Abstract: Fragile X syndrome is the most common form of inherited mental retardation. Mutations which abolish expression of an X-linked gene, FMR1, result in pathogenesis of the disease. FMR1 encodes a cytoplasmic RNA-binding protein which interacts with two autosomal homologs, FXR1 and FXR2. These proteins are highly expressed in neurons. In addition, the FMR1/FXR proteins are associated with ribosomes. Given their RNA-binding activity and association with ribosomes, these proteins are hypothesized to bind to specific RNAs and regulate their expression at translational levels in a manner critical for correct development of neurons. Much progress has been made in FMR1 research over the past several years, but little light has yet to be shed on the physiological function of these proteins. It will be critical to define the biochemical properties of these proteins, and identify potential downstream targets to clarify the molecular mechanisms underlying the potential roles of these proteins in translation. A basic understanding of the function of this new family of RNA-binding proteins should then allow us to begin to address the question of how the lack of FMR1 expression leads to symptoms in fragile X syndrome. J. Med. Invest. 47: 101-107, 2000

Keywords: fragile X syndrome, RNA-binding protein, translation
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![Diagram of FMR1 gene and its regulatory mechanisms]

The FMR1 gene is involved in the molecular mechanisms of fragile X syndrome (FRAX). The gene is located on the X chromosome and is responsible for coding the FMRP protein. A mutation in this gene can lead to the loss of FMRP function, which in turn affects the expression of other genes in the cell.

The diagram illustrates the transcription and translation processes regulated by the FMR1 gene. The gene is transcribed into mRNA, which is then translated into FMRP protein. The protein plays a crucial role in the regulation of gene expression, particularly in the brain, where it is highly expressed.

In the context of FRAX, the mutation in the FMR1 gene leads to a decrease in the expression of FMRP protein, causing various neurological and behavioral symptoms. This highlights the importance of understanding the molecular mechanisms underlying fragile X syndrome for the development of effective treatment strategies.
in vitro quaking
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