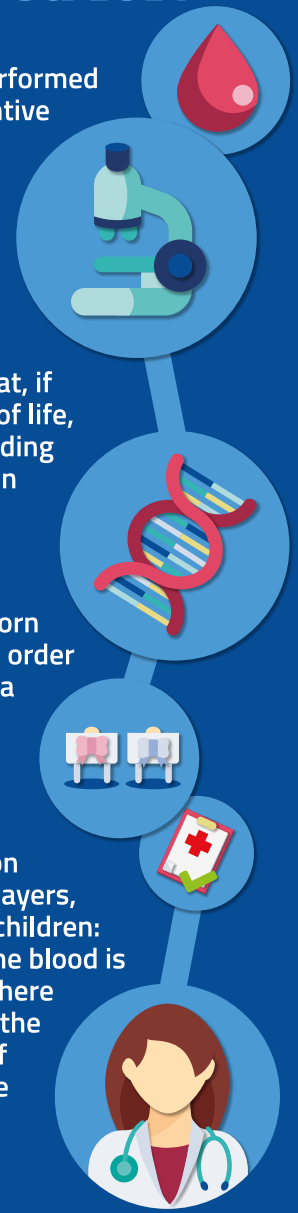
What is the neonatal screening performed on a blood drop and what is it used for?



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.

REGIONE CAMPANIA

The Campania Region takes care of you

To prevent diseases since the first day of life

ceinge BIOTECNOLOGIE AVANZATE

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

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www.ceinge.unina.it

CENTRO UNICO REGIONALE SCREENING NEONATALE

NEONATAL SCREENING SOLE REGIONAL CENTER

Screening Center Laboratory
Supervisor prof. Margherita Ruoppolo

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website
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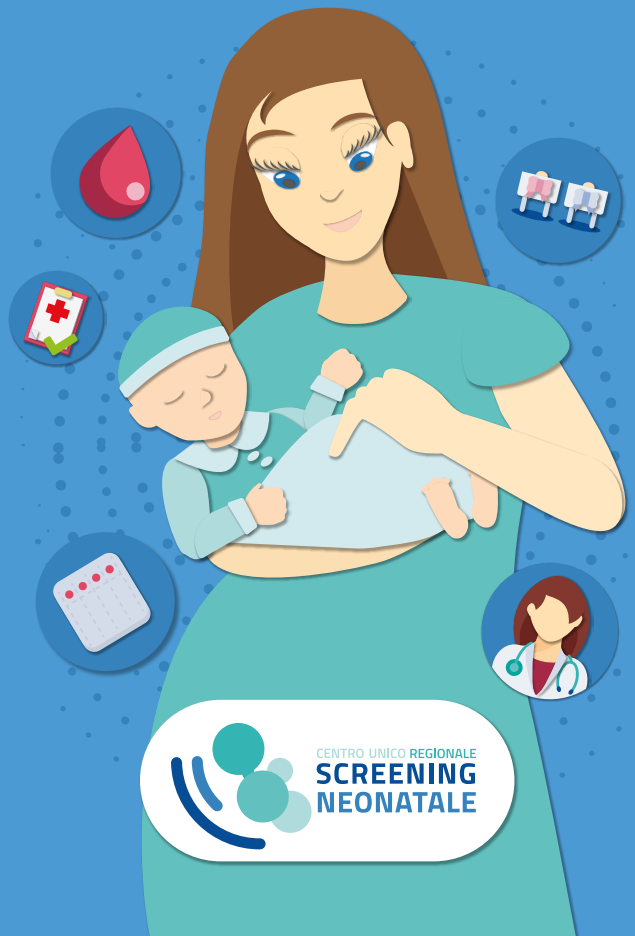
REGIONE CAMPANIA

FACT SHEET REGARDING THE

NEONATAL SCREENING

PERFORMED ON

A BLOOD DROP

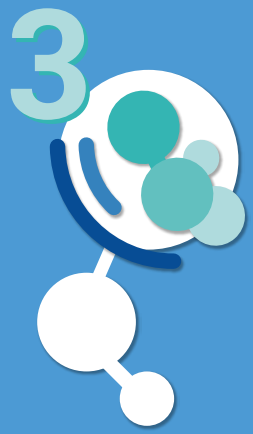


CENTRO UNICO REGIONALE SCREENING NEONATALE

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
- Biopterin defect in cofactor biosynthesis BIOPT□BS
- Biopterin 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-SCHAD
- 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.