

# Expanded newborn screening programme

Newborns shall be offered screening for 25 rare, congenital disorders for which early treatment is vital. The Norwegian newborn screening programme includes testing for 2 endocrinological conditions, 21 metabolic disorders, severe combined immunodeficiency (SCID) and other severe T-cell deficiencies, and cystic fibrosis.

The screening is performed by collecting a blood sample from the infant's heel 48-72 hours after birth. The blood sample, together with details of the mother's name, national identity number, address and telephone number, place of birth (hospital/clinic), time of birth, gestational length, birth weight and gender, is sent for testing at the Unit for Newborn Screening (Nyfødtscreeningen) at Oslo University Hospital (Oslo universitetssykehus).

The procedure requires that the parents have received information about the screening and given their consent. Parents who do not wish to have their baby screened should notify the maternity ward staff. It must be emphasised that there are also other congenital disorders that cannot be detected by this blood test.

## What might screening mean for my baby?

Although a baby may seem perfectly healthy just after it is born, in rare cases it may have a congenital condition such as a metabolic disorder. The longer the baby goes untreated, the greater the risk of permanent disability or death. It is therefore very important to detect any disorder as early as possible so the baby can be started on the necessary treatment.

Only a very small number of infants (about 1 in 1000) are born with the disorders screened for, but for these children early detection is vital.

## What happens after the tests have been done?

The test results are sent to the hospital/clinic where the baby was born. If the test result is normal, the parents will not be contacted. If the baby is suspected of having a disorder, the parents will be contacted immediately by a doctor to arrange for further tests. If the Unit for Newborn Screening receives a faulty blood sample, the parents will be contacted by the hospital/clinic where the baby was born and asked to repeat the test. Being asked to repeat the test does not necessarily mean that there is any suspected problem.

Follow-up of families where a disorder is detected is usually provided by the local paediatric department and/or Oslo University Hospital. If a test result indicates a disorder, the results of further tests will normally be available within 1-2 weeks.



## How reliable is the screening?

In rare cases, especially among premature babies, the test results may indicate a disorder even though the baby is actually normal and healthy. The next tests that are done will quickly remove any uncertainty.

## Storage and use of blood samples

After the screening has been done, the rest of the blood sample will be stored in de-identified form\* in the Unit for Newborn Screening's diagnostic biobank. Storing the sample allows for the possibility of repeating the test if there is any doubt about the diagnosis or of supplementing with other tests that were not available when the baby was born. The sample can also be used for quality assurance and for developing new screening techniques.

## Storage and use of personal data

To ensure best clinical practices in newborn screening, it will be necessary to use data from the screening programme, including test results, details of medication the baby was given or other factors that might affect the test results, and also any medical treatment already provided. The aim is to ensure that screening is performed as reliably as possible and to monitor the quality of treatment provided for the disorders screened for. This data is stored indefinitely.

Oslo University Hospital is legally responsible for data protection on the newborn screening programme. Once registered on the screening programme, the test subject has the right to gain access to the data and the right to have that data deleted (see [www.oslo-universitetssykehus.no/nyfodtscreeningen](http://www.oslo-universitetssykehus.no/nyfodtscreeningen)). If any registration errors have been made, the rights include correction of the data.

## Consent to newborn screening and to storage and use of personal data

The procedure requires that the parents have received information about the screening and given their consent. Parents who do not wish to have their baby screened should notify the maternity ward staff.

Parents wishing to have their baby screened, but who do not wish the sample to be stored must notify the Unit for Newborn Screening of this. Special forms for notifications are available from the maternity ward/hospital/clinic where the baby was born or at [www.oslo-universitetssykehus.no/nyfodtscreeningen](http://www.oslo-universitetssykehus.no/nyfodtscreeningen)

We also request your consent to allow the blood sample and personal data to be used for quality assurance and to improve the screening programme. Participation in this is voluntary. Parents who do not wish to give their consent are not required to provide reasons for their decision, and there are no consequences for any treatment received by the baby.

## Research

Blood samples and personal data from the Newborn Screening Programme may also be used for research. This requires separate consent in accordance with the ordinary rules for clinical healthcare research (see [www.oslo-universitetssykehus.no/personvern](http://www.oslo-universitetssykehus.no/personvern)).

## More information

The Unit for Newborn Screening can provide advice and counselling about the disorders screened for. The Unit for Newborn Screening's website [www.oslo-universitetssykehus.no/nyfodtscreeningen](http://www.oslo-universitetssykehus.no/nyfodtscreeningen) provides a description of these disorders and how they are treated. The website [www.helsenorge.no](http://www.helsenorge.no) also provides information about rare disorders and the treatment available for them.

For certain disorders, national advisory services have been established to provide advice and counselling. See [www.hesledirektoratet.no/funksjonshemninger](http://www.hesledirektoratet.no/funksjonshemninger) for a listing of these services. The Directorate of Health – Rehabilitation and rare disorders department's free helpline can also provide more information.

Unit for Newborn Screening: tel. 02770,  
e-mail [nyfodtscreeningen@ous-hf.no](mailto:nyfodtscreeningen@ous-hf.no)

\* Without the baby's name or date of birth, just a sample code