Ocular, bulbar, limb, and cardiopulmonary involvement in oculopharyngeal muscular dystrophy

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Résumé en anglais	OBJECTIVES: To assess skeletal muscle weakness and progression as well as the cardiopulmonary involvement in oculopharyngeal muscular dystrophy (OPMD). MATERIALS AND METHODS: Cross-sectional study including symptomatic patients with genetically confirmed OPMD. Patients were assessed by medical history, ptosis, ophthalmoplegia, facial and limb strength, and swallowing capability. Cardiopulmonary function was evaluated using forced expiratory capacity in 1 s (FEV1), electrocardiogram (ECG), Holter monitoring, and echocardiography. RESULTS: We included 13 symptomatic patients (six males, mean age; 64 years (41-80) from 8 families. Ptosis was the first symptom in 8/13 patients followed by limb weakness in the remaining 5 patients Dysphagia was never the presenting symptom. At the time of examination, all affected patients had ptosis or had previously been operated for ptosis, while ophthalmoplegia was found in 9 patients. Dysphagia, tested by cold-water swallowing test, was abnormal in 9 patients (17-116 s, ref <8 s). Six patients could not climb stairs of whom two were wheelchair bound and one used a rollator. Six patients had reduced FEV1 (range 23%-59%). No cardiac involvement was identified. CONCLUSIONS: Limiting limb weakness is common in OPMD and can even be the presenting symptom of the disease. In contrast, dysphagia was not the initial symptom in any of our patients, although it was obligatory for diagnosing OPMD before genetic testing became available. Mild respiratory dysfunction, but no cardiac involvement, was detected.
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