

RNA Editing: a Role in Health and Disease

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Introduction

RNA editing represents a critical post-transcriptional modification process that diversifies the transcriptome by altering RNA sequences after their initial transcription from DNA. Among the various types of RNA editing, adenosine-to-inosine (A-to-I) and cytosine-to-uridine (C-to-U) conversions are the most prevalent in mammals. These modifications exert significant influence over crucial cellular processes, including gene expression regulation, protein function, and the overall stability of RNA molecules, underscoring their fundamental role in cellular biology [1].

The A-to-I RNA editing, predominantly catalyzed by the ADAR (Adenosine Deaminases Acting on RNA) enzyme family, profoundly impacts the transcriptome and is especially vital in the complex environment of the nervous system. Its influence extends to key aspects of neuronal function, such as synaptic plasticity and the intricate mechanisms governing neuronal activity. Dysregulation of A-to-I editing has been increasingly implicated in the pathogenesis of both neurodegenerative diseases and various forms of cancer, suggesting its potential as a significant factor in disease development and progression [2].

Complementing A-to-I editing, cytidine deaminases, notably the APOBEC3 (Apolipoprotein B mRNA Editing Enzyme, Catalytic Polypeptide-like 3) family of enzymes, are responsible for C-to-U editing. This type of editing serves as a primary defense mechanism against the replication of retroviruses, including HIV, by introducing deleterious mutations into the viral genome. However, APOBEC3 activity is not without its complexities, as it can also inadvertently lead to genetic mutations within host cells, thereby contributing to the initiation and progression of tumorigenesis [3].

Given these diverse roles, the dysregulation of RNA editing processes has become intrinsically linked to a wide spectrum of human diseases. These include malignancies such as cancer, a multitude of neurological disorders affecting brain function, and infectious diseases stemming from viral infections. The pervasive involvement of RNA editing in disease pathogenesis highlights its critical importance in maintaining human health and positions it as a promising target for the development of novel therapeutic strategies [1].

In the context of cancer, RNA editing plays a significant and multifaceted role throughout its development and progression. Alterations in RNA editing patterns can profoundly affect the expression levels of key genes, including oncogenes that promote cell growth and tumor suppressors that normally inhibit it, ultimately leading to uncontrolled cellular proliferation and the metastatic spread of cancer cells. Consequently, therapeutic strategies that specifically target RNA editing enzymes are actively being explored for their potential in treating various forms of cancer [4].

The neurological implications of RNA editing are equally vast and profound, im-

pacting fundamental aspects of brain development and function. ADAR-mediated editing, in particular, is essential for proper neural development and for maintaining optimal brain function throughout life, influencing critical processes such as neurotransmission and the formation of complex neural circuits. Disruptions in these editing processes have been linked to a wide array of neurological disorders, including epilepsy, intellectual disability, and various neurodegenerative conditions [5].

The intricate relationship between RNA editing and the immune system is also an area of growing scientific interest and understanding. ADAR enzymes, for example, have been shown to modulate both innate and adaptive immune responses. They achieve this by editing a range of viral and host RNAs, thereby influencing the immune system's ability to recognize and respond to pathogens or self-antigens. Aberrant RNA editing has been associated with the development of autoimmune diseases and chronic inflammatory conditions [7].

RNA editing introduces an additional layer of post-transcriptional gene regulation, effectively expanding the proteomic diversity that can be generated from a relatively limited genome. Comprehending the precise specificity and regulatory mechanisms governing RNA editing enzymes is paramount to fully deciphering their multifaceted roles in both normal physiological states and various disease conditions. Significant advancements in high-throughput sequencing technologies have substantially enhanced our capacity to comprehensively profile the entire RNA editome [6].

Furthermore, the presence of specific RNA editing signatures is rapidly emerging as a valuable tool for disease diagnosis and prognosis. By meticulously profiling the RNA editome, researchers can identify unique patterns that are distinctively associated with particular types of cancer, specific neurological conditions, and different viral infections. This capability holds immense promise for the advancement of personalized medicine approaches, enabling tailored treatment strategies based on an individual's unique molecular profile [8].

Finally, the study of RNA editing mechanisms is continuously propelled forward by the ongoing development of innovative experimental and computational tools. Emerging techniques, such as individual-nucleotide resolution crosslinking and immunoprecipitation followed by sequencing (RIP-seq) and m6A-seq, coupled with sophisticated bioinformatic algorithms, are significantly improving our ability to identify and analyze editing sites, understand enzyme-RNA interactions, and elucidate their functional consequences within the cellular environment [9].

Description

RNA editing, a sophisticated post-transcriptional modification, plays a pivotal role in diversifying the RNA landscape and influencing gene expression. The predominant forms in mammals are adenosine-to-inosine (A-to-I) and cytosine-to-uridine

(C-to-U) editing. These modifications are not merely passive alterations; they actively impact gene expression, protein function, and RNA stability, thereby influencing cellular processes and organismal health. Consequently, any disruption or dysregulation in these editing pathways can have profound biological consequences, linking them to a spectrum of human diseases, from cancer to neurological disorders and viral infections. This highlights the critical importance of RNA editing in human health and its potential as a fertile ground for therapeutic interventions [1].

Adenosine-to-inosine (A-to-I) RNA editing, primarily orchestrated by the ADAR enzyme family, exerts a far-reaching influence on the transcriptome. Its functional significance is particularly pronounced within the nervous system, where it plays a crucial role in modulating synaptic plasticity and overall neuronal function. Emerging evidence increasingly implicates aberrant A-to-I editing in the pathogenesis of neurodegenerative diseases and various cancers, suggesting its involvement in disease mechanisms and its potential utility in diagnostic or therapeutic strategies [2].

Cytidine deaminases, notably the APOBEC3 enzymes, are central to C-to-U RNA editing. This type of editing serves as a vital innate immune defense mechanism, particularly against retroviral infections like HIV, by introducing deleterious mutations into the viral RNA. However, the activity of these enzymes is a double-edged sword; while protective against viruses, their promiscuous action can also lead to genetic mutations within the host genome, contributing to the development and progression of cancer. Therefore, a precise understanding and regulation of APOBEC3 activity are essential for both antiviral therapies and cancer prevention strategies [3].

The broader implications of RNA editing dysregulation extend to a wide array of diseases, underscoring its fundamental role in human health. Altered RNA editing patterns are associated with numerous pathologies, including the uncontrolled proliferation characteristic of cancer, complex neurological disorders that impair brain function, and susceptibility to or exacerbation of viral infections. This pervasive involvement emphasizes the critical nature of maintaining proper RNA editing homeostasis and identifies it as a significant therapeutic target [1].

In the realm of oncology, RNA editing is recognized as a key player in both the initiation and advancement of cancer. Aberrations in RNA editing can significantly alter the expression profiles of oncogenes and tumor suppressor genes, disrupting the delicate balance that controls cell growth and survival. This disruption can manifest as uncontrolled cell proliferation, enhanced invasiveness, and the development of metastatic potential. The prospect of targeting RNA editing enzymes therapeutically is therefore being actively investigated as a novel approach for cancer treatment [4].

The neurological consequences of RNA editing are extensive, impacting fundamental aspects of brain development and function. ADAR-mediated editing is indispensable for proper neural circuit formation, the modulation of neurotransmission, and the overall cognitive and motor functions of the brain. Consequently, defects or dysregulation in these editing pathways are linked to a diverse range of neurological conditions, including epilepsy, intellectual disability, and various debilitating neurodegenerative diseases, highlighting the critical role of RNA editing in maintaining neurological health [5].

The interplay between RNA editing and the immune system is increasingly recognized as a crucial determinant of health and disease. ADAR enzymes, for example, have the capacity to influence both innate and adaptive immune responses by modifying viral and endogenous RNAs. This modulation can affect how the body responds to infections and self-antigens. Dysregulation of RNA editing has been implicated in the pathogenesis of autoimmune diseases and chronic inflammatory disorders, suggesting a significant role in immune homeostasis [7].

RNA editing contributes to cellular complexity by expanding the proteome, generating functional diversity from a limited number of genes. Understanding the precise mechanisms governing the specificity and regulation of these editing enzymes is crucial for unraveling their roles in health and disease. Recent advancements in sequencing technologies have greatly improved our ability to comprehensively map and analyze the RNA editome, providing unprecedented insights into editing patterns and their functional consequences [6].

The identification of specific RNA editing signatures is emerging as a powerful diagnostic and prognostic tool across various disease contexts. By analyzing the global RNA editome, distinct editing patterns associated with specific cancers, neurological conditions, and viral infections can be identified. This diagnostic capability holds significant promise for the development of personalized medicine strategies, enabling tailored therapeutic interventions based on an individual's unique molecular profile [8].

Advancements in both experimental and computational methodologies are continuously enhancing our understanding of RNA editing. Techniques such as RIP-seq and m6A-seq, alongside sophisticated bioinformatics algorithms, provide increasingly detailed insights into the locations of editing sites, the interactions between editing enzymes and RNA substrates, and the subsequent functional outcomes of these modifications. This technological progress is crucial for dissecting the complex roles of RNA editing in biological processes [9].

Conclusion

RNA editing, particularly A-to-I and C-to-U conversions, is a crucial post-transcriptional process that alters RNA sequences, impacting gene expression, protein function, and RNA stability. Dysregulation of RNA editing is linked to numerous diseases, including cancer, neurological disorders, and viral infections. ADAR enzymes mediate A-to-I editing, vital for nervous system function and implicated in neurodegeneration and cancer. APOBEC3 enzymes catalyze C-to-U editing, acting as an antiviral defense but also contributing to cancer when dysregulated. Aberrant editing patterns affect oncogenes and tumor suppressors in cancer, making editing enzymes therapeutic targets. In the nervous system, ADAR editing is essential for development and function, with defects causing neurological diseases. RNA editing also modulates immune responses and is linked to autoimmune and inflammatory conditions. The RNA editome can serve as a biomarker for disease diagnosis and prognosis, paving the way for personalized medicine. Advances in sequencing and bioinformatics enhance the study of RNA editing mechanisms, and targeting RNA editing therapeutically is a promising area of research.

Acknowledgement

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Conflict of Interest

None.

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