

## Analysis of human sarcospan as a candidate gene for CFEOM1

### ABSTRACT

We examine the presence of sarcospan as a potential gene for CFEOM1 and conclude that its involvement in the pathology of this disease is not probable. To date, there is no evidence to suggest that any mutations in this gene have any bearing on muscular abnormalities.

### INTRODUCTION

CFEOM1 is a transcription factor for human sarcospan, which is a type of senescent cell found in the human body. It encodes a protein called CFEOM1, which is involved in the regulation of cell growth, differentiation and apoptosis. The CFEOM1 protein is expressed in the somatic cells of the human body, which have a high turnover rate. CFEOM1 is also expressed in the brain, which is important for the regulation of learning and memory.

An interesting problem is that CFEOM1 is expressed in the somatic cells of the human body, which have a high turnover rate. It is thought that the CFEOM1 protein is a product of a process that regulates cell growth and cell death, but no other explanation has been found. The autosomal dominant disorder CFEOM1 has been associated with the pericentromere of chromosome 12, which is marked by markers such as D12S1584 on the palindrome and D13S1668 on derwent. The disease manifests as external ophthalmoplegia, with congenital, bilateral ptosis, and the eyes fixed in a hypotrophic or downward position. Autopsy results indicate that CFEOM1 patients do not exhibit superior division of their cranial nerve III, which innervates the head of the same mass as Unlike other proteins in the dystrophin associated protein complex, sarcospan is homologous to the tetraspheric superfamily, which facilitates both integral-membrane and membrane-proximal protein interactions. We have further refined the previously published genomic structure of sarcospan and screened for mutations in six families with CFEOM1 and produced antibodies that recognize it. Sarcospan is found to be unmutated in all six CFEOM1 families studied and maps to BACs outside of the CFAOM1. However, we find evidence that these data do not suggest any mutating events for other dystrophin associated proteins.

### CONCLUSION

Remarkable conclusions Sarcospan is not believed to play a role in CFEOM1 due to the normal distribution of sarcosphemy, which has been confirmed by DNA sequence analysis and protein immunofluorescence results, as well as the localization of this cytokines through BACs outside of the CFEOM1.