

## **Information for parents about newborn screening**

The PKU test has been offered to newborns in Sweden since the mid 1960s. The purpose of the test is to find children with very rare but treatable metabolic diseases. An early diagnosis is crucial to enable a good prognosis for the child.

If any of the screening results are abnormal a paediatrician from your home hospital will contact you and arrange for further tests. The risk that your child has one of these diseases is very remote. Among the approximately 100,000 children born in Sweden each year, some 70 to 80 are found to have one of the diseases that we are testing for. From the beginning, the only disease in the screening was phenylketonuria, PKU. Over the years the number of included diseases has increased as screening methods and treatments have improved. A list of the diseases currently in the screening, including a short description of each, can be found on our web page [www.karolinska.se/pku](http://www.karolinska.se/pku). There you can also find more information about the PKU test.

The test is taken as soon as possible after the age of 48 hours. A few drops of blood are collected on a filter paper, allowed to dry and sent to the PKU laboratory for analysis. The results are ready within a week. Sometimes the sample is not sufficient in which case we will ask for a new sample. After analysis the sample is kept in a biobank for complementary analysis, quality control and method development. It is also available for research approved by an ethical committee. The sample is kept in accordance with the Swedish biobank law. The data we save is the mother's name and personal number, the child's personal number or date of birth and gender, the time of sampling, the length of the pregnancy and the name of the maternity ward. We need this information to keep track of the sample as well as to interpret the analysis results.

If you do not consent to have your child's sample saved in the biobank, you can ask for it to be discarded. This is done in writing by filling in a No-counterfoil, which you can get from the nurse who takes the sample. It must be signed by both legal guardians (alternatively the guardian if there is only one). The signed counterfoil is sent to *Stockholms Regionala Biobankscentrum, Box 4151, 141 04 Huddinge*. You can also download the No-counterfoil through the heading "Patient" at [www.biobanksverige.se](http://www.biobanksverige.se).

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