

Molecular aging: A primary or secondary trigger in late onset myopathies?

The genetic cause for OPMD is an alanine expansion in PABPN1. PABPN1 encodes for RNA binding protein, involved in multiple steps of RNA processing. The protein is ubiquitously expressed but symptoms in OPMD start only from midlife onwards, and initially are limited to only few muscles. This enigma is not fully understood.

Recent data correlating molecular and pathological changes with symptoms reveals the most prominent affected molecular pathway in OPMD and key regulators that cause muscle weakness in OPMD. A better understanding of OPMD pathology could lead to better therapeutical developments.