



T. C. Ministry of Health
SMA Carrier Screening Consent Form

Name- Surname : Date of Birth :
Turkish Identity Number : Telephone Number :
Address : Custodian (if any) :

INFORMATION ON GENETIC TEST

1. The purpose of the genetic test to be performed is to detect the highest possible carrier form of SMA (Spinal Muscular Atrophy) disease and does not include the diagnosis or carrier of other genetic diseases. With the result, another genetic test may be needed or the analysis may need to be repeated.
2. The method to be studied uses the Real-time quantitative Polymerase Chain Reaction (qPCR) technique applied to the DNA material obtained from the blood sample and is intended to detect changes in the 7th exon dose of the SMN1 gene. This method can show the form defined as "1+0", which is detected in approximately 90% of the carriage of SMA disease, with approximately 95% sensitivity and specificity. This test cannot show other rare SMA carriers ("2+0" form deletions, pathogenic sequence variants and mosaicism), changes in SMN2 gene dose, and carriers in other genes included in the differential diagnosis of SMA disease.
3. As with all other genetic testing, there is a small chance of failure or error in sample analysis. These conditions may be related to technique or rare variants seen in the person's DNA. Although extensive measures are taken to avoid these situations, a 100% guarantee cannot be given.
4. Samples may be requested again in cases where the sample is not sufficient, not received/sent under appropriate conditions. Information about the genetic test result is stored in a database that can only be accessed by the laboratory's authorities. The information and results of the person whom the sample is taken cannot be transferred to third parties other than the person himself/herself or his/her relatives that he/she give written permission, the relevant physician and legal obligations.
5. An individual whose screening result is found to be suspicious should apply to the nearest medical genetics polyclinic in order to receive genetic counseling.

Statement of the Sample Giver

I **READ** and **UNDERSTAND** the above explanations before giving my sample. Information about the transaction was transferred to me, my questions were answered.

In these conditions, for taking blood samples for genetic analyzes to detect the carrier of SMA disease, performing the test and destroying the biological sample taken from me at the end of the test, using the results of the tests made from biological samples in scientific studies, if necessary, on the condition that ethical rules are followed, without any pressure and obligation;

I ALLOW I DON'T ALLOW

NAME-SURNAME DATE SIGNATURE

The person being screened :

Doctor :

Custodian (if any) :