Support for Individuals Who Are Carriers of Spinal Muscular Atrophy: Genetic, Psychological and Social Approaches

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Received: December 28, 2024 **Accepted:** Dec 30, 2024 **Published:** December 31, 2024. **DOI: 10.5281/zenodo.14582724**

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Cite This Article as: Yanmaz G, Çakır OK. Support for Individuals Who Are Carriers of Spinal Muscular Atrophy: Genetic, Psychological and Social Approaches. Essentials Frontline Med J. 2024:1(1); 31-34.

Externally Peer-Reviewed.

ABSTRACT

Objective: Spinal muscular atrophy (SMA) is an autosomal recessive motor neuron disease with an estimated incidence of 1 in 10,000 live births and a carrier frequency ranging from 1/40 to 1/60. Carrier detection relies on genetic screening, highlighting the need for comprehensive support systems. This article evaluates support models for SMA carriers from a multidisciplinary perspective, emphasizing the critical roles of genetic counseling, psychological well-being, and social awareness initiatives.

Methods: A review of multidisciplinary approaches was conducted to assess the effectiveness of genetic counseling, psychological support frameworks, and social service models tailored to SMA carriers.

Results: The findings underline the importance of integrating genetic counseling into carrier detection processes. Furthermore, psychological support and social awareness programs are pivotal in addressing the broader impacts on carriers and their communities.

Conclusion: A multidisciplinary approach is essential to provide holistic support for SMA carriers. Genetic counseling, combined with psychological and social support, enhances the overall well-being and preparedness of affected individuals and families.

Keywords: Spinal muscular atrophy, spinal muscular atrophy carrier, diagnostic screening programs, social support, genetic counseling

INTRODUCTION

Spinal muscular atrophy (SMA) is a progressive neuromuscular disease caused by mutations in the SMN1 gene (1, 2). SMA is a disease characterized by widespread muscle weakness and atrophy in the proximal muscles (3). The clinical course of SMA is heterogeneous, ranging from a severe to a mild phenotype and is divided into 5 subclasses, from severe to mild: SMA Types 0, 1, 2, 3 and 4 (4). It is considered one of the most common genetically fatal diseases in childhood (1).

Rare diseases such as SMA have negative social, economic and psychological impacts on patients, families, carriers and society due to their devastating nature and have costly diagnostic and treatment processes (5). The primary way to mitigate the impact of rare diseases on society and government is to prevent their occurrence, if possible. This is achieved through two main methods: first is primary prevention through carrier screening, and second is secondary prevention through early detection screening (6).

While the worldwide carrier rate of SMA is approximately 2-3%, this rate can go up to 4-5% in countries with high rates of consanguineous marriages, such as Türkiye (7). In a study that estimated SMA carrier rate in Türkiye for the first time in 2022, the SMA carrier frequency in the Turkish population was found to be 1/27, which is higher than many other societies (8). Because of the high incidence rate and the severe course of the disease in some types, population-wide screening is recommended by the American College of Medical Genetics and Genomics (9).

Internationally, approaches to SMA screening vary considerably. Some countries have mandatory premarital carrier screening for SMA (Qatar), while others offer screening through government-supported health care plans (Israel, Australia) (10).

In Türkiye, SMA screening for individuals before marriage in primary care began in December 2021 (11). Screening tests are performed on individuals on an optional basis.

The aim of population-based SMA carrier screening is to identify couples at risk of having a child with SMA. Choosing to have SMA carrier testing done should be a conscious decision. The meanings of the couples' test are important. Because SMA is present in all populations, carrier screening should be offered to all couples regardless of race or ethnicity, but ideally to individuals who are premarital or planning a pregnancy (9). Counseling should be provided to everyone requesting the test and all risks of their children being carriers or the disease should be explained. In our country, where consanguineous marriages are common, knowledge and awareness about autosomal recessive genetic diseases with a high carrier rate, such as SMA, are becoming even more important.

A study of carrier screening in the general population in the United States found that approximately 60% of individuals seeking prenatal genetic counseling accepted SMA carrier testing. After the results were announced, it was seen that 98.7% of the patients were satisfied with continuing the test (12). Therefore, examining the genetic, psychological and social effects of SMA carriage on individuals is of critical importance to improve the quality of life of carriers.

2. GENETIC COUNSELING AND INFORMATION

2.1. Genetic Screening Process

Early detection of SMA carrier status enables couples to make informed reproductive decisions, reducing the risk of having individuals with SMA. The World Health Organization (WHO) recommends expanding genetic screening and supporting genetic counseling services. The best time to determine genetic risk is after clarification of carrier status and before pregnancy. Genetic counseling should be offered to individuals who are carriers or at high risk of being carriers.

SMA carrier screening is especially recommended for individuals before marriage or of childbearing age (12). It is suggested that earlier provision of genetic information and genetic screening programs could increase and expand the reproductive options of carrier couples and also reduce the number of births of children with genetic diseases (13). With the use of preconception screening, carrier couples can benefit from genetic technologies in their pregnancies and change their reproductive decisions to have a healthy child.

2.2. Reproductive Options for Carriers

Preimplantion Genetic Diagnosis (PGD): The pre-implantation stage is defined as testing embryos or oocytes for genetic defects. It is based on selecting healthy embryos before implantation (14).

Prenatal Diagnosis: The risk of SMA during pregnancy can be assessed through amniocentesis and chorionic villus sampling, and the decision of whether to continue the pregnancy can be left to the individual.

Use of Donors: Sperm or egg donation may be preferred to minimize genetic risks.

During genetic counseling, it is of great importance that individuals are informed, given freedom in decision-making processes, and privacy is protected (15). The decisions must be their own, but they can be helped to consider various factors that they may not have thought about.

3. Psychological Effects and Support Methods

Individuals who learn that they are SMA carriers often experience psychological effects such as anxiety, guilt, isolation and difficulty in making decisions (16). Uncertainties about the health of future children may cause anxiety in individuals. A study looking at the psychological impact of population-based carrier testing for cystic fibrosis found that carriers had a worse perception of their current health than non-carriers, despite being told that carrier status did not pose any disadvantage to their health (17).

In a study of the views of adults living with thalassemia and thalassemia carriers in the UK, the majority of the entire survey sample thought that carrier identity would influence people's choice of reproductive partners, suggesting strongly that carriers in particular are concerned about stigma (10).

Fear of stigma of being a carrier may increase feelings of loneliness. The mental burden of complex decisions about reproductive options can stress carriers. Surrogate couples, in particular, may experience intense emotional stress due to the uncertainty about their unborn child. It is of great importance to support carrier individuals to cope with these feelings.

Individual Psychotherapy: It can help individuals reduce anxiety and facilitate adaptation processes. Individual therapies can help carriers understand and manage their emotional processes. When applied together with genetic counseling, it can provide more holistic support to the individual.

Group Therapy: It helps to provide solidarity by communicating with individuals who have similar experiences. Carrier individuals can benefit from the solutions and experiences of others.

Family Counseling: Surrogacy can cause emotional and decision-making conflicts between couples. Family counseling can increase communication and understanding between spouses. It can help

individuals maintain their emotional balance during decision-making processes. Provides guidance to family members in the process of sharing and interpreting genetic information.

4. SOCIAL SUPPORT AND SOCIAL AWARENESS

4.1. The Role of Social Services

Social services facilitate access to health and psychological support services for carrier individuals. State-supported projects and the work of non-governmental organizations to raise SMA awareness in Türkiye are of great importance.

Non-governmental organizations have important roles, such as providing free genetic counseling and testing services for carriers, sustaining studies worldwide, and providing economic support or organizing donation campaigns for carrier families. Public institutions, such as the Ministry of Health, need to guide individuals regarding carrier screening and reproductive options.

4.2. Social Awareness Studies

Awareness campaigns targeting SMA carriers emphasize the importance of genetic screening and ensure that society develops a more conscious attitude towards genetic diseases. Increasing public awareness about genetic carrier status encourages early diagnosis and intervention. Campaigns should be targeted specifically at young individuals and couples in the pre-marital process.

Training programs can be organized for different segments of society such as health workers and teachers. These programs explain the scientific basis of SMA carriage and facilitate genetic counseling processes.

Informative content about SMA carrier status can be created through social media and digital platforms. In a study conducted in Türkiye, 197 individuals who were going to have a pre-marriage SMA carrier test were asked the question, "Where did you hear about SMA disease?" and 72.1% of the individuals answered "on social media" (18). According to these results, the role of social media is of great importance in terms of raising the correct awareness of individuals and society.

5. MULTI-DISCIPLINARY SUPPORT MODELS

Solving the genetic, psychological and social problems faced by carrier individuals requires a multidisciplinary approach. Collaboration between geneticists, psychologists and social workers increases the scope and effectiveness of support service.

6. THE ROLE OF FAMILY PHYSICIANS

In Türkiye, the SMA carrier screening program is implemented by primary health care institutions. Therefore, it is important that all primary health care workers, especially physicians, have sufficient knowledge about SMA and the related screening program and beyond. In a study, awareness of the screening program and subsequent genetic counseling was found to be higher in primary care than in secondary and tertiary care physicians (19). Studies emphasize that increasing the knowledge level of healthcare professionals through motivation and training plays a decisive role in the implementation of screening programs and counseling services (20).

Family physicians can inform individuals who are identified as SMA carriers about genetic screening tests and refer them to the relevant genetic specialist. They should help individuals address their concerns about fertility and having a healthy child. They should help individuals choose the most appropriate option by providing information about reproductive methods.

Since family physicians know the relationships of individuals with their families better than other physicians, they can support individuals by considering the problems they may experience in family relationships and their psychological and social effects as a whole.

In a study investigating the care and counseling services received by pregnant women, it was found that they received adequate care but counseling services were inadequate. It was observed that depression in pregnant women decreased with the care and counseling services they received (21). This study shows that primary care physicians who closely monitor pregnant women have a lot of work to do and that they should support individuals by increasing their counseling services.

They should inform individuals registered in their units, especially those of marriage and childbearing age, about the SMA screening program, emphasize its importance and guide them to make these tests widespread.

7. CONCLUSION AND RECOMMENDATIONS

SMA carrier status is not only a genetic reality, but also a multidimensional condition that affects the individual's quality of life, emotional state and social role. The effects of genetic information on individuals include not only biological but also psychological and social dimensions. Therefore, it is necessary to develop and expand services for carriers. In this regard, suggestions can be as follows:

- Making genetic counseling services more accessible.
- Expansion of psychological support services.

- Organizing training and campaigns to raise SMA awareness throughout society.
- Expansion of health policies to include SMA carriers.
- Increasing the continuous education of healthcare personnel.
- More effective promotion of screening programs
- To ensure more effective and conscious use of social media and digital platforms

DECLARATIONS

Contributions: All authors were involved in the conceptualization and design of the study. MB, SK, and DA conducted the search and performed the video evaluation and data extraction. MAN wrote the drafts of the manuscript and all other authors critically revised the manuscript. All authors read and approved the final manuscript.

Financial Support: No funding or support was received from any institution or organization for this study.

Conflict of Interest Statement: There are no potential conflicts of interest in this study.

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