2 Pre-printed registered Envelopes

Pre-printed plastic envelopes may be purchased directly from An Post by e-mailing Brian Beehan (brian.beehan@anpost.ie) with a copy to Noreen Hudson (noreen.hudson@anpost.ie)

Please send an email for the envelopes to both persons named above (in case one is on leave), stating the amount of envelopes required and quoting an HSE purchase order number. Envelopes retail @ €6.85 each and come in packs of 10 @ €68.50 per pack. A Pro Forma invoice, with the HSE purchase order number as reference, will then be returned to enable payment by EFT. Once the EFT payment has reached An Post's bank account the Philatelic Section will despatch the order.

9.3 Responsibility of Sender

If more than one NBSC is put in an envelope, they should be placed at 180° to each other (i.e. the bloodspots should not overlap and therefore not touch). The sender should state in writing how many NBS cards are in each envelope and include a list of the names of the babies on a separate page, a sample checklist is available to download from www.newbornscreening.ie

NBSCs, appropriately packaged, should be sent daily and not batched. It is the responsibility of the sender to dispatch the sample as soon as possible after collection, either by registered post or by courier. It is not appropriate to put the package into the post knowing that there may be a delay in it arriving at the NNBSL due to either a postal dispute (local or national) or over the Christmas period when the post is delayed. Alternative arrangements should be made by Maternity Hospitals/Units and LHOs to ensure NBSCs are despatched to NNBSL without delay. Parents should never be asked to post or deliver NBSCs to the NNBSL. This is the responsibility of the sample taker.

10 Procedure for Reporting Results

All Newborn Screening Reports are transmitted electronically via eReporting™ to the Maternity Unit/Hospital of birth and to the baby's LHO. Therefore, it is essential that the correct LHO is recorded on the NBSC, and the parent(s)/legal Guardians residence Eircode.

eReporting™ is a secure data protected means of transmitting Newborn Screening Reports over the internet. No paper copies of reports are now issued by the NNBSL. eReporting is password protected; only those with access to eReports can view results. It allows designated staff in the respective Maternity Unit/Hospital and LHOs to review the reports in pdf format.

The Reports are available to view on-line for 60 days from the date of issuing of the initial report; they are then archived in the NNBSL.

10.1 Setting up Authorised Users for eReports

Each Maternity Unit/Hospital and LHO nominates two Authorised Users; each will require access to the internet. Staff from the NNBSL will provide them with a User Name and User ID. They will also be provided with the URL link in order to access the eReporting portal.

10.2 Benefits of eReporting

- Authorised users can verify that a NBS sample has been received by the NNBSL before the results are available.
- Results are available as soon as they have been authorised by laboratory staff, usually within 2 to 3 days of receipt of the sample.
- As requests for repeat samples are made electronically, these repeat samples can be collected more quickly.
- A search facility can be used to find or link results; this includes babies or mother's name, UPI, Date of Birth or range of dates within the previous 60 days.
- Individual baby Newborn Bloodspot Reports can be printed locally and then filed in the Healthcare Record and/or a copy given to the Parent(s)/legal Guardian.

10.3 Requests for Repeat Sampling and responsibility for checking eReports

Authorised users must review eReports daily and notify colleagues if a repeat sample has been requested. The reason for requesting a repeat sample will be indicated on the individual report.

11 Procedures for Contacting Parent(s) with a suspected Positive Result

Parent(s)/legal Guardians of a baby with a suspected positive result are contacted directly by telephone by the Maternity Unit/Hospital. The procedure varies slightly depending on the condition under consideration and the result of the abnormal investigation. The NNBSL Steering Committee has set out Key Performance Indicators (KPIs) to ensure that babies detected with a condition are diagnosed and established on treatment within a specific time interval. The general procedure for contacting Parent(s)/legal Guardians is set out below with more specific details depending on the condition.

11.1 General procedure of contacting Parent(s)/legal Guardians with Query Positive Results

- The Clinical Liaison Officer in the NNBSL contacts a designated liaison nurse in the Maternity Unit/Hospital by telephone – the following information is given:
 - the baby's name, UPI, Date of Birth and address;
 - the disorder suspected and the result of the test.
- The designated liaison nurse in the Maternity Unit/Hospital will be asked to locate the baby and Parent(s)/legal Guardians and explain to them:
 - why the baby has to be referred to hospital;
 - what disorder has been suspected in their baby;
 - why a further NBS card or blood sample is required or thyroid scan if CHT suspected;
 - arrange for the baby to be brought directly to Temple Street Children's University Hospital or to the local Paediatric Unit as requested by the NNBSL. Parent(s)/legal Guardians should be advised that their baby might be kept in hospital for a few days, depending on the result of the repeat investigation. Therefore, they should bring a change of clothes for the baby and possibly for themselves.
- The designated liaison nurse will be given the mobile number of the Director of the NNBSL or his deputy. This number can be given to the parents, if they wish to contact him for more information before they arrive in the hospital.

At all times the designated liaison nurse must not instil any degree of anxiety when communicating with the Parent(s)/legal Guardians but must impart the information in a calm and professional manner, being fully informed of all the facts. These are rarely life-threatening conditions; therefore, Parent(s)/legal Guardians should be given plenty of time to make arrangements to travel to hospital.

Referral procedures for specific conditions are set out below

11.2 Query Positive Cases for PKU

The Clinical Liaison Officer in the NNBSL will book a bed for admission to Temple Street Children's University Hospital and liaise with the on-call metabolic team. The designated liaison nurse in the Maternity Unit/Hospital will be contacted and asked to:

- arrange for the baby to be transferred to Temple Street Children's University Hospital under the care of the on-call Metabolic Paediatrician;
- give the mobile number of the Director of the NNBSL or deputy to the Parent(s)/legal Guardians and invite them to contact him for more information, if they wish.

If the initial blood phenylalanine level is high then it is likely that the baby will be kept in hospital for a number of days until the level has fallen. During this time the baby will be started on lifelong treatment and the parent(s) will receive instruction on the monitoring and dietary management of their baby. This should be the only time that the baby will be admitted to hospital for the specific management of PKU.

Babies with a milder variant of the condition may be referred to the outpatient clinic. This information will be clearly given to the designated liaison nurse at the time of the initial contact.

11.3 Query Positive Cases for MSUD

The Director of the NNBSL will discuss the case with the on-call Metabolic Paediatrician. He will then, either contact the designated liaison nurse or the Paediatric Registrar in the Maternity Unit/Hospital directly, and arrange for the baby to be admitted, either to the local Special Care Baby Unit or directly to Temple Street Children's University Hospital. If the baby is to be admitted to:

- ***the local SCBU***
 - explain to the parents what disorder the baby is suspected of having;
 - on admission, examine the baby in detail and check the urine for the presence of ketones;
 - arrange for 1.3mL of whole blood collected into a lithium heparin tube for plasma branch chain amino acids to be sent immediately to the Metabolic Laboratory, Temple Street.

Contact the on-call Metabolic Paediatrician at Temple Street for further advice on management.

- ***Temple Street Children's University Hospital***
 - Parent(s)/legal Guardians must be informed that the baby will be admitted to hospital until the results of tests are known. If the test is positive, the baby will remain in hospital until the Metabolic Paediatricians are satisfied that the baby's condition is under control and that the parents will be able to cope at home.

11.4 Query Positive Cases for HCU

The designated liaison nurse will be asked to:

- locate the baby and Parent(s)/legal Guardian and explain to them why a blood sample is required and what disorder the baby is suspected of having;
- arrange for the baby to have a blood sample taken (1.3mL of whole blood collected into a lithium heparin tube) for methionine, total and free homocysteine and liver functions tests.
- The sample for free homocysteine must be de-proteinised immediately by the local laboratory staff; instructions will be given over the telephone.
 - if the plasma total and free homocysteine are raised and support the diagnosis, arrangements will be made for the Parent(s)/legal Guardians to attend the Metabolic Outpatient at Temple Street Children's University Hospital.
 - if the plasma methionine remains elevated but the total and free homocysteine are not raised and the baby is clinically well, further advice will be given to repeat the analysis. The baby will be followed up in the Metabolic Outpatient at Temple Street Children's University Hospital.

11.5 Query Positive Cases for Classical Galactosaemia

The Director or Clinical Liaison officer will contact the Paediatric Registrar in the Maternity Unit/Hospital to arrange immediate admission to the local Paediatric Unit. They will be asked to:

- locate the baby and Parent(s)/legal Guardians as a matter of urgency;
- explain to the parents the nature of the condition (Galactosaemia) and to bring their baby directly into the local paediatric unit where the baby will be admitted to hospital for further investigations.

On admission, all lactose and galactose containing feeds should be replaced by soya-based feeds. The baby should be examined and the following investigations performed:

- liver function tests;
- coagulation screen;
- blood cultures (to exclude, for example, *E coli* septicaemia);
- repeat NBSC.

As soon as the results of the investigations are available the local clinicians should either contact the Director of the NNBSL or the on-call Metabolic Paediatrician at Temple Street Children's University Hospital to discuss further action.

11.6 Query Positive Cases for Cystic Fibrosis

The results of the screening test will be available by approximately the third week of life:

- The Clinical Liaison Officer will contact the designated liaison nurse in the Maternity Unit/Hospital to obtain the Parent(s)/legal Guardians contact details and any relevant clinical information. She will then contact the CF Nurse Specialists in the appropriate HSE designated paediatric CF Centres, to give them the full contact details, relevant clinical information and the results of the mutational screen. These details will also be faxed.
- The CF Nurse Specialist will book a Sweat test appointment, and then contact the parent(s)/Legal Guardians to arrange for the baby to attend the nearest CF centre the following day.

On arrival, the parents will be fully informed as to what will happen; the baby will have a Sweat test, the results of which should be available by early afternoon on the same day. Depending on the results of the Sweat test, the parents will be informed that their baby has CF or is a carrier of the condition (and therefore unlikely to have CF). If the baby is considered to be a carrier, the parents will be referred for genetic counselling. See Fig 1.

HSE Designated Paediatric CF Centres

- Dublin North: Temple Street Children's University Hospital
- Dublin South:
 - Our Lady's Children's Hospital, Crumlin
 - National Children's Hospital (AMNCH), Tallaght
- Cork: Cork University Hospital
- Limerick: Mid-Western Regional Hospital, Limerick
- Galway: University College Hospital, Galway

11.7 Query Positive Cases for Congenital Hypothyroidism

The procedure may vary slightly depending on the age of the baby, the degree of elevation of the blood TSH level and referral hospital. However, clear instructions will be given. The designated liaison nurse will be asked to:

- locate the baby and parents;
- explain to the Parent(s)/legal Guardians what disorder the baby is suspected of having;
- arrange for the baby to attend the designated hospital to be examined, have blood tests performed and a technetium thyroid scan, following which the baby will usually be started on thyroid hormone replacement.

If the blood TSH level is confirmed as being very high and if the baby is approaching ten days of age thyroid hormone replacement may be started before they attend the hospital, in which case a thyroid ultrasound examination may be performed instead of a technetium scan.

Appendices

Appendix No. 1: Opt-Out Form

Appendix No. 2: Scan of Newborn Bloodspot Screening card

Appendix No. 3: e-Reporting user information

Appendix No. 4: Guide for Sample Takers

Appendix No. 5: Community Healthcare Organisations and LHOs

Appendix No.1 Opt-Out Form



NATIONAL NEWBORN BLOODSPOT SCREENING PROGRAMME

OPT- OUT FORM

Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

Baby's Surname <input style="width: 100%;" type="text"/>	Baby's First Name <input style="width: 100%;" type="text"/>
Date of Birth <input style="width: 100%;" type="text"/>	Baby's Unique Identifier <input style="width: 100%;" type="text"/>
Hospital/Place of Birth <input style="width: 100%;" type="text"/>	
Local Health Office <input style="width: 100%;" type="text"/>	
Baby's Address <input style="width: 100%;" type="text"/> <input style="width: 100%;" type="text"/>	
Mother's Surname <input style="width: 100%;" type="text"/>	
Mother's First Name <input style="width: 100%;" type="text"/>	

- I _____ being the parent/guardian of Baby _____, do not consent to allow the Newborn Bloodspot Screening Test (Heel-prick) to be carried out on my baby.
- I have read the Information leaflet on Newborn Bloodspot Screening and the test has also been explained to me.
- I fully understand the importance of the decision that I am taking by not allowing my baby to be tested.
- I understand that not detecting or treating one of the conditions, should baby have one, may result in severe intellectual or physical disability which could require long term care or result in premature death.

Signed (Parent/Guardian): _____

Full Name (PRINT): _____ Date: _____
(BLOCK CAPITALS)

Signed in the presence of (Midwife/PHN): _____ Date: _____

Position/Job Title: _____ Local Health Office: _____

OFFICIAL USE ONLY

6 copies of the completed form, signed by parent/guardian and midwife/PHN should be made. A copy to be given to the parent/guardian and a copy kept by the Midwife/PHN.

Copies are to be posted to the following:-

- The Director of Nursing/Midwifery • Director of Public Health Nursing • National Newborn Bloodspot Screening Laboratory
- the Baby's General Practitioner

	Director of Nursing/Midwifery	Director of Public Health Nursing	National Newborn Bloodspot Screening Laboratory	General Practitioner
Name:				
Address:			Children's University Hospital, Temple Street, Dublin 1	
Date Sent:				

Signed: (MIDWIFE/PHN) _____ Date: _____

Name: (BLOCK CAPITALS) _____

NNBSP April 2011

Parents have the right to opt-out from the programme on behalf of their baby and must sign this HSE Opt-out Form; this must be witnessed and signed by all parties.

The Opt-out Form can be downloaded from:

www.newbornscreening.ie or www.hse.ie/go/newbornscreening

[illegible]

Appendix No.3 Specimen Gate eReports™

Specimen Gate eReports™ allows authorised users with appropriate access rights in Hospitals and Public Health to view NBS reports via the internet and to view ‘pending’ samples that have been received but not yet reported. Each centre has two accounts only, one named and one generic. User names and passwords are only assigned to authorised users specific to their organisation / location following completion of a ‘User Authorisation Form’, available from NNBSL.

To access e-reports: In your internet browser type or copy the following URL and save to favorites. <http://sgateweblive.healthirl.net/ereports> or <http://10.0.37.81/ereports>

Each user, logins using their own ‘user name’ and ‘password’ assigned by the NNBSL, *see below*

National Newborn Bloodspot Screening Results

National Newborn Bloodspot Screening Laboratory: Ph: 01 878 4277

[Click here to add this page to your favorites](#)

Username :	<input type="text"/>
Password :	<input type="password"/>
<input type="button" value="Login"/>	<input type="button" value="Reset"/>

From here it is possible to search for a specific sample or download list or single reports.

- . Home
- . Specimen Search
- . Download Reports
- . Messages
- . Administration
- . Logout

Welcome to eReports

You have [0 unread message\(s\)](#).

Specimen Search

[Click for Help](#)

Surname:	<input type="text"/>	Mother's surname:	<input type="text"/>
DOB from (DD/MM/YYYY):	<input type="text"/>	DOB to (DD/MM/YYYY):	<input type="text"/>
UPI:	<input type="text"/>	Date of Collection (DD/MM/YYYY):	<input type="text"/>
<input type="button" value="Search"/>	<input type="button" value="Clear"/>		

Download Reports

[Click for Help](#)

Submitter	Reports
<input type="checkbox"/> RH (42)	20130522141146 ListReport 20130522122520 ListReport Show All Reports
1	

To Download Reports

- Select '**Download Reports**' from menu on left hand side – the most recent reports (list and single) for your location made available by the NNBSL will be visible to each user in pdf format.
- In this view, select '**List Report**' to open the latest List Report of normal results or select '**Single Report**' to open separately in pdf viewer each of the single reports.
- Select '**Show all Reports**' and all reports for the previous 60 days will be available to view / print which will enable users to print off their own copy reports if required.

Note: Those reports previously viewed will appear in a different colour. If reports have been archived after 60 days they will need to be individually regenerated. If reports are greater than 18 months they cannot be regenerated.

To do a search for a Specific Sample

To search for a particular sample or group of samples select '**Specimen Search**' from the left hand side menu using the criteria in the screen below.

The screenshot shows the 'Specimen Search' interface. On the left is a green sidebar menu with the following items: Home, Specimen Search, Download Reports, Messages, Administration, and Logout. The main content area is titled 'Specimen Search' and includes a 'Click for Help' link. Below the title is a form with six input fields arranged in two columns. The left column contains 'Surname:', 'DOB from (DD/MM/YYYY):', and 'UP:'. The right column contains 'Mother's surname:', 'DOB to (DD/MM/YYYY):', and 'Date of Collection (DD/MM/YYYY):'. At the bottom of the form are two buttons: 'Search' and 'Clear'.

A specific sample can be searched for using the above criteria or a group of samples can be searched for using DOB either for one day or a range of days

Example: To search for a Baby Murphy DOB 11/03/2013, enter the details as follows:

Note: DOB MUST be in the format dd/mm/yyyy.

This screenshot shows the 'Specimen Search' form with the following data entered into the input fields: 'Surname:' is 'Murphy', 'Mother's surname:' is empty, 'DOB from (DD/MM/YYYY):' is '11/03/2013', 'DOB to (DD/MM/YYYY):' is '11/03/2013', 'UP:' is empty, and 'Date of Collection (DD/MM/YYYY):' is empty. The 'Search' and 'Clear' buttons are at the bottom.

Select 'search' for the following screen:

Specimen Search [Click for Help](#)

Surname:	<input type="text" value="Murphy"/>	Mother's surname:	<input type="text"/>
DOB from (DD/MM/YYYY):	<input type="text" value="11/03/2013"/>	DOB to (DD/MM/YYYY):	<input type="text" value="11/03/2013"/>
UPI:	<input type="text"/>	Date of Collection (DD/MM/YYYY):	<input type="text"/>

Search Results

Report	Demographics	Lab number	Surname	First name	Date of Birth	Mother's surname	UPI	Date of Collection	Determination
View Report List	Demographics	078248	MURPHY		2013-03-11	MURPHY	724-3307114	2013-03-15	Normal
1									

Report: To view the report select 'View Report List' from left hand side 'Report' column to open. If the sample has been received in the laboratory but the results are not yet available then this will be viewed as 'Results Pending'.

Determination: The 'Determination' column will display 'Normal' if all the requested tests have a result of 'Not Suspected'. This column will display 'other' if a result for any condition is abnormal and/ or a repeat card is required.

Note:

1. To search for all samples from your location for a particular **DOB** enter the DOB range only i.e. for all babies in your location with DOB of 14/05/2013 enter from 14/05/2013 to 14/05/2013.
2. To search by UPI, please ensure a dash (-) is placed after the three digit HIPE Code and ensure that the letter (if any) is also provided eg. Coombe 930-B12345678.

Appendix No.4 Guide for Sample Takers



NEWBORN BLOODSPOT SCREENING TEST 10 STEP GUIDE FOR SAMPLE TAKERS



1

Equipment: Metered sterile lancet with tip no more than 2.5mm in depth, latex free gloves, cotton wool, newborn screening card, envelope, sharps bucket, paper towel, bowl of warm water and soap or cleansing heel.



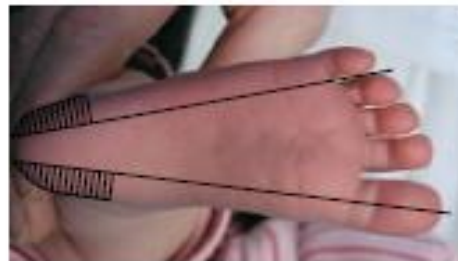
2

Explain to the parents the reason for the test and its importance. Obtain written consent. Complete all sections of the newborn screening card in clear print using a black ballpoint pen. Do not contaminate filter paper circles by allowing circles to come in contact with spillage or by touching before or after blood collection.



3

Instruct parents to keep baby's feet warm prior to test by applying two sets of socks or placing a set of socks beneath babygrow.



4

Select the puncture site on the heel. The preferred puncture and least hazardous site are indicated by the shaded areas. Warm the site prior to sampling by rubbing the skin for 1-2 minutes to increase blood supply. Preferably take the sample from the infant while the parent cuddles the baby, the test may be taken while the baby is breastfeeding. Breastfeeding during a painful procedure effectively reduces the response to pain in the newborn infants. Non-nurtive sucking e.g. pacifier and skin-to-skin contact reduce procedural pain in newborn infants. Current pharmacological treatments are not appropriate for pain relief during minor procedures like the heel prick in newborn infants.



5

Cleanse the heel with warm soapy water. Pat dry.



6

Puncture heel. Wipe away first drop of blood with cotton wool. Allow another large blood drop to form.



7

Touch the circle marked on the card gently to the hanging drop of blood so that the blood soaks through from back of the card to the other side.



8

Fill the two outer circles first; blood drops must be soaked through from the rear to the front of the card, filling all circles completely. Check that the blood has soaked completely through completing the circle on the front as well as the rear of the card.

To enhance blood flow, very gentle intermittent pressure may be applied to area surrounding puncture site.

9

Wipe away any excess blood with cotton wool. Press cotton wool firmly onto wound until bleeding stops. Do not use plaster over puncture sites as babies may swallow them. Air dry the newborn screening card before putting into the envelope. This may take up to two hours. Do not use excessive heating as this may invalidate the test.



10

Send the card by Registered post or by Courier to the newborn Screening Laboratory using the yellow fluorescent address labels to reach the Laboratory as soon as possible after collection. Keep a record of all samples sent in each envelope.

Appendix No.5 Community Healthcare Organisations and Local Health Offices

Community Healthcare Organisation	CHO Head Office	LHO	LHO Population	CHO Population
1	Upper Main St, Ballyshannon, Co. Donegal Tel: 071 9834000	Donegal	164,023	
		Sligo, Leitrim, West Cavan	95,598	
		Cavan, Monaghan	129,427	
				389,048
2	Merlin Park Hospital, Co. Galway Tel: 091 775044	Galway, Roscommon	250,653	
		Roscommon	64,065	
		Mayo	130,638	
				445,356
3	31/33 Catherine St. Limerick Tel: 061 483506	Mid-West (Clare, Limerick, North Tipperary/East Limerick)	379,327	
				379,327
4	Model Business Park, Model Farm Road, Cork Tel: 028 40404	Kerry	145,501	
		Cork - North	89,531	
		Cork – North Lee	181,567	
		Cork – South Lee	190,960	
		Cork – West Cork	56,530	
				518,588
5	Lacken, Dublin Road, Kilkenny Tel: 045- 882588	Carlow/Kilkenny	130,315	
		South Tipperary	98,105	
		Waterford	127,807	
		Wexford	145,273	
				501,500
6	Vergemount Hall, Clonskeagh, Dublin 6 Tel: 01 2680506	Dublin South East/Wicklow (Former - Dublin South East)	115,359	
		Dublin South East (Former - Dublin South)	130,563	
		Dublin South East/Wicklow (Wicklow)	118,542	
				364,464
7	Oak House, Lime Tree Avenue, Millennium Park, Naas, Co Kildare Tel: 045 880 400	Dublin South Central Former - Dublin South City)	144,858	
		Dublin South Central (Former - Dublin West)	146,332	
		Dublin South West Wicklow (Former - Dublin South West)	154,471	
		Dublin South West Wicklow (Former - Kildare/West Wicklow)	228,410	
				674,071
8	Arden Road, Tullamore, Co Offaly Tel: 057 9315850	Louth	122,098	
		Meath	187,880	
		Laois/Offaly	157,246	
		Midlands (Longford/Westmeath)	125,164	
				592,388
9	Swords Business Campus, Balheary, Swords, Co. Dublin Tel: 01 8131838	Dublin North	244,362	
		Dublin North City	337,124	
				581,486
				4,446,228

