the need of further information regarding different steroids and regimes to improve patient care.

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P1.35

Health-related quality of life in patients with Duchenne Muscular Dystrophy

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Advances in symptomatic therapy have improved the physical status and life span of patients with Duchenne Muscular Dystrophy (DMD). However, very little is known about quality-of-life in this patient population. Thirty-six patients with DMD (age range 10–24 years) completed either the DISABKIDS Questionnaire for the age group 10–16 years (18/36 boys) or the SF 36 for the age group older than 16 years (18/36 boys). To assess symptoms of depression in children and adolescents, the DIKJ (Depressionsiventar für Kinder und Jugendliche) was utilized; for adults the BDI (Beck Depression Inventory). Patients' scores were compared to published norms. Neurological examination included Vignos scales. In the older 18 patients (16–24 years) physical aspects of quality-of-life assessed with the SF36 were significantly reduced ($p < .001$) compared to published normative data. Interestingly, psychological aspects of quality of life were unaltered in our sample. In our younger 18 patients (10–16 years), we also observed significantly decreased physical quality-of-life, also when compared to normative data from other chronic conditions. Social and emotional aspects of quality-of-life were also significantly impaired ($p < .001$). Clinically relevant symptoms of depression were not observed in either the younger or older patient samples. An increase in both psychological and emotional impairment is expected while the physical impairment is constantly growing at the same time in the older group. However, this hypothesis is not supported by our results. While the illness is progressing, only the physical quality of life remains impaired, whereas the psychological quality of life appears to be no longer affected compared to the younger group, possibly because of psychological coping mechanisms.

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P1.36

Evaluation of the quality of life in patients with Duchenne Muscular Dystrophy

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Duchenne Muscular Dystrophy (DMD) is a severe recessive X-linked form of muscular dystrophy characterized by rapid progression of muscle degeneration leading to loss of ambulation that markedly influences the quality of life (QoL) of the patients. QoL includes subjectivity, multidimensionality, personal perception of symptoms and cultural influence. The analysis of QoL in DMD patients is useful for planning palliative therapy or evaluating new therapeutic perspectives. The objective of this study was to qualify and quantify QoL at different dominions of life and stages of DMD by means of the questionnaires AUQUEI, SF-36 and LSI-A, and comparing them regarding practicality and effectiveness. Ninety-five patients with DMD on steroid therapy were divided into four groups according to the stage of the illness: A = 5–7 years of age, B = 8–10 years, C = 11–13 years and D = older than 13 years. The questionnaires were applied four times in a period of one year. The results concerning the four applications and the reliability inter and intra-examiner were statistically examined. Comparing the different age groups using the LSI-A patients with DMD did not lose QoL, even with disease progression. Otherwise the results of AUQUEI and SF-36 showed that patients with DMD lose QoL progressively. SF-36 and AUQUEI received negative scores in QoL most probably because the excessive evaluation of motor and clinical aspects. LSI-A questionnaire fulfill the requirements of an evaluation of QoL for children with DMD by covering a greater range of circumstances in the life of the patient. The use of scales that embraces a great diversity of circumstances in patients' lives, without considering clinical aspects excessively, is a good alternative for assessing the QoL of these patients (Supported by CAPES).

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P1.37

Total energy expenditure (TEE) of patients with Duchenne muscular dystrophy

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The doubly labeled water (DLW) method is a gold standard method for measuring total energy expenditure (TEE) in free-living subjects of 1–2 weeks. We performed the first investigation of TEE measurement in patients with Duchenne muscular dystrophy (DMD). To measure total energy expenditure (TEE) patients with DMD. A total of 26 patients with DMD aged 14–38 year-old (mean age: 21.5 ± 7.2 years old, body weight: 40.1 ± 14.5 kg) were enrolled this study. Non-invasive positive pressure ventilation was performed in 13 patients and tracheostomy positive pressure ventilation in 5. TEE was measured by the doubly labeled water (DLW) method using stable isotope ($2H_2O$ and $H_2{}^{18}O$), resting energy expenditure (REE) was determined under conditions with the individual resting comfortably about 2 h after a meal and can be assessed by simple indirect calorimetry, and the PAL was calculated from TEE divided by REE. Subjects were required to record their total energy intake (TEI) using a three-day dietary record. This study was performed under approval of institutional review board and informed consent was obtained from all subjects. The average TEE, REE and PAL were 1134 ± 186 kcal/day (31.2 ± 7.8 kcal/kg/day), 1028 ± 328 kcal/day ($n = 18$), and 1.24 ± 0.79 ($n = 18$), respectively. TEI and TEI/TEE were 1303 ± 270 kcal/day and 1.15 ± 0.22, respectively. Significant low levels of TEE and PAL are consistent findings in patients with DMD and may be associated with decrease in skeletal muscle mass and activity.

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P1.38

ENDOMUS: "Clinical and diagnostic characterization of patients with neuromuscular disorders" – Epidemiologic study

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Creatine kinase (CK) is widely used as a screening test for suspected muscle disease. When an elevated level is found in a mildly symptomatic or asymptomatic patient diagnostic uncertainty is raised. We also know that in a certain number of muscle disorders, due to their rarity and complexity is difficult to achieve a final result. The investigators implemented a protocol to perform a population screening for six genetic muscle disorders to draw a diagnostic fluxogram for patients with high CK and/or limb-girdle weakness. Our aim is to present the screening program developed for population detection and clinical characterization of patients with Glycogenosis type II and IIB, CPT II deficiency, Limb-Girdle Muscular Dystrophy (LGMD) 2A, LGMD-2I and Congenital Muscular Dystrophy type 1C, and, reveal the preliminary results of this study. Data from patients with the above mentioned diseases and from undiagnosed patients with high CK and/or limb-girdle weakness, observed in 18 neuromuscular outpatient clinics in Portugal, during 1 year was collected. A dried blood sample was taken from undiagnosed patients, after written informed consent, and was used for tandem mass spectrometry measurement of lysosomal acid alpha-glucosidase activity and screening of frequent mutations for Glycogenosis type IIB, CPT II deficiency, LGMD-2A and 2I and Congenital Muscular Dystrophy type 1C. Up to now 82 patients were included in the data base, 45 females and 33 males with a mean age of 34 years (range: 2–79). Eleven of the patients had a previous diagnostic (5 Glycogenosis type II, 1 Glycogenose type IIb, 2 LGMD-2A, 3 LGMD-2I). For the undiagnosed patients the preliminary results permitted to diagnose 3 patients with Glycogenosis type II, and 3 with LGMD-2I. The genetic screening is ongoing as well as clinical data recording. We hope to achieve, the objective of drawing a diagnostic fluxogram.

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P1.39

Contribution of accelerometry to gait analysis during the six-minute walk test

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Physical impairment associated with neuromuscular disorders may be hard to quantify in an objective manner. For ambulant patients, gait is often considered as a relevant indicator of the functional capacity of patients in daily life and this partly explains the development of walking tests such as the six-minute walk test. Even though this test is useful to quantify the distance a patient is able to walk, it does not provide any information on the quality of gait and the specific strategies adopted by the patient to compensate for muscle weakness. It is possible to gather such information using a 3D accelerometer (Locometrix, Centaure Metrix, Evry, France) worn by the patient on a semi-elastic belt during the six-minute walk test. This device is light, wireless and can register signals on long period of times. Signals are analysed on ten second duration samples by a dedicated software and provide parameters on walking dynamics such as stride length and stride frequency. Other parameters like the power developed in the cranio-caudal, antero-posterior and medio-lateral directions, the percentage of high frequency shocks and the regularity index give useful information on the motor strategy of patients and the compensatory mechanisms developed. Several samples can be chosen along the six minutes of the test to study the evolution of gait parameters. Results demonstrate the different strategies adopted by subjects: some sparing their energy, others underestimating their capacities. Examples on two different adult neuromuscular pathologies (inclusion body myositis and maltase acid deficiency) are provided to highlight the potentiality of accelerometry to deliver useful information on physical impairment of neuromuscular patients.

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P1.40

Maximum isometric muscle force obtained by hand-held dynamometry in a healthy and eutrophic pediatric population

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The determination of muscle strength as a parameter of objective function of the motor system is relevant to the neurological assessment of children. This is most important when considering a suspected neuromuscular disease. The aim of this study was to determine, with a hand-held dynamometry, the maximal isometric muscle strength in the healthy, eutrophic, Chilean pediatric population, aged between 6 and 16 years, to establish force reference values to these ages. The secondary objective was to relate force with anthropometric parameters. Healthy children were recruited from different schools in the city of Santiago. After obtaining informed consent, age, weight and height were recorded. Nutritional status was determined using BMI (CDC/NCHS) and maximal isometric force was determined in 11 different muscle groups, using digital dynamometer (Lafayette Manual Muscle Test System, model 01163). We evaluated a total of 339 eutrophic children (BMI between p10 at <p85), 168 boys and 171 girls. Children were grouped together in 10 different age groups, each ranged by one year. Each age group included between 24 and 52 kids. Strength values obtained showed an increase directly related to both increased age and increased height, with a Pearson correlation coefficient of 0.84 for both age and height. The force values were similar between boys and girls up to 12 years of age, after this age force was significantly higher in boys than in girls. The results can be used as normal strength parameters for healthy and eutrophic children between 6 and 16 years old.

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IMAGING: POSTER PRESENTATIONS

P1.41

Design of a multi-center study to examine skeletal muscles of children with Duchenne muscular dystrophy using MRI/MRS

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A multi-center study is being implemented to evaluate the potential of magnetic resonance imaging (MRI) and spectroscopy (MRS) to monitor the progression of disease in children with Duchenne muscular dystrophy (DMD) and ultimately to serve as a surrogate outcome measure for clinical trials. Longitudinal MRI/MRS measures are being acquired over five years at three geographically distributed sites with a centralized data analysis center. In addition to MR, a battery of timed functional tests and muscle strength are assessed and immortalized fibroblasts deposited in tissue repositories. Initial efforts of this study have focused on establishing quality assurance procedures and minimizing variability in MR measures across sites, and from day-to-day. This has been accomplished using two-compartment coaxial phantoms and human subjects that visited each site. To date, 21 children have been enrolled in the study: 5 controls (9.2 ± 2.4 years, 29.2 ± 6.7 kg) and 16 ambulatory boys with DMD (9.3 ± 2.0 years, 32.4 ± 9.6 kg). MR scans include fat suppressed and unsuppressed transaxial 3D-gradient echo and spin echo images as well as localized ^{1}H-spectroscopy. During the initial visit MR measures are