

Supplementary figure.- Representations of protein changes caused by different PABPN1 gene mutations in OPMD. (A) Wild type (GCN) nucleotide sequence coding for a 10 alanine stretch at the N-terminus of PABPN1. (B) Most prevalent expanded alleles in OPMD. These expansions results in an increased length of the PABPN1 alanine tract (13, 14, or 15 residues). (C) A rare c.35G>C point muta_on in PABPN1 predicting a p.Gly11Ala amino acid change. This missense mutation originates an increase in the polyalanine stretch without triplet expansion (Robinson DO, Wills AJ, Hammans SR, Read SP, Sillibourne J. Oculopharyngeal muscular dystrophy: a point mutation which mimics the effect of the PABPN1 gene triplet repeat expansion mutation. J Med Genet 2006;43:e23). The methionine (Met) start residue is depicted

