When your baby is born, blood is taken from the baby to investigate some congenital diseases for which early diagnosis is important. These diseases are extremely rare and may be asymptomatic in the baby thus it can only be detected by testing. It can have serious consequences if not diagnosed and treated early and can be easily treated when diagnosed early. Also the disease can also be effectively controlled with medication or a special diet. A positive test for any of these diseases in your child in these screening results does not mean that your child is sick, it means that the disease is suspected. The baby with a positive screening test should definitely be examined further and it should be understood whether s/he is sick or not. In addition, since many of these diseases can cause progressive and irreversible damage over time, these examinations and assessments should be done as soon as possible.

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 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.

It has been determined that your baby may have ……………………………. as a result of the screening tests.

YOUR BABY NEEDS ADVANCED EXAMINATION AND CLINICAL EVALUATION IMMEDIATELY.

Your baby should be taken to the ……………………………………………………… Clinic for further examination, diagnosis and treatment. If you do not agree to take your baby to the clinic, you must mark and sign the relevant section after reading the form.

☐ I was informed that my baby's screening test result was suspicious, that my baby needed further examination and evaluation, and that I was also informed about the clinics where the evaluation could be made. When I requested, my questions were answered, I read and understood the text written above, although I DO NOT AGREE to take the baby to the relevant clinic. I accept all legal and criminal responsibility for the possible negative consequences for my baby and the consequences of undiagnosed diseases that may lead to disability or death of my baby.

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 :
Title :
Signature :

Physician
Name-surname :
Signature :

☐ The person was informed about the negative consequences that may arise for the baby if the referral was not followed. Her/his questions were answered, the text above was read and despite this, s/he refuses to take the baby to the relevant clinic and to sign the report.

Healthcare Worker
Name-surname :
Title :
Signature :

Physician
Name-surname :
Signature :
REFERRING THE NEWBORN SCREENING PROGRAM
CLARIFICATION AND REFUSAL FORM