
Preface

Trinucleotide repeats are relatively common in the human genome. These simple repeats have received much attention since epoch-making discoveries were made that particular trinucleotide repeats are expanded in the causal genes of human hereditary neurological disorders. For example, the CGG repeat is expanded in fragile X syndrome at the 5' untranslated region (UTR) of its causal gene. In myotonic dystrophy, it is the CTG repeat that is expanded at the 3' UTR of its causal gene. The CAG repeat was also found expanded in coding regions of the genes responsible for X-linked spinal and bulbar muscular atrophy, Huntington's disease, spinocerebellar ataxia, and other disorders.

On the other hand, expansion of the GAA repeat was identified in the intron of the gene responsible for the Friedreich's ataxia. For these trinucleotide repeat diseases, the longer the trinucleotide expansion, the earlier the age of onset and the more severe the syndrome. Thus, these findings that showed the intriguing link between a particular trinucleotide expansion and its associated neurological disorders have led to a new field of intensive study. Active research addressing the underlying mechanisms for trinucleotide repeat diseases has employed various approaches ranging from DNA biochemistry to animal models for the diseases. In particular, animal models for the triplet repeat diseases have provided excellent resources not only for understanding the mechanisms but also for exploring therapeutic interventions.

Dr. Bates and Dr. Hay have introduced an overview of trinucleotide repeat diseases in Chapter 1, using mouse models. Our book, *Trinucleotide Repeat Protocols*, covers a broad range of biochemical, histochemical, and molecular biological methods, as well as animal model systems. In the protocols, one will find how each of these techniques has been used effectively to address the unique questions relevant to trinucleotide repeat diseases. Most methods described in *Trinucleotide Repeat Protocols* are also essential and widely applicable in modern biology, not necessarily related to trinucleotide repeat diseases. All protocols are written in a self-sufficient manner so that one can repeat the entire course of the experiment without needing to refer to other texts.

Trinucleotide Repeat Protocols is divided into five sections: a review chapter, analysis of trinucleotide repeat DNA and RNA, analysis of polyglutamine-containing proteins, establishment of animal and culture models of trinucleotide repeat diseases, and their applications. All authors in *Trinucleotide Repeat Protocols* have made important milestone discoveries toward a better under-

standing of the mysterious characteristics of trinucleotide repeat sequences at the levels of DNA, RNA, and proteins to elucidate the mechanisms of hereditary neurological diseases and develop effective therapeutic interventions. *Trinucleotide Repeat Protocols*, which covers a wide range of modern molecular-biology techniques, should be of high utility not only to scientists interested in neuronal genetic diseases, but also postgraduate students and established investigators in related fields.

I wish to take this opportunity to express my great appreciation for the efforts of all authors in preparing the protocols.

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