European Respiratory Society
Annual Congress 2013

Abstract Number: 3033

Publication Number: P2535

Abstract Group: 4.1. Clinical respiratory physiology, exercise and functional imaging

Keyword 1: Lung function testing **Keyword 2:** Congenital lesion/malformation **Keyword 3:** Sleep disorders

Title: Hypercapnic ventilatory response in patients with Steinert's myotonic dystrophy type 1

Dr. François 15720 Bughin fbughin@hotmail.com MD [1], Dr. Fares 15721 Gouzi f-gouzi@chu-montpellier.fr MD [1], Dr. Matthieu 15724 Desplan m-desplan@chu-montpellier.fr MD [1,2], Prof. Yves 15732 Dauvilliers y-dauvilliers@chu-montpellier.fr MD [2] and Prof. Maurice 15733 Hayot m-hayot@chu-montpellier.fr MD [1]. [1] Clinical Physiology, INSERM U-1046, CHRU Montpellier, University of Montpellier I and II, Montpellier, France, F-34295 and [2] Neurology, National Reference Network for Narcolepsy, Sleep-Disorders Center, CHRU Montpellier, INSERM U-1061, University of Montpellier I, Montpellier, France, F-34000 .

Body: Steinert's myotonic dystrophy type 1 (DM_1) leads to hypercapnic respiratory failure which is often considered as secondary to respiratory muscle weakness. Because of frequent central sleep disorder breathing, low central ventilatory drive is likely to be involved in the pathogenesis of hypercapnia but data are scarce and conflicting. Thus, the aim of our study was to compare the hypercapnic ventilatory response in 20 consecutive DM_1 patients versus 22 matched healthy subjects (HS) of similar age (33±10 vs.39±14 yrs). The ventilatory response to carbon dioxide was assessed by the rebreathing method with measures of occlusion pressure (P0.1). DM_1 patients performed spirometry, arterial blood gas analysis (ABG) and inspiratory maximal pressure (PImax). In response to hypercapnia, DM_1 patients vs. HS showed lower forced vital capacity, minute ventilation (VE), mean inspiratory flow rate (VT/Ti), and P0.1 ($p<0.001$, $p<0.009$, =0.06 and <0.05, respectively). There was no significant difference in hypercapnic ventilation response corrected by the predicted maximal ventilation (cHCVR) (Jones et al., Respiration,1993) (HS: 1.91±0.79 vs. DM: 2.10±1.58 %; $p=0.82$). PImax was only slightly reduced in patients (66±38% predicted values). In DM_1 patients, HCO3- was significantly correlated with cHCVR ($r=-0.48$; $p<0.05$), but PImax was not correlated with ABG parameters. Hypercapnic vs. normocapnic patients showed lower cHCVR and P0.1 ($p<0.05$). In contrast to previous studies, our results suggest a contribution of ventilatory control dysfunction to chronic respiratory failure in DM_1 patients. More data are now needed to identify the phenotypes of patients experiencing such central abnormalities.