WHAT IS SPINAL MUSCULAR ATROPHY (SMA)?

SMA is an inherited, progressive lower motor neurone disease. When left untreated, motor neurons in the spinal cord that control the muscle movements of our body are affected, resulting in lack of energy in addition to feeding and breathing problems. In later stages, the clinical picture may be accompanied by varying degrees of limitation in joint movements, deformities in the spine, nutritional and respiratory problems.

On average, one out of every 50 individuals within the society is a carrier of SMA disease. If the father and mother are both carriers of the disease, the probability of the disease in each pregnancy is 25%. Although consanguineous marriage increases the incidence of the disease, since the carrier rate is high in the society, babies of unrelated individuals may also be born with it. It is necessary for you and your family members to receive genetic counselling.

Coincidentally, the probability of marriage between two individuals who are carriers of SMA is very high.

Since the carrier rates are high in our country, SMA testing is recommended before pregnancy for all couples with or without a sick baby in the family.

When it is determined that the couple is a carrier, genetic counselling, prenatal or pre-implantation diagnostic test options can be offered for a healthy child.

PRE-MARITAL SPINAL MUSCULAR ATROPHY (SMA) CARRIER SCREENING PROGRAMME

STATEMENT OF INFORMATION FOR FEMALE SPOUSE

STATEMENT OF INFORMATION FOR THE FEMALE SPOUSE

In the Spinal Muscular Atrophy carrier screening test, I was informed that I was not a carrier as a result of the sample I gave because my spouse was a carrier, but that I could apply to the medical genetics polyclinic with my result.

I listened/read the information given to me about the disease.

I received information about the places where I can receive genetic counselling.

Date: ...../...../.....

Female Spouse / Prospective Spouse

TCKN

First and Last Name:

Signature

Informing Physician

First and Last Name:

Signature:

TR ID NO: Republic of Turkey Identity Number

This is the copy for the health institution.

SPINAL MUSCULAR ATROPHY (SMA) CARRIER SCREENING TEST INFORMATION FOR A PROSPECTIVE FEMALE SPOUSE WHOSE PARTNER IS A SMA CARRIER

If only your spouse/partner is a carrier, SMA disease due to deletion mutation **is not expected** to occur in your children. Therefore, you will be excluded from the screening programme.

However, depending on the complexity of the disease genetics of SMA and the screening method applied, you **may not be detected** in the screening test in 5% of cases even if you are a carrier. Therefore, new mutations and other rare causes cannot completely exclude the possibility of SMA.

You can apply to the **Medical Genetics Outpatient Clinic** where you can receive genetic counselling with your result.