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Title: Respiratory involvement in myotonic dystrophy type 1: Experience of pulmonology consultation in a pediatric hospital

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Body: Introduction: Myotonic dystrophy type 1 (DMT1) is a genetic muscle disease, with prevalence of 1-9/100 000 and variable clinical expression. In children may be associated with respiratory involvement. Aims and Objectives: Respiratory characterization of pediatric patients with DMT1. Methods: Study conducted in a pediatric hospital covering 2.3 million inhabitants. Descriptive and retrospective analysis of clinical records of patients followed between Jan-1985 and Feb-2013. Evaluated: sex, age, genetics, respiratory symptoms, ancillary exams, therapy and follow-up. Results: Eighteen patients were followed. Ten, genetically confirmed, were referred to pulmonology consultation (7 males): 2 newborns; 1, 27 month-old and 7 adolescents. The newborns presented respiratory failure requiring noninvasive ventilation (NIV). One died at 22 months (M) and the other maintained NIV during 27M, abandoning thereafter. The toddler snored and had recurrent respiratory infections. Polysomnography (PSG) showed obstructive hypoventilation. He goes on ventilated for 105M with good compliance and no respiratory symptoms. Adolescents were referred between 9-14 years: 5 snored; 1 had excessive daytime sleepiness. Two began NIV for sleep apnea and obstructive hypoventilation, maintaining it with good compliance and clinical response. The remaining have been followed for 2-15M. PSG already performed in 2 showed sleep respiratory disturbance. Conclusion: Most patients showed respiratory involvement as neonatal respiratory failure or sleep respiratory disorder. Even in the absence of respiratory symptoms assessment must include PSG. NIV was used with good compliance and favourable response.