ATATÜRK ÜNİVERSİTESİ YAYINLARI
ATATURK UNIVERSITY PUBLICATIONS

Review *Derleme* DOI: 10.62425/esbder.1497446

Challenges Faced By Families of SMA Patients

SMA Hastalarının Ailelerinin Karşılaştığı Zorluklar

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ABSTRACT
SMA, a genetic neuromuscular disease that affects the control of muscle movement and results in severe motor disorders, is among the rare diseases. Due to the low prevalence of rare diseases and serious problems with correct diagnosis, there may be delays in diagnosis. When the studies on SMA are examined, it is known that the issues related to diagnosis are mostly emphasised, but a limited number of studies have been conducted on the current issues of patients with SMA or their caregivers. In addition to studies on treatment, it would be useful to consider the patient and his/her environment together in studies on the quality of life of patients and caregivers. Examining the concept of quality of life in SMA disease will form the basis for studies on quality of life, and at the same time, the continuity of studies on the subject will be ensured. In this sense, current issues the treatment of patients diagnosed with SMA was addressed in this study. The fact that these patients experience significant deficiencies such as respiratory impairment, malnutrition and skeletal deformity causes them to face difficulties in meeting their basic needs, especially in nutrition and toileting. These problems reduce the quality of life of patients and their relatives. Therefore, early diagnosis and providing medical, psychological, and social support to patients and their relatives will be effective in their quality of life. In this review, recommendations were made to address the current issues of SMA patients and their relatives.
Keywords: Genetic neuromuscular disease, spinal muscular atrophy, types of spinal muscular atrophy, treatment

ÖZ
Kas hareketinin kontrolünü etkileyen ve ciddi motor bozukluklarla sonuçlanan genetik bir nöromüsküler hastalık olan SMA, nadir görülen hastalıklar arasında yer alıyor. Nadir hastalıkların görülme sıklığının düşük olması ve doğru tanıda ciddi sorunlar yaşanması nedeniyle tanıda gecikmeler yaşanabilmektedir. SMA ile ilgili yapılan çalışmalar incelendiğinde çoğunlukla tanıya ilişkin konuların vurgulandığı ancak SMA hastası veya bakım verenlerin güncel sorunlarına yönelik sınırlı sayıda çalışmanın yapıldığı bilinmektedir. Tedaviye yönelik yapılan çalışmaların yanında hastaların ve bakım verenlerin yaşam kalitesine yönelik çalışmalarda hasta ve çevresinin birlikte ele alınmasının yararlı olacaktır. SMA hastalığında yaşam kalitesi kavramının ele alınarak incelenmesi, yaşam kalitesi ile ilgili çalışmalara temel oluşturacak, aynı zamanda konu ile ilgili çalışmaların sürekliliği sağlanacaktır. Bu anlamda bu çalışmada SMA tanısı alan hastaların tedavisindeki güncel konulara değinilmiştir. Bu hastaların solunum yetmezliği, yetersiz beslenme ve iskelet deformitesi gibi önemli eksiklikler yaşaması, beslenme ve tuvalet başta olmak üzere temel ihtiyaçlarının karşılanmasında zorluk yaşamalarına neden olmaktadır. Bu sorunlar hasta ve yakınlarının yaşam kalitesini düşürmektedir. Bu nedenle erken teşhis ve hasta ve yakınlarına tıbbi, psikolojik ve sosyal destek sağlanması yaşam kaliteleri üzerinde etkili olacaktır. Bu derlemede, SMA hastalarının ve yakınlarının güncel sorunlarına yönelik önerilerde bulunulmuştur.
Anahtar Kelimeler: Genetik nöromüsküler hastalık, spinal müsküler atrofi, spinal müsküler atrofi çeşitleri, tedavi

Geliş Tarihi/Received 07.06.2024
Kabul Tarihi/Accepted 01.10.2024
Yayın Tarihi/Publication Date 09.10.2024

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Cite this article: Aysu, B., Yılmaz, S., Aral, N., & Aydoğdu, F. (2024). Challenges Faced By Families of Sma Patients. *Journal of Midwifery and Health Sciences, 7*(3), 541-547.

Introduction

SMA, a genetic neuromuscular disease that affects the control of muscle movement and results in severe motor disorders, is among the rare diseases. Due to the low prevalence of rare diseases and serious problems with the correct diagnosis, there may be delays in their diagnosis. Most of the such diseases either have no treatment or no effective treatment can be applied. For this reason, the quality of life in individuals trying to live with this disease may be at a low level (Peña-Longobardo et al., 2020; Parker, 2007; Oudgenoeg-Paz & Rivière, 2014).

The first clinical description was made by Dr Guido Werdnig in 1891, after which the broad clinical spectrum of SMA was recognised with varying degrees of detailed clinical and anatomical description. Since the first diagnosis of SMA, great progress has been made in understanding its pathophysiology. The identification of the molecular basis of SMA in 1995 and the establishment of animal models are considered as an important step (Oskoui et al, 2017).

When the studies on SMA are examined, it is seen that the issues related to treatment (Barkats, 2020; Eisenkölbl, 2021; Glowinski & Blazejewski, 2020; Kim et al., 2020; Mercuri et al., 2020; Nizzardovd., 2015) and diagnosis (Canary et al., 208; Lin et al., 2015) are mostly emphasised. However, it is found that there is limited study on the quality of life of patients with SMA or their caregivers (Lloyd et al., 2019; Vaidya & Boes, 2018; Vega et al., 2020; Peña-Longobardo, 2020). SMA, one of the rare diseases, is an issue that should be emphasised (Peña-Longobardo et al., 2020). Quality of life is important in the treatment of patients diagnosed with SMA (Vega et al., 2020) and health-related quality of life measurements should be followed regularly (Mercuri et al., 2020). It is thought that it would be useful to expand the studies on the quality of life of patients and caregivers and to address the patient and his/her environment together in addition to the studies on treatment. For this reason, examining the concept of quality of life in SMA disease will provide a basis for studies on quality of life and will also contribute to the continuity of studies on the subject.

Spinal Muscular Atrophy

This condition results from the degeneration of specific nerve cells known as "motor neurons" located in the spinal cord and brain stem, which is the region of the brain linked to the spinal cord. (Parker, 2007). The symptoms include pronounced muscle weakness caused by the breakdown of nerve cells in the front part of the spinal cord. The progression of the disease varies significantly depending on the individual. In this disease, proximal limb muscles are affected more than distal muscles and lower limb muscles are affected more than upper muscles (Oudgenoeg-Paz & Rivière, 2014). Loss of motor neurons leads to weakness and shrinkage (atrophy) of muscles used for activities such as crawling, walking, sitting, and controlling head movement. In severe cases of SMA, the muscles used for breathing and swallowing are also affected (Parker, 2007). This condition can be caused by a microdeletion on chromosome 5q13, i.e., a genetic defect resulting from the deletion of part of the gene sequence (Oudgenoeg-Paz & Rivière, 2014). The genetic defect is on the long arm of chromosome 5, which is why this disease is also called 5q-related SMA. Inheritance is autosomal recessive (Eisenkölbl, 2021). In the normal population, the carrier frequency is 1 in 35 to 40, which means that approximately 1 in 6,000 to 10,000 newborns are affected (Eisenkölbl, 2021).

Classification

SMA is classified according to the age of onset of symptoms, which is based on the age of onset, developmental milestones, and life span (Oudgenoeg-Paz & Rivière, 2014). The later the age of onset, the slower the progression. The earlier the age of onset, the higher the likelihood of bulbar motor neuron involvement and respiratory adiposity in addition to signs of anterior hom cell involvement. The types vary depending on the age of onset (Marcus & Jacobson, 2012).

SMA is seen as a total of five types (0, I, II, III and IV). Three types of this disease are known to affect children before the age of one year. Type 0 begins before birth and presents as a very severe form of spinal muscular atrophy. The first sign of type 0 is a decrease in fatal movement, which is noticed between 30 and 36 weeks of gestation (Parker, 2007). The infantile form, type I, is called "Werdnig-Hoffinan Disease". Type I is a severe form of disease that is evident at birth or in the first few months of life. In Type I, newborn babies move very little after birth, have difficulty in swallowing and breathing, and cannot sit without support (Marcus & Jacobson, 2012; Parker, 2007). Although SMA Type I is the most frequently diagnosed subtype, its overall prevalence is the lowest due to its high mortality rate. Infants with SMA Type I (also known as Werdnig-Hoffmann disease) typically exhibit early symptoms of muscle weakness in the legs rather than the arms within the first six months of life. These infants are unable to roll over independently and never achieve the ability to sit without support. They often develop a chest with a bell shape due to weak intercostal muscles and exhibit paradoxical breathing using the diaphragm. Over the first year of life, bulbar weakness progresses, leading to increased feeding difficulties as the

lower cranial nerves become affected. These babies take longer to feed. They are also at risk of aspiration pneumonia and growth retardation. Cognition is normal. Infants with the onset of symptoms in the first week of life are classified as SMA Type I (sometimes referred to as SMA Type 0 when symptoms begin before birth) and die soon after birth (in less than 1 month) (Oskoui et al, 2017). Eisenkölbl (2021) emphasised in his study that SMA I in the infantile form usually leads to death within the first 24 months of life.

Type II is referred to as a "moderate/retained form of spinal muscular atrophy" and is observed at 18 months and before. In Type II, the infant can sit without support but cannot stand or walk without support (Marcus & Jacobson, 2012). These children present to the hospital with proximal weakness, hypotonia and areflexia affecting the legs rather than the arms (Oskoui et al., 2017).

Type III is defined as a juvenile form or "Kugelberg Welander Disease" and is milder than Type 0, I or II (Marcus and Jacobson, 2012; Parker, 2007). Type III is considered to start after 18 months in some sources, usually between 5-15 years of age (Marcus & Jacobson, 2012), and in some sources, it is accepted to occur between early childhood (older than 1 year) and early adulthood (Parker, 2007). In Type III, the child can sit and walk, but the need for support emerges with advancing age. The child usually loses these abilities later in life (Marcus & Jacobson, 2012; Oskoui et al, 2017).

Type IV is defined as "adult-onset spinal muscular atrophy" and is seen in two forms as Type IV and Finkel type. Type IV is a very rare form that starts in adulthood and usually occurs after the age of 30 (Marcus & Jacobson, 2012; Parker, 2007). Symptoms of type IV are typically mild to moderate and include muscle weakness, tremor, and twitching (Parker, 2007). In 75% of patients, muscle twitching (fasciculation) is associated with occasional muscle cramps (Oskoui et al., 2017).

Spinal muscular atrophy types (0, I, II, III, and IV) are inherited through autosomal recessive patterns. Typically, parents of individuals with autosomal recessive disorders carry one altered gene copy each without displaying symptoms. In contrast, Finkel-type spinal muscular atrophy follows an autosomal dominant inheritance pattern, where having one altered gene copy in each cell is enough to cause the disorder (Parker, 2007). Pérez-García et al. (2017) noted the lack of longitudinal pathological data in SMA patients and suggested further research to identify pathological changes in all cell types and problems affected by SMA. Further research is considered necessary to develop new SMA therapies that will be successful (Pérez-García et al., 2017).

Newborn screening for SMA is of great interest because it allows early diagnosis and treatment. The ideal time to initiate treatment is before the first degeneration of motor neurons. Thus, newborn screening can also help identify non-symptomatic individuals (Mercuri et al., 2020). Therefore, studies for SMA show the importance of starting treatment as early and pre-symptomatic as possible and including SMA in newborn screening. Newborn screening ensures a pre-symptomatic treatment initiation and thus the best possible therapeutic success (Eisenkölbl, 2021). Vill et al. (2021) reported that genetic newborn screening enabled the identification of newborns with infantile SMA and rapid treatment specific to SMA. Çankaya (2010) also emphasised that prenatal diagnosis is the most appropriate approach for SMA. Early diagnosis of SMA contributes to early supportive care and reduction in patient and carer stress (Lin et al., 2015). Identifying infants before the period of the greatest motor neuron loss enables early treatment to begin. In addition, it is necessary to carry out studies that will help each child affected by SMA to reach their strengths and to inform families about the multidisciplinary care of these children (Oskoui et al., 2017). In this process, it is emphasised that there should be a short time interval between the screening result and referral to a treatment centre ready for treatment (Vill et al., 2021).

Difficulties experienced by patients with SMA and their families

While SMA mainly impacts voluntary muscle strength, its resulting complications are numerous. These include impaired breathing, malnutrition, skeletal deformities, and other issues. Minimizing these complications is crucial for SMA patients. Effectively managing these complications involves addressing medical, psychological, and social aspects, making it a multifaceted endeavor. Identifying and bringing together existing knowledge about quality care can improve the quality of life of patients with SMA and their families (Crawford, 2017).

The complexity of medical care requires the recognition that not only the child with SMA but also the stakeholders around the child with SMA are affected by this diagnosis. These stakeholders can be expressed as parents, siblings, other family members, therapists, teachers, doctors, nurses, health centres and hospitals, and private and public funding sources. Physicians should think about the therapies to be applied to the individual with SMA primarily from the perspective of their patients and take into account the concerns of stakeholders because these stakeholders indirectly affect patients (Crawford, 2017). When designing and evaluating any strategy or intervention for patients with SMA, the economic impact of new treatments in this field

should also be considered (Peña-Longobardo et al., 2020). Specific policies are needed to support families who must live with the disease, not only in terms of their deteriorating quality of life but also because of the significant economic burden that the disease brings (Marcellusi et al., 2019). Peña-Longobardo et al. (2020) emphasised the importance of family support or non-medical assistance. Michalík (2014) stated that there is also a need to focus on better support for caregivers of a child with SMA because long-term care of a child with a serious, incurable disease implies a significant change in life situation, involving not only personal growth but also social, partner, health, and economic aspects (Michalík, 2014). SMA has non-health costs such as hospitalisations, emergencies, medical tests, medications, visits to general practitioners and specialists, transport of medical supplies and healthcare, and social and informal care (Peña-Longobardo et al., 2020). Peña-Longobardo et al. (2020) included 86 children with SMA, of whom 26.7% were Type I and 73.3% were Type II or III in their study and found that the average annual cost associated with SMA reached €54,295 in England, €32,042 in France and €51,983 in Germany. In the same study, direct non-health costs vary between 79-86 per cent of total costs and informal care costs are the main component of these costs. SMA, therefore, has a high socioeconomic impact in terms of health and social costs. It has also been observed that the quality of life of affected children is extremely reduced. When the hours of care were analysed, it was found that informal carers in England provided an average of 12.5 hours of care per day, while people in France and Germany provided 10.65 and 9.31 hours of care per day, respectively. Although the intensity of caring was lower in France (compared to England and Germany), the burden of caring was found to be higher. Chambers et al. (2020) found that the average total indirect healthcare costs for all types of SMA were $63,145 per year and the families of children diagnosed with SMA II were the most affected families. It was determined that three out of four caregivers (78%) experienced economic problems due to their care duties. Belter et al. (2021) found that those affected by a less severe form of SMA and those with higher functional status had a higher quality of life.

When assessing the disease burden, the impact of conventional care on children with SMA is evaluated across three dimensions: life expectancy, the pain and suffering linked to the disease and its complications, and the ability to work independently. These dimensions have distinct criteria that do not overlap, but the latter two—pain and suffering related to the disease and its complications, and the capacity for independent work—are crucial factors determining quality of life. Many therapies, in themselves, serve only one of these goals, because some therapies pit one of these goals against the other and can improve one while worsening the other. For example, mouth and throat suctioning is an uncomfortable but life-prolonging procedure. It is the caregiver's responsibility to consider the balance between burden and benefit for any intervention and to reconsider frequently at each point. Since the criteria for each of these three goals for standardised care are so different, an objective assessment of the net balance is often impossible. Given all dimensions of complexity, those with individual experience, i.e., medical and family carers and patients alike, can have very different views on the value of any intervention (Crawford, 2017).

It is known that even typically developing children need full support for care such as feeding and toileting. Therefore, it is important to investigate the activities of daily living and care burden in the first year of life in infants diagnosed with SMA. For this purpose, conducting studies on how the need for care and the level of independence change with increasing age in typically developing infants may help to determine the direction of research on children diagnosed with SMA. This will be even more important considering the fact that clinical trials and new treatments increasingly target pre-symptomatic infants, and a detailed follow-up will be required (Mercuri et al., 2020).

As a result of the study conducted by Fischer et al. (2021), it was found that the agreement between parents who serve as the primary caregivers of children with SMA and their children regarding their perceptions of the disease is weak and that parents perceive the severity of SMA more than their children. In other words, parents think that SMA has a greater impact on their children compared to their children. Weaver et al. (2020) found that the quality of life of children was higher than that of caregivers. When children's perceptions of their own illness are negative, there may be a decrease in their quality of life scores. In line with the strong relationship between children's illness perceptions and quality of life, it is emphasised that health professionals should evaluate the child's illness perceptions and change them if necessary (Fischer et al., 2021).

Yao et al. (2021) found that exercise training and multidisciplinary team management improved the quality of life in SMA. In particular, children with type I and type II SMA, as well as caregivers, had a lower quality of life compared to those with type III SMA. Strengthening the standard of care in a multidisciplinary team was recommended to improve the quality of life of SMA patients, and it was stated that quality of life should be emphasised in clinical practice to

improve understanding of the effects of SMA and to make better treatment decisions. Glowinski & Blazejewski (2020) found that the SPIDER device, which was developed as a rehabilitation tool that controls changes in the centre of gravity in patients with neurological disabilities including SMA and can make continuous adjustments during any patient's movement, significantly improved movement, balance and coordination ability, gait and mobility, and reduced motor function and fall risk after rehabilitation. In addition, exercise is also very important in patients with SMA. However, it is recommended that personalised, patient-specific interventions should be used to see the benefits of exercise (Houdebine et al., 2019). Salem & Jaffee Gropack (2010) applied aquatic therapy to a 3-year-old girl with type III SMA twice a week with 45-minute sessions for 14 weeks. The intervention included water activities designed to improve gross motor skills and age-appropriate functional mobility. As a result of the study, improvement was found in the child's gait, walking speed, and step length. In a study by Vega et al. (2020), it was found that the quality of life of children with SMA with higher motor function was higher.

Recommendations

In line with the results obtained from the studies, the following can be suggested:

- Developing policies to support families socially and economically,
- Carrying out studies for early diagnosis of SMA and disseminating existing diagnostic methods
- Making evaluations according to the type of SMA the child has while developing applications for quality of life,
- Involving the child's stakeholders (parents, siblings, other family members, therapists, teachers, doctors, nurses, health centres and hospitals, and private and public funding sources) in programmes designed to improve the quality of life of children with SMA.
- Conducting more studies on current issues in children and families diagnosed with SMA.

Peer-review: Externally peer-reviewed.
Author Contributions: Concept - FA-BA; Design- FA-BA; Supervision- SY-NA; Resources- FA-BA ; Data Collection and/or Processing- FA-BA; Analysis and/or Interpretation-SY-NA; Literature Search- FA-BA; Writing Manuscript- FA-BA ; Critical Review- SY-NA
Conflict of Interest: The authors have no conflicts of interest to declare.
Financial Disclosure: The authors declared that this study has received no financial support.
Hakem Değerlendirmesi: Dış bağımsız.
Yazar Katkıları: Fikir- FA-BA; Tasarım- FA-BA; Denetleme- SY-NA; Kaynaklar- FA-BA; Veri Toplanması ve/veya İşlemesi FA-BA; Analiz ve/veya Yorum- SY-NA; Literatür Taraması- FA-BA; Yazıyı Yazan- FA-BA; Eleştirel İnceleme- SY-NA
Çıkar Çatışması: Yazarlar, çıkar çatışması olmadığını beyan etmiştir.
Finansal Destek: Yazarlar, bu çalışma için finansal destek almadığını beyan etmiştir.

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Genişletilmiş Özet

Çocuklarda görülen nadir hastalıklardan biri de spinal müsküler atrofidir. Bu hastalığın tedavisi güçtür. Tedavisi olsa bile yaşam kalitesinde veya yaşam beklentisinde sorunlar yaşanmaktadır. Spinal müsküler atrofi (SMA), motor nöronların kaybına bağlı olarak kas zayıflığı ve atrofi ile ortaya çıkmaktadır. Kas hareketinin kontrolünü etkileyen, şiddetli motor bozukluklarla sonuçlanan genetik nöromüsküler bir hastalık olan SMA nadir hastalıklar arasında yer almaktadır. Nadir hastalıkların prevalanslarının düşük olması, doğru teşhiste ciddi sorunlar yaşanması nedeniyle tanı ve teşhiste gecikmeler yaşanabilmektedir. İlk klinik tanımlama 1891'de Dr. Guido Werdnig tarafından yapılmıştır ve ardından SMA'nın geniş klinik spektrumu, çeşitli derecelerde ayrıntılı klinik ve anatomik tanımlamalarla tanınmıştır. SMA'nın ilk teşhisinden bu yana, patofizyolojisinin anlaşılmasında büyük ilerleme kaydedilmiştir. 1995'te SMA'nın moleküler temelinin tanımlanması ve hayvan modellerinin oluşturulması önemli bir adım olarak kabul edilir.

SMA, omurilikte ve omuriliğe bağlı beyin bölgesi olan beyin sapında bulunan "motor nöronlar" olarak bilinen belirli sinir hücrelerinin dejenerasyonundan kaynaklanır. Semptomlar arasında omuriliğin ön kısmındaki sinir hücrelerinin bozulmasından kaynaklanan belirgin kas güçsüzlüğü yer alır. Hastalığın ilerlemesi bireye bağlı olarak önemli ölçüde değişir. Bu hastalıkta, proksimal uzuv kasları distal kaslardan daha fazla etkilenir ve alt uzuv kasları üst kaslardan daha fazla etkilenir. Motor nöronların kaybı, emekleme, yürüme, oturma ve baş hareketini kontrol etme gibi aktiviteler için kullanılan kasların zayıflığına ve küçülmesine yol açar. Şiddetli SMA vakalarında, nefes alma ve yutma için kullanılan kaslar da etkilenir Bu durum kromozom 5q13'teki bir mikrodelesyondan, yani gen dizisinin bir kısmının silinmesinden kaynaklanan genetik bir kusurdan kaynaklanabilir. Genetik kusur kromozom 5'in uzun kolundadır, bu nedenle bu hastalığa 5q ile ilişkili SMA da denir. Kalıtım otozomal resesiftir. Normal popülasyonda taşıyıcı sıklığı 35 ila 40'ta 1'dir, bu da yaklaşık 6.000 ila 10.000 yenidoğanda 1'inin etkilendiği anlamına gelir. SMA, semptomların başlama yaşına göre sınıflandırılmakta olup bu sınıflandırma başlangıç yaşına, gelişimsel dönüm noktalarına ve yaşam süresine dayanmaktadır. SMA'nın temel olarak beş tipi bulunmaktadır: Tip 0, Tip I, Tip II, Tip III ve Tip IV. Tip 0, doğumdan önce başlamakta ve çok şiddetli bir spinal müsküler atrofi şekli olarak ortaya çıkmaktadır. Tip I, bozukluğun doğumda veya yaşamın ilk birkaç ayında belirgin olan ciddi bir şeklidir. Tip I'de doğumdan sonra yeni doğan bebekler çok az hareket etmekte, yutma ve nefes almada güçlük çekmekte, desteksiz oturamamaktadır. Tip II, 18 ay ve öncesinde görülmektedir. Bebek, Tip II'de desteksiz oturabilmekte ancak desteksiz ayakta duramamakta veya yürüyememektedir. Tip III'de çocuk oturabilmekte, yürüyebilmekte ancak ilerleyen yaşla birlikte destek ihtiyacı ortaya çıkmaktadır. Tip IV erişkin yaşta başlayan, genellikle 30 yaşından sonra ortaya çıkan çok nadir bir formdur. SMA birincil olarak yalnızca bir işlevi yani istemli kas gücünü etkilese de kas zayıflığının aşağı yönlü komplikasyonları çok fazladır. Bunlara solunum bozukluğu, yetersiz beslenme, iskelet deformiteleri ve diğer sorunlar dahildir. Tipik olarak gelişen çocukların bile beslenme ve tuvalet gibi bakım için tam desteğe ihtiyaç duyduğu bilinmektedir. Bu nedenle, SMA teşhisi konan bebeklerde yaşamın ilk yılında günlük yaşam aktiviteleri ve bakım yükünü araştırmak önemlidir. Bu amaçla, tipik olarak gelişen bebeklerde bakım ihtiyacının ve bağımsızlık düzeyinin artan yaşla nasıl değiştiğine dair çalışmalar yürütmek, SMA teşhisi konan çocuklarla ilgili araştırmanın yönünü belirlemeye yardımcı olabilir. Klinik denemelerin ve yeni tedavilerin giderek daha fazla semptom öncesi bebekleri hedef alması ve ayrıntılı bir takip gerekmesi gerçeği göz önüne alındığında bu daha da önemli olacaktır. Bu komplikasyonları en aza indirmek SMA hastaları için çok önemlidir. Bu komplikasyonları etkili bir şekilde yönetmek, tıbbi, psikolojik ve sosyal yönleri ele almayı içerir ve bu da onu çok yönlü bir çaba haline getirir. Bu nedenle kaliteli bakım hakkında mevcut bilgileri belirlemek ve bir araya getirmek, SMA'lı hastaların ve ailelerinin yaşam kalitesini iyileştirebilir. Tıbbi bakımın karmaşıklığı, yalnızca SMA'lı çocuğun değil, aynı zamanda SMA'lı çocuğun etrafındaki paydaşların da bu tanıdan etkilendiğinin kabul edilmesini gerektirir. Bu paydaşlar ebeveynler, kardeşler, diğer aile üyeleri, terapistler, öğretmenler, doktorlar, hemşireler, sağlık merkezleri ve hastaneler ve özel ve kamusal fon kaynakları olarak ifade edilebilir. Doktorlar, SMA'lı bireye uygulanacak terapileri öncelikle hastalarının bakış açısından düşünmeli ve paydaşların endişelerini dikkate almalıdır. SMA'nın hastaneye yatışlar, acil durumlar, tıbbi testler, ilaçlar, pratisyen hekim ve uzman ziyaretleri, tıbbi malzeme ve sağlık hizmetlerinin taşınması, sosyal ve gayri resmi bakım gibi sağlık dışı maliyetleri vardır.Sadece yaşam kalitelerinin bozulması açısından değil, aynı zamanda hastalığın getirdiği önemli ekonomik yük nedeniyle de hastalıkla yaşamak zorunda kalan aileleri desteklemek için özel politikalara ihtiyaç vardır.

Sonuçlar doğrultusunda; aileleri sosyal ve ekonomik olarak desteklemek için politikalar geliştirilmesi, SMA'nın erken tanısı için çalışmalar yapılması ve mevcut tanı yöntemlerinin yaygınlaştırılması, yaşam kalitesine yönelik uygulamalar geliştirilirken çocuğun sahip olduğu SMA tipine göre değerlendirmeler yapılması, SMA'lı çocukların yaşam kaliteleri geliştirmek üzere hazırlanan programlara çocuğun paydaşların da (ebeveynler, kardeşler, diğer aile üyeleri, terapistler, öğretmenler, doktorlar, hemşireler, sağlık merkezleri ve hastaneler, özel ve kamu finansman kaynakları) dâhil edilmesi ve SMA tanısı alan çocuklar ve ailelerde yaşam kalitesi ile ilgili daha fazla çalışma yapılması önerilmektedir.