



T.C. SAĞLIK BAKANLIĞI

SMA CARRIER SCREENING CONSENT FORM

First and Last Name : Date of Birth :
TR ID No : Tel :
Address : Guardian, if any :

INFORMATION ON THE GENETIC TEST

1. The purpose of the genetic test is to determine the possible carrier status of SMA (Spinal Muscular Atrophy) and does not include the diagnosis or carrier status of other genetic diseases. The result may require another genetic examination, or the analysis may need to be repeated. It is recommended that a couple planning a pregnancy wait for the results of the screening test.
2. The method to be studied uses the Real-time quantitative Polymerase Chain Reaction (qPCR) technique applied to DNA material obtained from a blood sample and is intended to detect changes in the dose of exon 7 of the SMN1 gene. This method can show the form defined as "1+0", which is detected in approximately 90% of SMA disease carriage, with a sensitivity and specificity of approximately 95%. This test cannot show other rare SMA carriers (deletions in the "2+0" form, pathogenic sequence variants and mosaicism), changes in the dose of the SMN2 gene and carriers in other genes included in the differential diagnosis of SMA disease.
3. As with all other genetic tests, there is a small chance of failure or error in sample analysis. These may be related to the technique or to rare variants in the individual's DNA. Although extensive precautions are taken to avoid these situations, 100% guarantee cannot be given.
4. In cases where the sample is not sufficient, not taken/sent under appropriate conditions, a repeat sample may be requested.
5. Information about the genetic test result is stored in a database that can only be accessed by the authorities of the laboratory. The information and results of the individual from whom the sample was taken cannot be transferred to third parties other than the individual himself/herself or his/her relatives who have given written permission, the relevant physician, and legal obligations. Individuals can also obtain their test results from the family health centres to which they are affiliated.
6. Individuals who are found to be carriers should apply to the nearest medical genetics outpatient clinic for genetic counselling.

Declaration of the Person from whom the Sample was taken

I have **READ** and **UNDERSTOOD** the above explanations before giving my sample. I was informed about the procedure and my questions were answered.

Under these conditions, I give my consent for a blood sample to be taken from me for genetic analyses to determine the carrier of SMA disease, for the test to be performed and for the biological sample/genetic material taken from me at the end of the test to be stored/destroyed, to be de-identified and used for further examination and scientific purposes, to be shared on scientific platforms, provided that ethical rules are followed, without any pressure and obligation;

YES

NO

FIRST – LAST NAME

DATE

SIGNATURE

The person being screened:

Doctor:

Guardian, if any: