



Triple Repeat Diseases of the Nervous Systems (Advances in Experimental Medicine and Biology)

Lubov T. Timchenko

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World of Unstable Mutations The book "Triplet Repeat Diseases of the Nervous System" overviews the latest data on several disorders associated with unstable mutations. This field of research is progressing extremely fast. The number of polymorphic mutations and diseases caused by these mutations is increasing almost every month. There is a strong interest to molecular bases of triplet repeat disorders. This is explained by growing necessity to develop molecular approaches for cure of these diseases. Therefore, the authors of this book describe unstable mutations with the emphasis on molecular pathology. Broad discussion is presented on how polymorphic expansions cause cell dysfunction. The first chapter of the book focuses on the molecular pathological processes that originate "unstable" mutations. The authors review several available models by which normal "stable" region of DNA become pathogenic and discuss possible mechanisms causing DNA instability. The other chapters of the book describe inherited diseases associated with different types of unstable mutations. Based on the location of mutation in the disease gene, polymorphic expansions of the nervous system can be divided into two major groups. First group includes disorders with unstable expansions within the open reading frame of the gene such as Spinocerebellar Ataxias caused by polyglutamine expansions. The second group includes diseases caused by expansions situated within the untranslated regions of the gene.



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