

Apolipoprotein E gene polymorphism is not a strong risk factor for diabetic nephropathy and retinopathy in Type I diabetes: case-control study

ABSTRACT

Diabetic retinopathy is not a known genetic susceptibility for Type I diabetes patients, regardless of whether APOE gene polymorphism is present. The relationship between apoE-related genes and diabetic nephropathy may be weak or moderate, but not very strong.

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 spirochaplasi, 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 dystobrevins 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 desmin and desmin can colocalize with each other. During co-immunoprecipitation experiments, it was discovered that the desmuslin and -dystrobrevin interaction involves the region of protein encoded by exons 8-16 of desmin (precisely similar to human cDNA) and domains 1A-2A of the desmin 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 desmin (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

Results: Traces of the mRNA differential display were used to identify genes with altered expression in *Pneumocystis carinii*-infected hosts, and the exact sequence of one of these fragments (gene encoding the mitochondrial ATPase 6 of F₀F₁, a subunit of this complex) was found to be homologous to the nucleotide of an expressed gene). The ATPase 6 gene is overexpressed during *P. carinii* infection, as indicated by the northern blot analysis of total RNA extracted from rat lung infected with PCA and mock-infused rabenoviruses. By in situ hybridization of cells

found to be distal and apical of the respiratory tree and of alveoli that expressed the phosphatases 6 and 8 gene, it was shown that these regions were more than 120 genes and some of those on the distal parts of mice mice. The over-expression of the ATPase 6 gene in *P. carinii* infection is thought to be caused by type II pneumocytes and Clara cells, as indicated by the presence of SP-B gene expressed through a two-color fluorescent in situ hybridization.