supposed to be severe. The muscular dystrophy wards may be requested to offer the circumstances for those who have difficulties in continuing HMV.

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P1.12

Duchenne muscular dystrophy and optoelectronic plethysmography: A longitudinal study of respiratory function

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Respiratory muscle weakness starts as early as the second decade of life in Duchenne Muscular Dystrophy (DMD) but signs and symptoms are subtle and not easy to detect. Identify early markers of respiratory insufficiency and rule out the role of pharmacological and surgical therapies (i.e. steroids and spinal fusion) through a longitudinal study in a large group of DMD patients, in which multidisciplinary clinical evaluations are combined with Optoelectronic Plethysmography (OEP). 114 DMD patients spanning the whole range of disease severity (age range of 3–30 years). Clinical protocol: Muscular and functional evaluation (MRC and MFM scale, North Star Ambulatory Assessment and 6 min walk test), degree of scoliosis – cardiac function (ECG, 24 h ECG and echocardiography) – respiratory function (spirometry, MIP, MEP, PCF, polysomnography). OEP: abdominal contribution to chest wall volume variations (ABD) during quiet breathing, slow vital capacity and cough. The DMD patients subdivided into 4 groups according to age, showed that ABD during quiet breathing is a strong indicator of diaphragm impairment, which occurs at different times in different patients. A subgroup of 40 adolescent DMD patients showed differences in ABD related to the time spent with low oxygen saturation during night, despite similar spirometric parameters. The influence of steroids, scoliosis and spinal fusion are under analysis. Additionally, the inadequate pre-inspiration and insufficient expiratory flow, particularly of the rib cage muscles, seemed to be the cause underlying the progressive inefficient cough typical of the natural course of the disease. Our results are able to identify early signs of respiratory impairment and initial alterations of some mechanisms such as cough, that could improve the correct timing of the interventions with non invasive ventilation or cough-assisting devices.

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P1.13

Respiratory pattern during water swallowing in patients with Duchenne muscular dystrophy and myotonic dystrophy type 1

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To analyze the respiratory change during swallowing is important for the management of dysphagia in patients with muscular dystrophy. We evaluated the respiratory cycle at rest and after water swallowing, and apnea/hypopnea during water swallowing. We evaluated the respiratory patterns during water swallowing in 10 patients with Duchenne muscular dystrophy (DMD) (19.6 ± 3.2 years old, %FVC: $22.6 \pm 18.1\%$), 10 patients with myotonic dystrophy type 1 (MD-1) (46.5 ± 11.6 years old, %FVC: $55.2 \pm 16.6\%$), and 10 healthy volunteers as control subjects (43.9 ± 10.3 years old). The respiratory patterns were evaluated by the simultaneous recording of cervical swallowing sound during water swallowing. A thermistor was used for pneumography and a hypersensitive microphone was used for detecting cervical sound. The means of four continuous respiratory cycles at rest and after 3 ml water swallowing were used for analysis. The respiratory cycle, in which the amplitude was smaller than half of that at rest, was defined as apnea/hypopnea. In DMD patients, the respiratory cycle was 2.8 ± 0.6 s at rest, and 2.9 ± 0.7 s after swallowing. In MD-1 patients, that was 2.6 ± 0.5 s at rest, and 3.0 ± 0.6 s after swallowing. In control subjects, that was 4.2 ± 0.8 s at rest and 4.0 ± 0.9 s after swallowing. The apnea/hypopnea duration during water swallowing were 8.4 ± 4.1 s in DMD patients, 3.7 ± 1.6 s in MD-1 patients, and 3.2 ± 1.6 s in control subjects. The respiratory cycles in DMD and MD-1 patients were shorter than that in control subjects. The apnea/hypopnea duration in DMD patients was longer than those in MD-1 patients and control subjects. Prolonged apnea/hypopnea was observed during water swallowing in DMD patients.

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P1.14

Cardiac transplantation in Duchenne muscular dystrophy: A case report

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Despite significant advances in the management of Duchenne muscular dystrophy (DMD), dilated cardiomyopathy and heart failure remain causes of morbidity and mortality. Although cardiac transplantation is an accepted form of therapy for most patients with end stage dilated cardiomyopathy, the severely limited donor pool excludes patients with poor long-term survival and quality of life. We report the case of a 14-year-old patient with intermediate DMD and severe dilated cardiomyopathy who underwent successful cardiac transplantation. The patient was diagnosed at age three, when he presented with delayed motor milestones and weakness. Genetic testing demonstrated duplication of exon 2 in the dystrophin gene and $<2\%$ dystrophin in skeletal muscle. He was treated with deflazacort for six years, and remained ambulatory. Cardiac screening at age 10 years demonstrated dilated cardiomyopathy. He was treated with digoxin, enalapril, and carvedilol. Decompensated congestive heart failure ensued at age 14 and he was referred for cardiac transplantation. Although ambulatory, he had marked activity intolerance. Echocardiography demonstrated a left ventricular shortening fraction of 12%. He was treated with percutaneous intravenous home milrinone infusion with clinical improvement. Pulmonary function testing demonstrated mild respiratory muscle weakness which predicted he would not require ventilatory support for 5–10 years. Following discussions with medical, surgical, and bioethical personnel, he was listed. Two months later, he underwent uncomplicated cardiac transplantation. His cardiac immunosuppression includes Prograf, Imuran, and Prednisone, and he has had no rejection or serious infections. He remains ambulatory four years following cardiac transplantation. Our experience suggests that cardiac transplantation can be successful in selected patients with DMD who have a phenotype of severe cardiomyopathy with preserved pulmonary function and skeletal muscle integrity.

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