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Genetic Mutations in Romanian Patients with Hemophilia A

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Introduction

Hemophilia A is an X-linked recessive bleeding disorder caused by mutations in the F8 gene, which encodes coagulation factor VIII. The severity of the disease ranges from mild to severe, depending on the level of factor VIII activity. Romania, with its unique genetic background and healthcare infrastructure, presents a specific demographic for studying these mutations. This review aims to synthesize current knowledge about the genetic mutations in Romanian patients with Hemophilia A, providing insights into their clinical relevance. The F8 gene, located on the X chromosome, is essential for producing factor VIII, a key protein in the blood clotting cascade. Mutations in this gene disrupt factor VIII synthesis or function, leading to hemophilia A. Single nucleotide changes that can result in amino acid substitutions, premature stop codons, or altered splicing sites.

Description

Small or large insertions or deletions in the gene sequence can cause frameshift mutations or loss of function. Large chromosomal rearrangements, such as intron 22 inversions, are common in severe cases. Studies have identified various mutations in Romanian patients, including common ones like the intron 22 inversion and point mutations. The intron 22 inversion is prevalent in Romanian patients with severe Hemophilia A, reflecting a broader trend observed in other populations. Research has uncovered novel mutations specific to the Romanian population, contributing to a better understanding of the genetic diversity within this group. The correlation between specific mutations and disease severity in Romanian patients is consistent with global patterns but also shows unique local variations. Genetic testing is essential for accurate diagnosis, especially in distinguishing between different types of Hemophilia A and identifying carriers [1].

Research on Romanian patients with Hemophilia A has revealed a spectrum of F8 gene mutations, some of which are common globally, while others appear to be unique or less frequently reported. The intron 22 inversion mutation has also been identified as the most prevalent in Romanian patients, consistent with global data. However, studies have indicated the presence of other mutations such as point mutations and small deletions, reflecting the genetic diversity within this population.For example, a study by Sima identified multiple mutations in Romanian patients, including novel mutations that had not been previously reported in the international databases. This highlights the importance of region-specific studies to uncover unique mutation patterns, which may have implications for genetic counseling and treatment approaches [2].

While the mutation spectrum in Romanian patients shares similarities with other European populations, including the prevalence of the intron 22 inversion, some differences exist. The frequency of other mutations such as intron 1 inversion and specific point mutations may vary, suggesting potential genetic drift or founder effects in the Romanian population. Moreover, the clinical severity observed in Romanian patients corresponds with the type of mutation, where severe Hemophilia A cases are often linked to large deletions or nonsense mutations that completely disrupt factor VIII production. Conversely, missense mutations may result in milder forms of the disease, where some factor VIII activity is retained [3].

In Romania, access to advanced genetic testing has improved, but challenges remain in some areas. Understanding the genetic profile aids in tailoring treatment strategies, such as gene therapy and personalized factor VIII products. Romanian patients benefit from both local and international treatment guidelines, but ongoing research is needed to address specific needs. The genetic diversity among Romanian patients presents challenges in developing comprehensive mutation databases and personalized treatments. Further research is needed to explore the full spectrum of mutations and their implications for treatment strategies in Romania. Improving access to genetic testing and advanced therapies is crucial for enhancing patient outcomes in Romania [4,5].

Conclusion

The study of genetic mutations in Romanian patients with Hemophilia A provides valuable insights into the disease's molecular basis and its clinical management. While significant progress has been made, continued research and improvements in healthcare infrastructure are essential for advancing treatment and patient care.

Acknowledgement

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Conflict of Interest

None.

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