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LECTURE

Trinucleotide repeats

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Abstract

Trinucleotide repeat (TNR) expansions are present in a wide range of genes involved in several neurological disorders, being directly involved in the molecular mechanisms underlying pathogenesis. The molecular mechanisms that correlate TNR expansions with disease are multifactorial and divergent depending on the affected gene. In fact variations in TNR can alter either gene expression and/or the function of the RNA or protein it encodes. This talk will present an overview of TNRs, with a particular emphasis on CAG repeat expansions that are translated into polyQ repeats in the affected proteins. Detail will be given to the interplay between polyQ repeat expansions and flanking regions in modulating the protein dynamics, self-assembly and aggregation of the carrier protein, which culminates in neuronal toxicity and cell death.

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