

What is the neonatal screening performed on a blood drop and what is it used for?

The neonatal screening performed on a blood drop is a preventive medical activity which allows an early identification of newborn babies who are affected by a hereditary disease even before the first symptoms occur.

These are rare diseases that, if identified in the first days of life, could be treated, thus avoiding severe and sometimes even fatal consequences to the baby.

Therefore, it is of vital importance that ALL newborn babies undergo this test in order to guarantee their right to a good health.

The neonatal screening is not only a test but an integrated and multidisciplinary prevention program involving many players, besides parents and their children: the Birth Centers, where the blood is drawn, the laboratories, where the test is performed, and the clinical centers in charge of the newborn babies in case the babies are affected by one of the diseases.



The Campania Region takes care of you

To prevent diseases since the first day of life



CEINGE
CENTRALIZED ADMISSION
SERVICE

*For information regarding the booking
of a genetic consultation and the
access to the diagnostic services:*

Hours of operation - phone number

9.00 a.m. - 1.30 p.m. ■ +39 081 7462436

11.00 a.m. - 1.30 p.m. ■ +39 081 7463169

2.30 p.m. - 5.30 p.m. ■ +39 081 3737781

+39 081 3737727

e-mail

accettazione@ceinge.unina.it

website

www.ceinge.unina.it



CENTRO UNICO REGIONALE
**SCREENING
NEONATALE**

**NEONATAL SCREENING
SOLE REGIONAL CENTER**

*Screening Center Laboratory
Supervisor prof. Margherita Ruoppolo*

Phone numbers

+39 081 3737933

+39 081 3737776

e-mail PEC

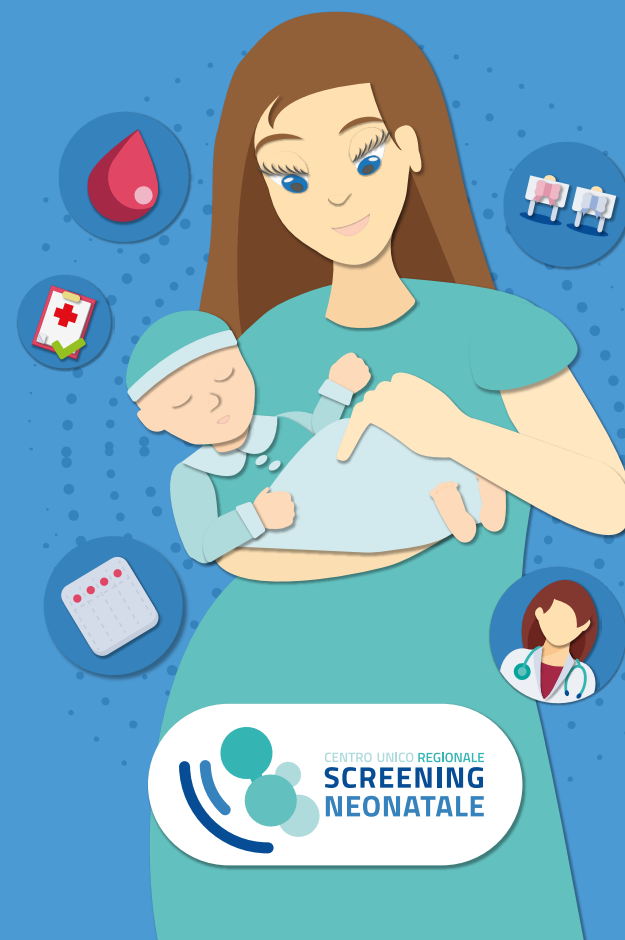
labsne@pec.ceinge.unina.it



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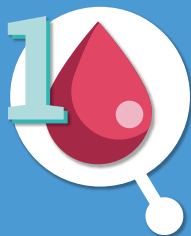


FACT SHEET REGARDING THE **NEONATAL SCREENING** PERFORMED ON **A BLOOD DROP**



CENTRO UNICO REGIONALE
**SCREENING
NEONATALE**

How is the test performed?



The screening begins with the drawing of a blood drop, usually from the heel, performed on the baby's second or third day of life before he is discharged from the Birth Center.

The blood drops are collected on a special blotting paper which is part of a **card** containing all the newborn identification data.



The **card** is collected every day and sent to the

Neonatal Screening SOLE REGIONAL CENTER

located at the

Advanced Biotechnologies CEINGE

Via Gaetano Salvatore, 486
Naples, Italy

where the tests necessary to identify the diseases included in the screening (see Table 1) are performed.

Once the analyses are completed, the **card** is stored at the CEINGE location for 5 years in order to allow further analyses related to the scope of the screening, if necessary.



The personal data are used solely for the scope of the screening and according to the current privacy laws and regulations.

TABLE 1

Pathologies subject to neonatal screening performed on a blood drop in the Campania Region

AMINO ACID DISORDERS

Phenylketonuria PKU
Hyperphenylalaninemia HPA
Bioppterin defect in cofactor biosynthesis BIOPT $\times$ BS
Bioppterin defect in cofactor regeneration BIOPT-REG
Tyrosinemia type I TIR1
Tyrosinemia type II TIR 2
Maple syrup urine disease MSUD
Homocystinuria CBS
Homocystinuria MTHFR
Citrullinemia type II CIT 2

UREA CYCLE DISORDERS

Citrullinemia type I CIT 1
Argininosuccinic aciduria ASA
Argininemia ARG

BETA-OXIDATION DISORDERS

Carnitine uptake defect CUD
Carnitine palmitoyltransferase deficiency I CPT 1
Carnitine palmitoyltransferase deficiency II CPT 2
Carnitine/acylcarnitine translocase deficiency CACT
Very long-chain acyl-CoA dehydrogenase deficiency VLCAD
Trifunctional protein deficiency MTP/LCHAD
Medium-chain acyl-CoA dehydrogenase deficiency MCAD A

Medium/short-chain 3-OH acyl-CoA dehydrogenase deficiency M-SHAD
Glutaric acidemia type II GA2/MADD

ORGANIC ACIDURIAS

Glutaric Acidemia type I GA 1
Isovaleric Acidemia IVA
Beta-ketothiolase deficiency BKT
3-hydroxy 3-methylglutaric aciduria HMG
Propionic Acidemia PA
Methylmalonic acidemia (Mut deficiency) MMA
Methylmalonic acidemia (ICbl A deficiency) MMA
Methylmalonic acidemia (Cbl B deficiency) MMA
Methylmalonic acidemia with Homocystinuria (Cbl C deficiency) MMA-HCYS
Methylmalonic acidemia with Homocystinuria (Cbl D deficiency) MMA-HCYS
2-methylbutyryl-CoA dehydrogenase deficiency 2MBG
Multiple carboxylase deficiency MCD
Malonic aciduria MA

OTHER DISEASES

Galactosemia
Biotinidase deficiency
ADA SCID

OTHER GENETIC DISEASES

Cystic fibrosis FC
Congenital hypothyroidism (IC)

When and how will you be informed of the screening results?

If abnormal values are not observed, which means the test is negative, the Birth Center will receive the communication of the negative screening within few days after the blood is drawn.

In a small percentage of cases, it could be necessary to repeat the test and, therefore, the parents could be contacted again by the Birth Center.



WARNING

Being called back does not mean that the child is sick but it means that further analyses are necessary.

In fact, it could happen that the blood sample is insufficient or inadequate which would make it necessary to draw more blood.

If later tests show that the patient is positive for one of the pathologies, the newborn baby is admitted immediately in the pediatric clinical centers of the Federico II University Hospital Agency (AOU) and of the Santobono-Pausillipon Hospital (AORN) for cures and clinical assistance, for constant monitoring, and for the most appropriate therapeutic strategies.

Although this screening is provided for by law, the possibility of opposing to it is available by expressing your dissent before the blood is drawn.

Such dissent will be added as a note to the baby's medical records.