

Abstract

Oculopharyngeal muscular dystrophy (OPMD) is a late-onset muscular dystrophy caused by a polyalanine expansion mutation in the first exon of the poly (A) binding protein nuclear 1 (PABPN1) gene. It is characterized by progressive eyelid drooping, swallowing difficulties and proximal limb weakness. Currently there is no effective treatment for OPMD. Here, we will highlight the current translational research research advances in the treatment of OPMD. Both In vitro and in vivo OPMD disease models will be described. We will discuss different experimental therapeutic approaches for OPMD including gene editing, RNA molecules, and drug therapies.