SMA consent form

GC: ..... Date: ...../...../…….

Spinal Muscular Atrophy (SMA) is an inherited neuromuscular disorder caused by mutations in the SMN1 gene. It follows an autosomal recessive pattern of inheritance. Based on SMA status, individuals fall into three categories:

1. Healthy non-carriers who have at least two copies of the SMN1 gene.
2. Healthy carriers who possess one normal and one mutated copy of the SMN1 gene. Carriers are not at risk of developing the disease, but they can pass the mutated gene to their offspring.
3. Affected individuals, who have inherited two defective copies of the SMN1 gene (one from each parent), and exhibit varying degrees of disease severity.

Important Notes on SMA Carrier Detection:

- Carrier screening methods such as MLPA are designed to detect deletions in the SMN1 gene. Therefore, if the mutation is a point mutation, it may not be detected by such tests.
- In some individuals, although two SMN1 copies are present, both copies may reside on the same chromosome (cis configuration). These individuals may still be carriers, and this situation cannot be identified by standard SMA screening methods such as MLPA. Overall, carrier detection sensitivity is estimated at 90–95% due to these limitations.
- If only one partner in a couple is tested and found to be a carrier, the other partner must also be tested. Additionally, it is recommended that other family members undergo carrier testing.
- If one or both partners are found to be carriers, prenatal diagnosis (PND) is advised.
- In about 98% of SMA cases, the child inherits the condition from carrier parents. However, in approximately 2% of cases, a de novo (new) mutation is responsible. Therefore, even if both parents test negative as carriers, there remains a 2% residual risk.

Prenatal Diagnosis (PND) Must Be Performed in Two Stages:

- **Pre-conception stage: If definitive results are obtained, the couple may proceed with pregnancy.** However, couples must understand that even if one or both test non-carrier, due to test limitations, a small residual risk of being a carrier and having an affected child remains. Hence, PND can still be performed if only one partner is identified as a carrier. According to Iranian population data, 3% of SMA cases involve only one carrier parent.
- **Post-conception stage: The couple should contact the center again in the 9th week of pregnancy to schedule sampling of chorionic villi (CVS) or amniotic fluid, to assess the fetus for SMA.**

* This center performs SMA testing by checking for deletions of exons 7 and 8 of the SMN1 gene, which account for approximately 95% of SMA cases. The remaining 5% involve point mutations that cannot be detected by standard methods .

Final Result and Turnaround Time:

Test results are typically available in approximately two weeks. In some cases, additional samples may be required. If any issues arise or the turnaround time is extended, the family will be notified by phone. If the first stage is completed and results are confirmed, the turnaround time for CVS is 1–2 weeks, and for amniotic fluid it is around 1 month (due to the need for cell culture).

Note: As in all stages, repeat testing may be necessary, and no additional fees will be charged for such repeats.

It has been explained to me/us that the technical results of the test performed may be used for scientific and research purposes, provided that all identifying personal information remains confidential. I/we hereby give my/our consent for such use.

It has also been clarified that the laboratory is obligated to provide technical information to relevant national authorities upon request, in accordance with the regulations approved by the Ministry of Health.

We, Ms..... and Mr. ....., the undersigned, having read all the items stated in this consent form, received adequate explanations, and been given sufficient opportunity to ask questions, hereby give our informed consent freely and voluntarily, without any pressure or coercion. We fully understand the potential outcomes of the aforementioned tests, including possible emotional, psychological, and physical consequences.

We hereby sign this consent form and request the center to perform prenatal diagnostic testing for Spinal Muscular Atrophy (SMA). We understand that once the test has been accepted, it cannot be cancelled.

Name, Surname, and Signature (Woman):

Name, Surname, and Signature (Man):

Name, Surname, and Signature of Genetic Counselor: