SIMMESN

The Italian Society for the Study of Hereditary Metabolic Diseases and Newborn Screening

Expanded Newborn Screening

Information pack for parents

Contents

Introduction

SIMMESN and the entire Board of Directors have supported and promoted the creation of this information pack, confident that it will be a valuable resource for all parents.

The Chairman
Andrea Pession

Dear Parents,

This brochure aims to provide you with essential information about **Expanded Newborn Screening**, to help you understand the importance of life-saving diagnostic testing for your baby.

The birth of a child is a joyful moment, but it can also bring worries and uncertainties. During this time, you will receive a great deal of information. Learning about newborn screening is crucial for your **baby's future well-being** and that of your entire family.

This brochure does not replace the advice of your doctor or paediatrician, but we hope it will serve as a helpful resource, especially if your baby is called back for further testing following a positive screening result.

Knowledge leads to understanding!

Italian Psychologists
Hereditary Metabolic Diseases

Developed by the SIMMESN Working Group of Psychologists Specializing in Hereditary Metabolic Diseases

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For more information on ENBS and on SIMMESN visit
https:// www.simmesn.it/en/

Scan the QR code with your smartphone to download the digital information pack

What is Expanded Newborn Screening (ENBS)?

Expanded Newborn Screening (ENBS) is a comprehensive, integrated, multidisciplinary program for secondary health care.

ENBS aims to **identify** newborns at risk of developing an hereditary Metabolic Disease (see list) across the entire neonatal population. This allows for the early **diagnosis** of affected infants and, in confirmed cases, the initiation of life-saving treatment as soon as possible.

In most cases, ENBS detects the disease before clinical symptoms appear, allowing for the early start of the most effective treatment and often improving the severe prognosis of these conditions. The Italian National Health Service fully funds ENBS.

Recently, Italian legislation (Law 167 of 2016, Appendix 1, and Ministerial Decree of October 13, 2016) expanded Newborn Screening nationwide to include a list of around 40 Metabolic Diseases (see list on pages 12 and 13). The law outlines the screening criteria, target populations (both regional and national), and the procedures, as well as the list of diseases.

How is it done?

BLOOD SAMPLE

Expanded Newborn Screening is conducted at the birth facility between the 48th and 72nd hour of the newborn's life. A small blood sample is taken and placed on a Guthrie card, along with the newborn's personal details.

What happens to the Guthrie card after the sample is collected?

EXECUTION OF THE ANALYSIS

After the sample is collected, the Guthrie card is promptly sent to the screening laboratory for testing.

The laboratory is also responsible for managing the newborn's data (including sensitive information).

If one or more biomarkers are found to be abnormal, suggesting a potential disease covered by the law, the following actions may be taken:

- A second Guthrie card may be requested from the birth facility;

or

- An evaluation at a specialized Clinical Center may be required;

or

- Immediate admission to a specialized Metabolic Clinical Center may be required, as the detected values could indicate a health risk to the newborn.

The Clinical Center will take over the newborn's care and immediately start the appropriate treatment. The center will also manage ongoing clinical and biochemical monitoring and complete the diagnostic process with additional investigations, including molecular testing, if necessary.

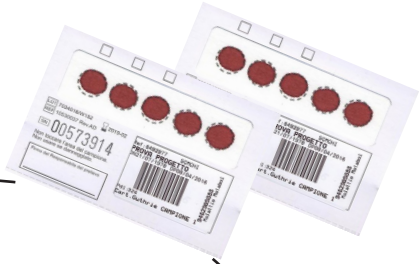
PLEASE NOTE!

A recall for screening does not mean a diagnosis of disease. A diagnosis can only be confirmed through the diagnostic confirmation process.

Summary

Collection of the sample on the Guthrie card at the birthing facility

The Guthrie card with the blood sample is sent to the screening laboratory



If the result is positive, the birth facility is asked to repeat the sample and send a second Guthrie card

Alternatively, you may be instructed to visit a specialized Metabolic Clinical Center, where a multidisciplinary team will address your questions and support you, including counseling.

Why is ENBS an important tool for your baby's health?

The diagnosis of a hereditary Metabolic Disease is a complex process that can only be performed at a few specialized centers in Italy. Therefore, the ability to identify diseases that are difficult to diagnose through just a few drops of blood—diseases that can only benefit from targeted interventions—is crucial for the newborn's future.

ENBS is a health prevention program. Its goal is to identify individuals with biochemical abnormalities indicative of certain diseases, proceed with diagnostic confirmation, and, in the case of a confirmed diagnosis, initiate specific treatment for the disease and provide ongoing care.

This leads to an improved quality of life for both the newborn and their family.

Avoid searching the internet...

... but if you can't hold back, share the source (website, journal, etc.) of the information with the specialist at the Clinical Center. This will help the parent build a solid foundation of accurate and useful information.

Fighting misinformation can be tough, but this is a great place to start!

Which diseases are included in the screening?

List of conditions currently included in screening, along with their abbreviations. Italian Law 167 of October 16, 2016, Appendix 1, and Ministerial Decree of October 13, 2016.

Amino Acid Disorders	
Phenylketonuria	PKU
Benign Hyperphenylalaninemia	HPA
Biopterin Cofactor Biosynthesis Deficiency	BIOPT(BS)
Biopterin Cofactor Regeneration Deficiency	BIOPT(REG)
Tyrosinemia Type I	TYR I
Tyrosinemia Type II	TYR II
Maple Syrup Urine Disease	MSUD
Homocystinuria (CBS Deficiency)	HCY
Homocystinuria (Severe MTHFR Deficiency)	MTHFR
Tyrosinemia Type III	TYR III
Glycine N-Methyltransferase Deficiency	GNMT
Methionine Adenosyltransferase Deficiency	MAT
S-Adenosylhomocysteine Hydrolase Deficiency	SAHH
Organic Acidemias	
Glutaric Aciduria Type I	GA I
Isovaleric Acidemia	IVA
Beta-Ketothiolase Deficiency	BKT
3-Hydroxy 3-Methylglutaric Aciduria	HMG
Propionic Acidemia	PA
Methylmalonic Acidemia (Mut)	MUT
Methylmalonic Acidemia (Cbl-A)	Cbl A
Methylmalonic Acidemia (Cbl-B)	Cbl B
Methylmalonic acidemia with homocystinuria (Cbl C deficiency)	Cbl C
Methylmalonic acidemia with homocystinuria (Cbl D deficiency)	Cbl D
2-methylbutyryl-CoA dehydrogenase deficiency	2MBG

Malonic Aciduria	MAL
Multiple Carboxylase Deficiency	MCD
3-Methylglutaconic Acidemia	3MGCA
3-Methylcrotonyl-CoA Carboxylase Deficiency	3MCC
2-Methyl 3-Hydroxybutyryl-CoA Dehydrogenase Deficiency	2M3HBA
Isobutyryl-CoA Dehydrogenase Deficiency	IBG
Urea Cycle Disorders	
Citrullinemia Type I	CIT I
Citrullinemia Type II (Citrin Deficiency)	CIT II
Argininosuccinic Acidemia	ASA
Argininemia	ARG
Beta-Oxidation Disorders	
Carnitine Transport Deficiency	CUD
Carnitine Palmitoyltransferase 1A Deficiency	CPT Ia
Carnitine-Acylcarnitine Traslocase Deficiency	CACT
Carnitine Palmitoyltransferase II Deficiency	CPT II
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	VLCAD
Mitochondrial Trifunctional Protein Deficiency	TFP
Long-Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	MCAD
Medium/Short Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	M/SCHAD
Glutaric Aciduria Type II	GA II/MADD
Short-Chain Acyl-CoA Dehydrogenase Deficiency	SCAD
Other Conditions	
Galactosemia	GALT
Biotinidase Deficiency	BTD
Cystic Fibrosis	CF
Congenital Hypothyroidism	CHT

Diseases included in screening but not covered by the Ministerial Decree of October 13, 2016

Some Italian regions offer optional screening programs for certain hereditary metabolic disorders. Parents can consent to include these conditions in the diagnostic process through the analysis of the collected drops.

Lysosomal Diseases	Immunodeficiencies
Leukodystrophies	Congenital Adrenal Hyperplasia
Spinal Muscular Atrophy (SMA)	

Summary

If your child is recalled following a positive screening result,
it does not necessarily mean that they have an illness

Follow the instructions provided and
return to the birth facility for a second test

or

go to a specialised Clinical Center

Finally...
if your child is called back due to a positive screening result
you can ask questions and get clear, detailed information from the doctors
at the Metabolic Clinical Center.

Notes

SIMMESN

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