In case of detection of carrier in terms of SMA:

- Consult the relevant specialists through your family doctor to receive genetic counseling.
- The correct interpretation of your screening test results is very important.
- Please inform your family (such as siblings and cousins) regarding the extended implementation of the screening program.
- If two carriers are married, couples should receive genetic counseling before pregnancy is planned, and they should be informed about diagnostic methods before pregnancy and delivery.

Spinal muscular atrophy (SMA) carriage;

- It is very common in society.
- It is hereditary.
- It is not a disease.
- It does not require treatment.
- It does not turn into a disease.
- It is not an obstacle to marriage.
- It is not an obstacle for having children.

Consult your FAMILY PHYSICIAN to get information about the premarital Spinal Muscular Atrophy (SMA) screening test.

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For Healthy Children

Premarital Screening

PREMARITAL SPINAL MUSCULAR ATROPHY (SMA) SCREENING TEST

What is Spinal Muscular Atrophy (SMA)?

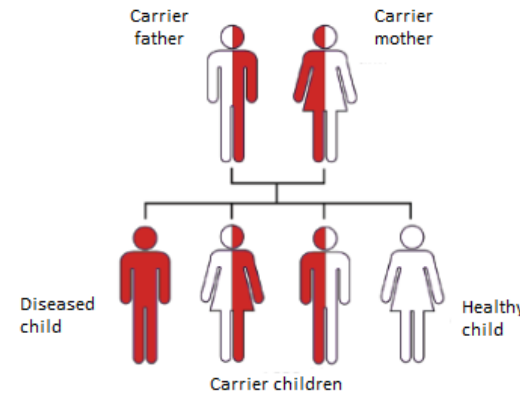
SMA is a progressive inherited muscle disease. The disease affects the anterior horn motor neuron cells in the spinal cord, which control the muscle movements of our body, and weakness, feeding and respiratory problems occur. In the following periods, varying degrees of limitation of joint movements (contractures), spinal deformities (scoliosis), nutrition and respiratory problems may be seen.

- Weakness and muscle wasting occur in voluntary muscle movements. Vision, hearing and cognitive functions are not affected.
- There are different types according to the age of onset and the gained motor development stage.
- The incidence of SMA is 1 in 6,000-10,000 newborn babies.
- Although the natural course of the disease has changed with treatment options in recent years, all studies indicate that patients who are diagnosed and followed up at the time of the onset of symptoms should be followed up under long-term treatment.
- The response to treatment shows individual differences, and SMA is a serious disease that involves systems other than motor neurons.

How is SMA inherited?

- Human beings have two genes for a trait; one passes down from the mother and the other from the father. If one of the genes inherited from the mother and father change, the person becomes a carrier and it continues throughout life.
- SMA is inherited autosomal recessively. It occurs as a result of survival motor neuron (SMN) protein deficiency. *The SMN1 gene encodes the SMN protein.*
- Individuals with SMA have two non-functioning copies of the SMN1 gene. Due to the genetic features of the disease, one of the non-functioning SMN1 gene copies is inherited from the father and the other from the mother in 95% of the patients. Parents of children with SMA are carriers and do not have the disease. In very rare cases (2%), SMA disease may occur as a result of spontaneous (de novo) errors.
- The genetics of SMA disease are quite complex. For this reason and depending on the screening method applied, the screening test may give false-negative information in terms of carriage at a rate of 5%.

- **The carrier frequency of SMA in the community varies between 1/40-1/60. <u>On average, one out of every 50 individuals in the community is a carrier for SMA disease.</u>**
- **Two individuals who are coincidental SMA carriers are more likely to marry.**
- **For each pregnancy, the children of the father and mother who are carriers of SMA disease;**
- 25% will have SMA disease.
- 50% are carriers.
- 25% do not carry the disease and are healthy.



- **In particular, <u>consanguineous marriages</u> increase the risk for hereditary diseases due to the high probability of carrying similar diseases within the same family.**