NEWBORN SCREENING PROGRAM HEEL BLOOD SAMPLING
CLARIFICATION AND REFUSAL FORM
…… FAMILY HEALTHCARE UNIT/HOSPITAL

Baby name-surname	:
Baby ID number	:
Date of birth	:
Mother's name-surname	:
Mother's ID number	:
Father's name-surname	:
Phone	:
Address	:

Type of Sample
☐ First sample
☐ Second sample
☐ Repeated sample
☐ Incorrect feedback sample
☐ SMA First sample
☐ SMA Second sample

A few drops of blood will be drawn from your baby's heel to be tested under the National Newborn Screening Program. With the blood sample taken, diseases which are currently in the program; Spinal Muscular Atrophy, Phenylketonuria, Congenital (Congenital) Hypothyroidism, Biotinidase Deficiency, Cystic Fibrosis and Congenital Adrenal Hyperplasia will be examined and you will not be charged for these tests.
These diseases are extremely rare and may not present immediately in the baby, but they can be detected by tests. These diseases are extremely rare and may not present themselves immediately, but they can be detected by tests. If not diagnosed and treated early, they may have serious consequences, and when diagnosed early, with drug therapy and with a special diet it can be controlled or treated. Therefore, it will be in your baby's best interest to have the test done. Heel blood is sent to the Newborn Screening Laboratories of the Ministry of Health in Ankara and Istanbul, where biochemical methods are used for the screening of Phenylketonuria, Congenital Hypothyroidism, Biotinidase Deficiency, Cystic Fibrosis and Congenital Adrenal Hyperplasia, and molecular genetic methods are used for the screening of Spinal Muscular Atrophy (SMA). SMA screening only checks for mutations in the SMN1 and SMN2 genes. Blood samples are not taken out of the institution, screening results are not shared with third parties and institutions. Two different blood samples are taken from the babies before they are discharged from the hospital and within the first week. Sometimes, more blood may be required from your baby due to insufficient blood taken, repeating the test with questionable results, or similar reasons. These tests are for screening purposes and may require further examination and clinical evaluation for definitive diagnosis.

What is phenylketonuria?
Phenylketonuria is a congenital metabolic disease. Children born with this disease cannot absorb an amino acid called phenylalanine found in protein foods. As a result, increased phenylalanine and its residues in the blood and other body fluids damage the developing brain of the child and cause the child to have severe intellectual disability and many more symptoms related to the nervous system.

What is congenital hypothyroidism?
Congenital hypothyroidism is a thyroid hormone deficiency that begins with birth. Thyroid hormones are located in the front of the neck and secreted from the thyroid gland which is an endocrine gland. In infants with hypothyroidism, the thyroid gland may be underdeveloped or undersized or even if it has a normal appearance, in cases where it does not work well, thyroid hormone secretion is insufficient.Thyroid hormones are essential for growth in infants and children, and for the normal progression of brain development in infants.Therefore, thyroid hormone deficiency (hypothyroidism) negatively affects growth and mental development.

What is biotinidase deficiency?
Biotinidase deficiency is a congenital metabolic disease. Due to a congenital vitamin deficiency in children born with this disease, there is a disruption in the process of fuel production in the body. As a result of this vitamin deficiency, skin lesions that start around the eyes, mouth, nostrils and diaper area

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can spread to the body in a short time. Hair, eyelashes and eyebrows begin to shed simultaneously with these skin symptoms. Due to the accumulation of acidic substances, the patient starts to breathe frequently and deeply, and there is a change in consciousness that progresses to coma. Untreated crisis can lead to death. Some acid substances accumulated in the body during the non-crisis period could have a negative effect on the visual and auditory nerves in the long term. Therefore, permanent vision and hearing loss may develop.

What is cystic fibrosis?

Cystic Fibrosis is a congenital disease that mainly affects the lungs and digestive system. In infants with cystic fibrosis, the disease and symptoms can occur at various ages. The most common complaints are: recurrent lung infections, getting sick frequently, having lots of greasy, foul-smelling stools and not gaining enough weight because they can't digest the food they take. Cystic fibrosis patients who are diagnosed early thanks to newborn screening are treated with appropriate diet, medications and physiotherapy. Although there is no definitive treatment for cystic fibrosis, patients can have a longer and healthier life thanks to new treatments being found every day.

What is congenital adrenal hyperplasia?

Congenital adrenal hyperplasia (CAH) is a disease in which the adrenal glands cannot produce enough cortisol (and sometimes the hormone that regulates the body's salt balance: aldosterone), which is necessary for life. The production of these hormones occurs with the help of some enzymes in the adrenal gland, and these hormones are essential for the body to deal with stress and maintain salt balance. In its deficiency, adrenal insufficiency develops which is life threatening and it causes death due to exposure to serious infections and diarrhea in infancy. With early diagnosis, medical and surgical treatment is possible, thus patients can have a healthy life.

What is spinal muscular atrophy (SMA)?

SMA is the name given to a group of inherited neuromuscular (related to muscles and nerves) diseases. The disease is known for progressive muscle weakness and muscle wasting. There are four types of the disease, and the most common is SMA Type I, with an incidence of approximately 60-70%. SMA Type I is the most common type and is the most severe and fatal, resulting in severe respiratory failure and eventual death before the age of two. Other types occur later and progress with milder clinical symptoms. If the disease is diagnosed early, the disease can be brought under control with new treatment options and the quality of life of the patients increases.

If you do not agree to have a heel blood sample taken from your baby, you must mark and sign the relevant section after reading the form.

☐ I was informed about the heel blood screening and the diseases screened, my questions were answered if I demanded, I read and understood the text written above, although **I DO NOT ALLOW** my baby's heel blood sample to be taken.

Baby's Relative (Parent-guardian)
Name and surname :
Relation :
Signature :

The mother/father/guardian of the baby, whose information is given above, was informed about the subject on the date of ……/……/…….....

Healthcare Worker
Name-surname :

Physician
Name-surname :

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Title :
Signature :

Signature :
Signature :

☐ The person was informed about the heel blood screening and the diseases screened, her/his questions were answered, the text above was read, but s/he refuses to take the heel blood sample of the baby and to sign the report.

Healthcare Worker
Name-surname :
Title :
Signature :

Physician
Name-surname :
Signature :