Oculo-Pharyngeal Muscular Dystrophy: Clinical and Neurophysiological Features.

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Introduction:
Oculo-opharyngeal muscular dystrophy (OPMD) is a late-onset autosomal dominant muscle disorder characterized by progressive palsy of the eyelids, dysphagia, and muscle weakness in the limbs (Mirabella et al., 2000). Most cases are transmitted in an autosomal dominant fashion, although some occur in a recessive or sporadic pattern (Brais et al., 1999). Autosomal dominant OPMD is caused by short (GCC) 8-13 triplet-repeat expansions in the polyadenylation binding protein 2 (PABP2) gene, which is localized in chromosome 14q11, while autosomal recessive OPMD is caused by a double dose of a (GCC)7 PABP2 allele (Brais et al., 1999). The histopathologic, molecular, and genetic features of OPMD have become increasingly well-characterized (Mirabella et al., 2000).

Although OPMD is generally considered a muscular pathology, few papers described patients of having clinical and pathological evidence for a concomitant neuropathy. However, the frequency of associated neuropathy varies consistently among different reports from 85% (Hardiman et al., 1993) to 6-7% (Mirabella et al., 2000; Jones and Harper, 2010).

Patients and Methods:
We reviewed neurophysiological reports of OPMD patients admitted at the Neuromuscular Centre of the Catholic University between 2000 and 2013 in order to evaluate electrophysiological pattern, particularly the presence of an associated polyneuropathy. In all patients clinical findings suggested OPMD. Muscle biopsy and genetic analysis confirmed the diagnosis.

Results:
Demographic, clinical and neurophysiological features are summarized in Table 1.

Conclusions:
-Our retrospective analysis confirms that polyneuropathy is not frequently associated with OPMD (Mirabella et al., 2000; Jones and Harper, 2010).
-We also observed in three cases a mixed recruitment: mixed pattern of MUPs has been already described in OPMD (Mirabella et al., 2000) and in our cohort.
-Understanding of the spectrum of neurophysiological changes associated with OPMD may help the clinicians to differentiate it from other neuromuscular disorders and to address a correct genetic evaluation, especially in atypical cases or in the absence of a family history.

References: