Muscle Specific Fragile X Related Protein 1 Isoforms are Sequestered in the Nucleus of Undifferentiated Myoblast

ABSTRACT

During myogenesis, FXR1P isoforms exhibit a distinct pattern of subcellular partitioning that differs from other families of FMR proteins. As the role of this protein in Fragile X syndrome is still unknown, the model system described here should be viewed as primarily focusing on building models to understand the structure-function relationships among various members of the FARM family.

INTRODUCTION

The absence of dystrophin and its associated proteins can result in the phenotype of muscular dystrophy, as they are believed to play a crucial role in maintaining the integrity of the extracellular matrix and the membrane of muscle cells. The DAPC is composed of various protein complexes that are either directly or indirectly linked to dystrophin. The four transmembrane proteins known as sarcoglycans are organized by a fifth protein called spirochaplasia, which is believed to play cAMP signalling roles at the cell membrane. The dystroglycan complex, which interacts directly with dystrophin in the cytoplasm and laminin on the extracellular matrix, serves as a structural link between the interior and exterior of the cell. A third subcomplex includes dystobruvines and syntrophines, both of which have an unknown function. Recently, the yeast two-hybrid method was used to identify desmuslin (DMN), an -dystrobrevin-interacting protein. Both mRNA and protein are expressed mainly in cardiac and skeletal muscle and contain genes that encode a novel intermediate filament (IF) protein of 1253 amino acids. Electron microscopic analysis indicates that cessnin and desmin can colocalize with each other. During co-immunopreciptation experiments, it was discovered that the desmuslin and -dystrobrevin interaction involves the region of protein encoded by exons 8-16 of etanocellulones (precisely similar to human cDNA) and domains 1A-2A of the demineralin rod domain. Desmuslin is hypothesized to act as a mechanical support for the muscle myofibers by creating an unrecognized interface between the extracellular matrix and the Z-discs through desmin and plectin. Human genetic disorders, such as congenital and adult onset myopathies, have been associated with the involvement of several IF proteins, including duns (desmin), which may also play a role in myopathy. This possibility is supported by the exclusive expression of DMN in skeletal and cardiac muscle. We examined 71 patients with different forms of muscular dystrophy and myopathy for mutations in the DMN gene, finding 9 single-nucleotide polymorphisms (SNPs) that do not alter the protein sequence but 12 that modify the residue they encode. Our research has revealed that no controls are probable origins of the phenotype, but our findings are applicable for disequilibrium studies of this region of chromosome 15q26.3 and for studying mutation analysis and association in other genetic disorders.

CONCLUSION

Research on gene expression in the postnatal developing murine brain has revealed that only 1% of genes transcribed are regulated by development. The aim of this study is to provide researchers with an alternative approach to identify specific transcripts with unique developmental consequences without having to perform a comprehensive screening. RNA fingerprints that contain a subset of developmentally

controlled transcripts are presented here, with all the information required for isolation and identification of individual transcripti. We have identified 131 developmentically controlled transcriptions in three major expression profiles, totaling 141. Roughly 7% of the participants were categorized as C, and the rest were classified as A (61) or B (61). Our research supports the idea that modifying DDRT-PCR expression profiles indicates actual changes in expression levels during brain development. Remember that alterations in expression profiles are linked to changes in RNA level per microgram total ARN throughout the brain and, given that the postnatal brain is not a homogeneous system (i.e.