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Macroglossia in adult Duchenne muscular dystrophy

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We present three adult patients with, genetically and immunohistochemically proven, Duchenne muscular dystrophy of age 18, 20, and 21 respectively, with a classical Duchenne muscular dystrophy phenotype including progressive (proximal predominant) tetraparesia, joint contractures, cardiac deficit, and respiratory insufficiency. All patients showed initial calf and tongue muscle hypertrophy, and developed later generalized limb muscle atrophy in presence of persisting macroglossia (Fig. 1). The exact pathophysiology of muscle hypertrophy/pseudohypertrophy in dystrophinopathy patients is unclear (Cros *et al.*, 1989; Jones *et al.*, 1983).

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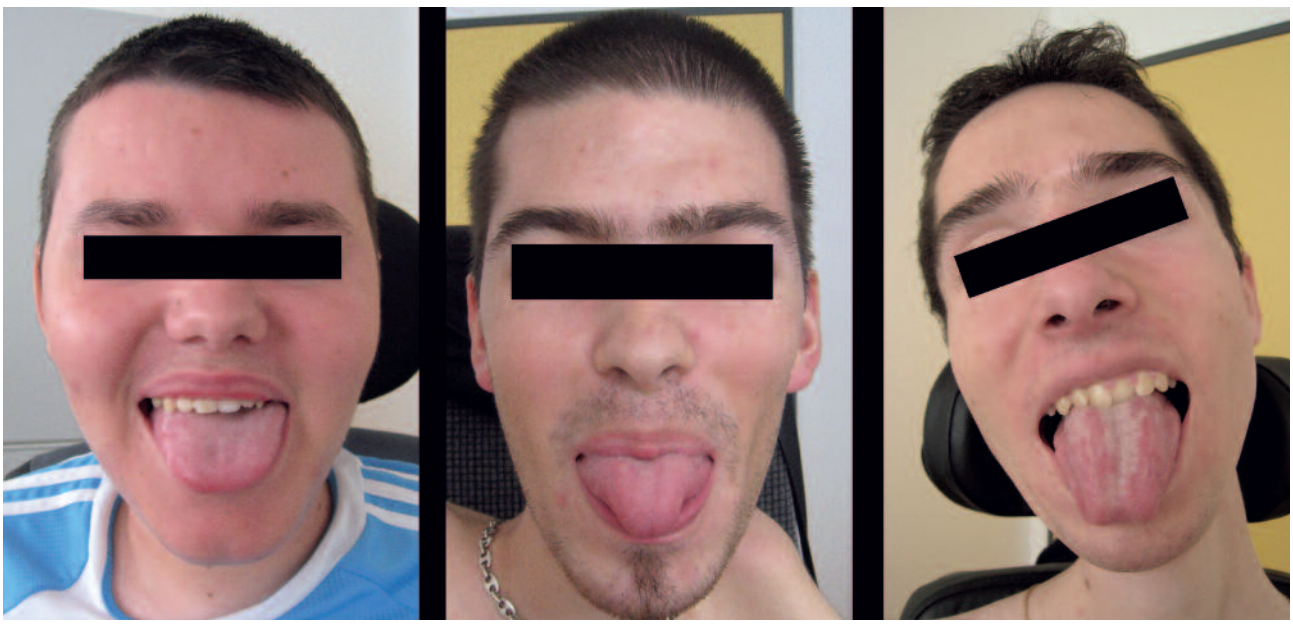

Fig. 1. — Pictures of three DMD patients showing persistent macroglossia at adult age