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Review Article

Molecular Mechanisms Causing SMA Pathology

Aydan I. Dadashova[1] ✉ **and Mehraj A. Abbasov**[2] ✉

[1]Department of Natural Sciences, School of Advanced Technologies and Innovation Engineering, Western Caspian University, 17 A, Ahmad Rajabli Street, III Parallel, AZ1072 Baku, Azerbaijan
[2]School of Agricultural and Food Sciences, ADA University, 61 Ahmadbay Aghaoglu Street, Narimanov District, AZ1008 Baku, Azerbaijan

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Abstract

Disorders in genes and chromosomes lead to various genetic diseases. These diseases can be inherited from parents to future generations or arise from mutations that form spontaneously in genetic material. Researching genetic diseases is extremely important because it helps to understand the mechanisms that cause the disease and to provide effective interventions. As a result, diagnostic methods are being improved and targeted treatments and preventive measures are being developed. One of the crucial genetic diseases is Spinal Muscular Atrophy (SMA). SMA has a significant impact on quality of life, causing severe physical disabilities and, in some cases, life-threatening complications. The study of SMA also serves as a model for understanding other neurodegenerative and genetic diseases.

Keywords: spinal muscular atrophy, SMN1 and SMN2 genes, SMN protein, splicing mechanism, alternative splicing

1. Introduction

Spinal muscular atrophy (SMA) is a devastating genetic disorder that primarily affects motor neurons in the spinal cord. This neuromuscular disease results in progressive muscle wasting and muscle weakness. Spinal muscular atrophy is considered one of the most common inherited causes of child mortality worldwide. The disease is caused by mutations in the survival motor neuron (SMN) genes, namely SMN1 and SMN2. Mutations in the survival motor neuron (SMN) genes, namely SMN1 and SMN2, are the main cause of the disease. Knowledge about the processes that cause SMA has increased significantly in recent years, providing insight into the complex interplay of cellular processes that underlie the pathogenesis of the disease. This article examines the roles of the SMN1 and SMN2 genes, RNA splicing processes, and molecular genetic mechanisms involved in the pathogenesis of SMA [1], [2].

2. General Information About SMA

Mutations or deletions in the SMN1 (survival motor neuron) gene, located on chromosome 5q13, are the main cause of the genetic disease spinal muscular atrophy (SMA). Motor neurons are nerve cells in the spinal cord that allow impulses to be sent from the spinal cord to the muscles. The deficiency of the SMN protein, which is essential for the structure and function of motor neurons, is caused by mutations in the SMN gene. Insufficient and progressive loss of SMN protein results in motor neuron (synaptic) dysfunction and degeneration, resulting in muscle atrophy and weakness [1], [2]. Approximately one in 10,000 newborns worldwide is affected by SMA. SMA is thought to be one of the most common hereditary causes of neonatal death. SMA has an autosomal recessive inheritance pattern. Accordingly, the disease can only be manifested in an individual who inherits two copies of the mutant SMN1 gene, one from each parent. Carriers have one mutant and one normal copy of the

SMN1 gene. Studies suggest that about 1 in 40 to 60 people carry the defective SMN1 gene. Carriers can pass the defective gene on to their offspring, but they do not show symptoms of the disease. The symptoms of SMA vary depending on the type and severity of the disease. There are 5 types of SMA (Type 0, I, II, III, IV) [3], [4], [5].

The disease is usually caused by a homozygous deletion. Either exons 7 and 8 are deleted together, or exon 7 alone is deleted. In the remaining cases, small intragenic and de nova mutations have been identified [5], [6]. Both SMN1 and SMN2 genes are located on chromosome 5 in the region q11-13 [1], [2]. The SMN1 and SMN2 genes, which have a similar structure, are composed of 10 exons. Although the SMN gene was previously reported to consist of nine exons, recent studies have shown that it consists of ten exons. Exon 6b was discovered by exon fission of an Alu element in intron 6 [7]. Although the SMN1 and SMN2 genes are 99% identical (homology), there are 5 nucleotides that differentiate them. There are 3 differences in introns 6 and 7 in the non-protein-coding regions, and 2 differences in exons 7 and 8 in the coding regions. Mutations in the SMA gene cause a decrease in SMN protein levels. The severity of the disease phenotype is related to the copy number of the paralogous gene, SMN2, which produces a less stable form of the SMN protein. The milder symptoms of the disease are associated with a higher SMN2 copy number. The copy number of the SMN2 gene differs between the types of the disease, with fewer in Type 0 and more in Type 4 [8]. However, low levels of methylation of the SMN2 gene are associated with milder symptoms of the disease, as the disease may develop differently due to epigenetic processes that methylate DNA and silence the SMN2 gene. Studies have found that patients with different types of SMA have different levels of methylation at positions -296 and -290 in two CpG islands of SMN2 [9]. A hallmark of SMA pathology is aberrant splicing of SMN2 pre-mRNA transcripts. The C-to-T transition of exon 7 damages a potential exonic splicing enhancer (ESE). This is the main molecular abnormality responsible for SMA. The silent nucleotide exchange results in the replacement of a C in the SMN1 gene (TTC → TTT at position 280) with a T in the SMN2 gene. This region surrounding exon 7 is important because it causes a change in the resulting mRNA. In most SMN2 transcripts, the loss of SMN1 is not fully compensated for by a single nucleotide difference that causes exon 7 skipping. In alternative splicing, deletion of exon 7 in the SMN2 gene results in the production of a truncated and unstable SMN protein isoform. This isoform lacks critical functional domains and is termed SMNΔ7. This impairs its ability to participate in snRNP assembly and other cellular processes essential for the survival of motor neurons [10], [11], [12].

3. Factors Affecting the Regulation of Splicing

In general, several factors influence the regulation of SMN gene splicing: a combination of regulatory proteins, cis-acting elements, and trans-acting factors. To influence splicing, several proteins, including RNA-binding proteins and splicing factors, interact with specific sequences present in the pre-mRNA from SMN. For example, the SMN protein controls the splicing of its own transcripts by forming complexes with other splicing factors, such as gemin proteins. The two most common forms of RBP are heterogeneous ribonucleoprotein (hRNP) and serine/arginine-rich (SR) proteins. When RNA-binding proteins bind to the ISS or ESS, exon removal is promoted, and the formation of the spliceosome complex is inhibited. The same gene transcript can produce different end products depending on the tissue to which the RBPs bind. The different splicing mechanisms of these proteins result in different combinations of exons and introns [12]. SMN genes have certain sequences called cis-acting elements that act as binding sites for regulatory protein binding. These cis-acting components, which promote or prevent the inclusion of exon 7 in SMN2, include exon splicing enhancers (ESEs) and exon splicing silencers (ESSs). SMN splicing can also be influenced by RNA or proteins synthesized by other genes. These are trans-acting factors. For example, various physiological signals or environmental influences can affect the activity or expression level of splicing regulatory proteins.

The transition in the protein-coding region of the SMN2 gene results in the skipping of exon 7. The Tudor domain, an important component of the SMN protein that is involved in binding to various RNA and protein molecules, is encoded by exon 7. In the absence of exon 7 in SMNΔ7, the function of the Tudor domain is impaired. This, in turn, disrupts the protein's interaction with other molecular proteins. This loss of function is

associated with the reduced stability and activity of SMNΔ7 when compared to the full-length SMN protein produced by SMN1.

Deletion of exon 7 of the SMN2 gene occurs through alternative splicing. This results in the production of a less functional SMN protein. Some splicing factors influence the skipping of exon 7 [13], [14]. These include positive and negative regulatory splicing factors. For example, SRSF1 (serine/arginine-rich splicing factor 1) and Tra2B (transforming protein 2 homolog β) are positive regulatory factors. Both splicing factors bind to the enhancer regions of exon 7, called SE1 and SE2. In this case, exon 7 is not skipped. Conversely, exon 7 is spliced out when negative regulatory factors bind to SE1. Examples of these include heterogeneous nuclear ribonucleoprotein A1 (hnRNP A1) and the src-associated substrate in mitosis 68. Negative regulation may also occur through the binding of hnRNP A1 to the SE2 and N1 regions [15], [16], [17]. SMN1 expression is dominated by positive splicing regulation. Cooperative binding of hnRNP G and SRSF9 is enabled by TRA2B, further enhancing the beneficial effects of exon 7 recognition. The U1 and U2 snRNPs are therefore recruited to exon boundaries, and inclusion of this exon depends on recognition by this snRNP. The U1 and U2 snRNPs are attracted to exon boundaries for these reasons, and inclusion of this exon depends on recognition by this snRNP. SMN2 transcripts contain a C to U substitution in the SE1 region, which allows the negative regulator hnRNP A1 to bind to this site. SAM68 can bind to this site and exert a similarly damaging effect. SAM68 can bind to this site and exert a similarly damaging effect. It is known that there are two additional hnRNP A1 regions in the SE2 region and the intronic silencing element ISS-N1 that cause exon 7 exclusion. There are negative effector proteins that reduce the recognition of the exon by U1 and U2 snRNPs, leading to the exclusion of exon 7. Inclusion of exon 7 in both SMN1 and SMN2 transcripts is facilitated by the binding of hnRNP M to a region in SE2 that overlaps with the TRA2B binding site (Figure1) [8].

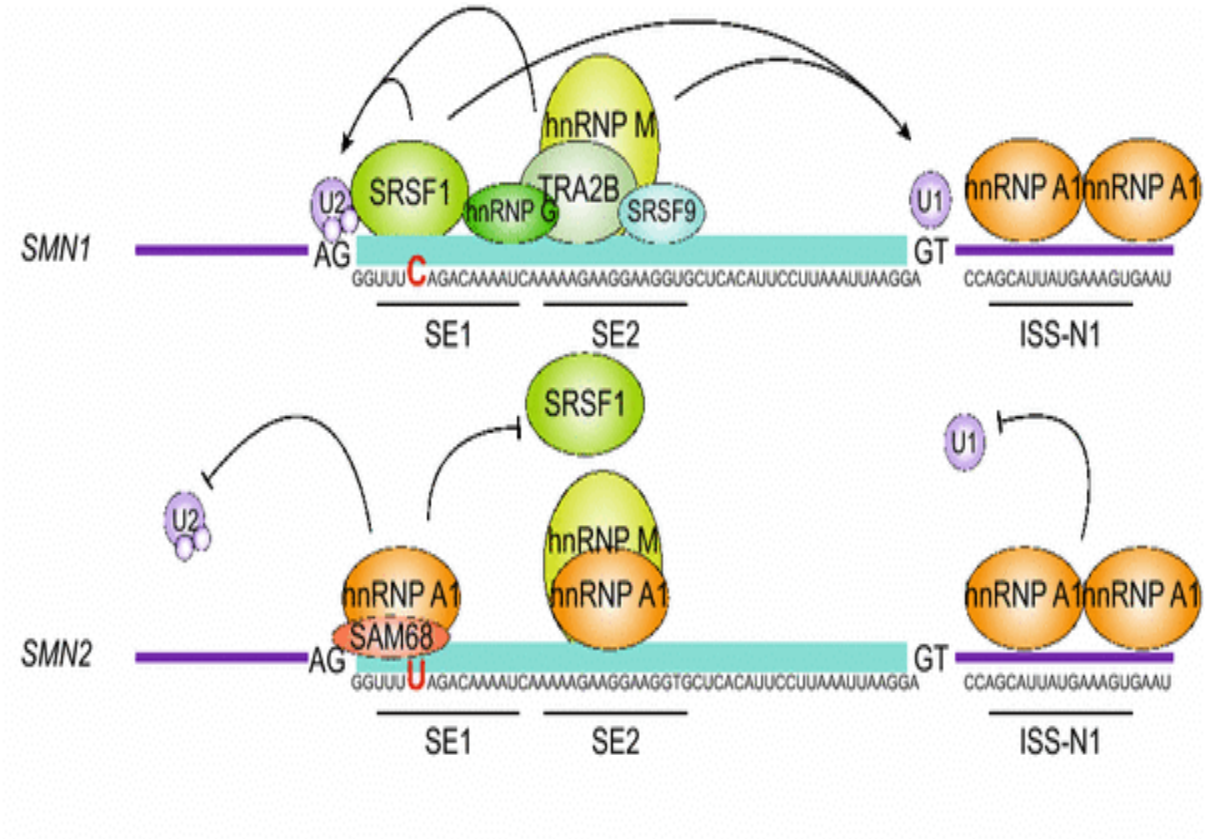

Figure 1. SMN1 and SMN2 exon 7 splicing regulators [8].

The SMN protein plays a significant role in snRNP formation and mRNA transport, which are essential for the proper functioning of motor neurons. Its structural domains allow it to interact with various proteins and RNA,

which contribute to necessary biological processes. SMN is essential for the functioning of motor neurons, as deficiencies in the protein cause severe neurodegenerative defects. Deficiency of this protein leads to degeneration and loss of motor neurons, resulting in muscle weakness. It is the SMN protein that plays a crucial role in the assembly of snRNPs, which are essential components of the spliceosome. The process of pre-mRNA splicing, a crucial step in mRNA development, is carried out by the spliceosome. It affects physiological processes such as transcription and RNA metabolism by forming multiprotein complexes. The SMN protein Gemin 2-8 and Unrip combine to form the SMN-Gemin complex, which plays an important role in the biogenesis of snRNPs. Smn proteins bind to snRNPs in a complex to form small nuclear ribonucleoprotein particles (snRNPs). Without the SMN protein, snRNPs cannot assemble, and motor axon functions are impaired. This complex facilitates the transport of snRNPs from the cytoplasm to the nucleus and their assembly. The SMN protein plays a crucial role in the transport and translation of mRNAs in axons in motor neurons. SMN's function in axonal transport, particularly the movement of β-actin mRNA from motor neurons to axonal growth cones, has been studied. Reduced levels of SMN protein are associated with impaired β-actin transport to axon terminals via microtubules [18], [19], [20]. The maintenance of motor neurons and muscle function depends on the function and plasticity of synapses. They play a role in the appropriate activity of lower motor neurons in the anterior horn of the spinal cord and the bulbar nuclei of the cranial nerves. SMN protein is found in the nucleus and cytoplasm. In the cytoplasm, SMN is found in granules that are involved in the transport of mRNA and ribonucleoprotein complexes along axons. In the nucleus, SMN is assembled into subnuclear structures called "gems". These are considered to be sites of snRNP biogenesis and recycling. The amount of protein varies between different organs and tissues of the body, such as the kidney, liver, heart, skeletal muscle, fibroblasts, and lymphocytes [21]. SMN protein co-localizes with various SMN complex components in axons, and most SMN granules do not contain SMN proteins located in the axonal compartment. Neurochondrin, a neuronal-specific protein, co-localizes with snRNPs and participates in cytoplasmic localization. SMN protein also has the ability to bind to the -COP component of the COPI vesicle. In primary cortical neurons, neurite outgrowth in NSC-34 cells is reduced in the absence of -COP. As a result, SMN localization and accumulation are altered. SMN and -COP are required simultaneously for proper neurite production [22]. The SMN protein is approximately 38 kDa in size and consists of 294 amino acids [23], [24]. Structural analysis of the SMN protein suggests that it consists of several major domains with specific functions. Exons 2a-2b contain a potential nucleic acid binding site, located at the N-terminal end. This domain binds to Gemin2, another protein in the SMN complex. This interaction is essential for the stability and function of the SMN complex. The Tudor domain binds to symmetrically dimethylated arginine (sDMA) residues on target proteins and facilitates interactions with various RNA-binding proteins. This domain is located in exon 3. The domain located in exon 5 is a proline-rich region, located in the central part of the protein and involved in protein-protein interactions. The Y/G box is a glycine-rich region towards the C-terminus of the protein involved in RNA binding and oligomerization and is located in exon 7. The 3'-untranslated region is localized in the region of exon 8 [25], [26].

Recent therapeutic advances in SMA have focused on restoring functional SMN protein levels to improve disease symptoms and clinical outcomes. Numerous therapeutic approaches have been developed to promote the inclusion of exon 7 in SMN2 transcripts and thereby increase the production of full-length SMN protein. These strategies include antisense oligonucleotides (ASOs) designed to mask exon splice sites, small molecules that modulate splicing factors, and gene editing techniques to correct underlying mutations [27], [28]. In particular, ASOs and small molecules act by modulating the splicing of the SMN2 gene. Antisense oligonucleotides, which are short chains of synthetic nucleic acids, prevent the skipping of exon 7 in the SMN2 mRNA transcript. The first drug approved by the FDA was nusinersen, which contains ASOs. This drug, which only aims to restore SMN expression in the central nervous system, also has side effects such as headaches, back pain, vomiting, constipation, lower respiratory tract infections, etc. Other drugs that act as splicing modifiers are small molecules such as Risdiplam, which act as an ASO and increase SMN protein expression. Despite some side effects, it improves motor function by targeting both the central and peripheral nervous systems [29], [30].

4. Conclusion

The molecular mechanisms underlying the pathology of spinal muscular atrophy (SMA) are complex and multifaceted. It involves a cascade of genetic, molecular, and cellular events, primarily resulting from a deficiency of the SMN protein. Understanding splicing defects and other dysfunctions is clarifying how SMN deficiency causes muscle atrophy, leading to the development of promising therapeutic approaches aimed at restoring SMN protein levels.

Author Contributions

A.I. Dadashova wrote the manuscript under the supervision and review of M.A. Abbasov. Both authors reviewed and approved the final manuscript.

Conflict of Interest

The authors declare no conflicts of interest.

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Abbreviations

Spinal Muscular Atrophy (SMA), Survival Motor Neuron (SMN), Deoxyribonucleic Acid (DNA), Ribonucleic Acid (RNA), Messenger Ribonucleic Acid (mRNA), Small Nuclear Ribonucleoprotein (snRNP), RNA-Binding Protein (RBP), Heterogeneous Ribonucleoprotein (hRNP), Heterogeneous Nuclear Ribonucleoprotein (hnRNP), Exonic Splicing Enhancers (ESE), Exonic Splicing Silencers (ESS), Intronic Splicing Silencer (ISS), Serine/Arginine-Rich (SR), Serine/Arginine-Rich Splicing Factor 1 (SRSF1), Transforming protein 2 homolog β (Tra2B), Src-Sssociated in Mitosis, 68 kDa (SAM68), Symmetrically Dimethylated Arginine (sDMA), Coat Protein Complex I (COPI), Antisense Oligonucleotides (ASOs), Food and Drug Administration (FDA).

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