

P4.12**Vitamin D and bone health in Duchenne muscular dystrophy (DMD) patients**

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Background: Osteoporosis is common in DMD patients. The use of chronic steroids has resulted in steroid induced osteopenia and increased risk of fractures. The role of vitamin D in bone health of DMD patients has not been ascertained. **Objectives:** To: (1) ascertain the prevalence of vitamin D deficiency/insufficiency; (2) study the seasonal variation in 25 OH D levels; (3) assess the impact of D3 supplements on 25 OH D levels; and (4) study the relationship between 25OH D levels and bone mineral density (BMD). **Methods:** IRB approved retrospective review of data of bone health parameters of 288 DMD patients at a major pediatric neuromuscular comprehensive care center between January 2003 and September 2009. **Results:** Mean age of patients – 7.8 years (SD 4.0, range 0.6–25.0). Without D supplements and steroids, 36% had vitamin D insufficiency (25 OH D level of 20–30 ng/ml) and 54% had vitamin D deficiency (25 OH D levels <20 ng/ml). There was a seasonal variation of 25 OH D levels – mean level of 23.7 ng/ml (SD 9.5) for July–December compared to 20.2 ng/ml (SD 6.2) for January–June ($p < 0.0001$). Fifty percent of patients on a daily mean D3 intake of 1280 IU for a year had no increase in 25 OH D levels. The reasons for the lack of increase in levels are unclear. The remaining 50% on a mean daily D3 intake of 1267 IU for a year had a mean increase of 25 OH D by 12.8 ng/ml (CI 10.9–14.8). Hypercalciuria was not seen in these responders to D3 supplements. There was no correlation between 25 OH D levels and DEXA lumbar spine and distal femur R1 z-scores. 14.4% of DMD patients had a history of fractures at first visit and 10.3% developed fractures during the year of D3 supplementation. **Conclusions:** Vitamin D insufficiency/deficiency and fractures are common in DMD patients and the role of vitamin D in bone and non skeletal health in DMD should be studied prospectively.

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P4.13**Growth hormone improves growth in Duchenne muscular dystrophy with steroid-induced growth failure**

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Background: Glucocorticoid (GC) treatment is considered standard therapy in Duchenne muscular dystrophy (DMD). However, GCs cause growth failure, weight gain, and osteoporosis. GC-induced growth failure may be due to suppression of growth hormone (GH) production, GH resistance, or direct effects on bone. GH therapy has shown beneficial effects for growth in pediatric patients with GC-induced growth failure. Efficacy and safety data of GH in DMD is lacking. **Objective:** To evaluate efficacy and safety of GH in DMD boys with GC-induced growth failure. **Methods:** We report a case-series of DMD boys on daily GCs, followed at the Cincinnati Neuromuscular Clinic. The boys were treated with GH for severe growth failure and followed at pre-treatment, GH treatment initiation, 6mo, 12mo and 24mo treatment time periods. Outcomes measured included growth velocity, height, weight, BMI, neuromuscular/cardio-pulmonary function, and side effects. **Results:** DMD (39) patients with GC-induced growth failure were treated with GH for 4–32mo (mean = 11mo). They had received daily GCs for 5 ± 2.2 years. There

was an increase in mean height growth velocity by 4 ± 0.5 cm/year in the 1st year post GH treatment initiation compared to pre-GH treatment ($p < 0.0001$), and by 5 ± 1.7 cm/year ($p = 0.01$) in the 2nd year post GH treatment. No significant changes between time points for timed Gower's maneuver were noted. Thirty-foot run times were not significantly different at 6mo and 12mo compared to times at GH treatment initiation. GH was well tolerated with 3/39 experiencing side effects by 1 year. **Conclusions:** GH treatment in DMD with GC-induced growth failure improved growth. Neuromuscular function was not adversely affected by the GH treatment. Prospective studies are needed before conclusions can be drawn regarding long-term safety and efficacy.

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P4.14**Combination of steroids and ischial weight-bearing KAFOs in DMD prolongs ambulation past 20 years of age – A case report**

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Background: Patients with Duchenne Muscular Dystrophy (DMD) lose ambulation by age 12 years. Chronic steroids have increased the duration of independent ambulation by 2–4 years. Ischial weight-bearing KAFOs (IWBKAFOs) have been shown to provide an extra 2–3 years of ambulation. The combination of these two therapeutic approaches may further prolong ambulation. **Objective:** To report the outcome of the ambulatory status of a patient with DMD with a combination of chronic daily steroid therapy and ischial weight-bearing KAFOs. **Case report:** Patient is a 20.5 years old male diagnosed with DMD (with a muscle biopsy) at the age of 2.5 years after he presented with difficulty in rising from the floor. He was subsequently found with a nonsense mutation in exon 58 of the dystrophin gene. Patient has been on daily Deflazacort therapy since age 7 years. He lost the ability to arise from the floor and to walk up steps at 14.7 years of age and was not able to stand or take steps independently by age 16.3 years. Patient was evaluated at age 16.5 years and was found with MRC grade 4 strength for bilateral shoulder abduction and elbow flexion; MRC grade 2 strength for hip flexion and knee extension; bilateral hip flexor contractures of 20°, knee flexion contractures of 15° and equinus ankle contractures of 45°. Patient was fitted with IWBKAFOs at age 16.5 years following correction of his ankle and knee contractures and was able to ambulate independently in his IWBKAFOs till about age 17.5 years. At age 20.5 years, patient is still able to take slow steps for an average of 500 feet with mild to moderate two hands' support and has no significant scoliosis. **Conclusion:** A combination of daily steroids and ISWBKAFOs has extended the duration of ambulation beyond that of natural history DMD patients and that of steroids or ISWBKAFOs alone.

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P4.15**Improvement of the pulmonary functions after the use of steroids in Duchenne muscular dystrophy patients from different regions of Brazil**A.J. Godoy^a, J. Veloni^b, Y. Almeida^a, B. Delfino^a, R. Gatti^a^aUniversity City of Sao Paulo, Sao Paulo, Brazil, ^bUniversity of Ribeirao Preto, Ribeirao Preto, Brazil

Introduction: Several reports have shown the benefits of using steroids for Duchenne Muscular Dystrophy (DMD) patients. All agree the need of wheelchair can be postponed by using those drugs. What it is still controversial it is if we can get other good outcomes. We decided to analyze the pulmonary functions of DMD patients taking steroids. **Method:** We selected 35 DMD patients from different regions of Brazil seen in an outpatient clinic and checked who was taking steroids. Standardized parameters regarding pulmonary functions were analyzed. We also checked muscle strength, cardiac function, creatinokinase levels and quality of life. A table was prepared to compare the subgroups of patients using the drugs with those not using them. **Results:** All patients are between 20 and 25 years of age. Six patients were under that medication and 29 were not. Patients taking steroids used prednisone (0.5 - 1.0 mg per Kg per day). The age of onset, regarding the use of the drug varied from 3 to 14 years. The duration of use ranged between 2 and 7 years. Fifty per cent of patients taking steroids needed ventilatory assistance, between 2 and 10 hours per day while 75% of the patients not using the medication were in that condition (between 10 and 20 hours per day). **Conclusion:** Our results strongly suggest the importance to prescribe steroids for DMD patients, even after they lost ambulation. Keeping those patients in a good shape will give them a chance to be enrolled in a near future in clinical trial, for example, with exon-skipping or stem cell therapy.

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P4.16 **Effects of different exercises on hemodynamic responses in Duchenne muscular dystrophy**

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Aim: The aim of this study was to investigate the effects of different exercises on heart rate and oxygen saturation of children with Duchenne Muscular Dystrophy (DMD). **Subjects and method:** 30 subjects (mean age: 7.87 ± 1.45 years) with DMD in early stages (stage1/2) of disease according to the Brooke Functional Classification included in this study. 12 of 30 children were required to climb up and down the standardized 5 stairs during 3 minutes. 23 of 30 children did cycling during 40 minutes after a 5 minutes warming up period with stretching exercises. The day after cycling, the physical therapy exercises including specific stretching and aerobic exercises for both upper and lower extremities according to the functional status of children were applied by a physical therapist to 10 of 23 children during 40 minutes. The hemodynamic responses including heart rate and oxygen saturation of children were recorded before and just after the exercise applications. **Results:** The heart rate was elevated after climbing stairs and physical therapy exercises ($p < 0.05$), although didn't show a significant difference after cycling. Mean heart rate was % 58 of maximal heart rate as submaximal only just after physiotherapy exercises. There were no significant differences in oxygen saturation after 2 exercise applications ($p > 0.05$). **Discussion:** Heart rate is mostly affected by exercises including both upper and lower extremities than exercises restricted to lower extremities in DMD patients in early stages of disease. Submaximal heart rate was only achieved after physiotherapy exercises.

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P4.17

Effects of exercises on muscle strength and fatigue level in Duchenne muscular dystrophy

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Aim: The aim of this study was to investigate muscle strength responses and fatigue levels of children with Duchenne Muscular Dystrophy (DMD) after two different exercises. **Subjects and Method:** 23 children with DMD in early stages of disease (stage 1/2) according to the Brooke Functional Classification included in this study. All of them did cycling on a stable bicycle actively during 40 minutes after a 5 minutes passive warming up period with stretching exercises. 10 of 23 children was also applied physical therapy exercises by a physiotherapist including both of upper and lower extremities during 40 minutes the day after cycling. Lower extremity muscle strengths of children were measured with a hand-held dynamometer and fatigue levels were assessed with an effort rating scale called 'Pictorial Variant of the Children's Effort Rating Table' by self-report before and just after the exercises. **Results:** The fatigue levels were increased after two exercise applications ($p < 0.05$). The strengths of quadriceps and hamstrings and total lower extremity muscles decreased after cycling ($p < 0.05$), while didn't show a significant difference after physical therapy exercises. **Discussion:** The perceived exertion by children were similar after two exercises. It is thought that the decrease in muscle strength after cycling is due to the exhaustive effect of cycling specifically on quadriceps and hamstring muscles in supported sitting position. The muscle strength was maintained just after physical therapy exercises. The exercise programme which was planned and applied by a physiotherapist showed no risk of specific muscle fatigue.

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P4.18

Asphyxia in a Duchenne muscular dystrophy patient due to tracheal compression by brachiocephalic artery

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Background: Trachea and brachiocephalic artery are anatomically crossing. This anatomical location sometimes causes asphyxia and other respiratory problems due to tracheal compression by brachiocephalic artery (TCBA), as known as tracheomalacia, in patients with neuromuscular diseases. Scoliosis, opisthotonus and laryngo-tracheal separation are the risk factors of TCBA. Typical symptoms of TCBA are recurrent asphyxia and/or stridor. We present a Duchenne muscular dystrophy boy with TCBA and summarize the key points of the diagnosis and management of TCBA on our experiences with other patients with neuromuscular diseases with TCBA. **Case presentation:** A wheelchair-bound 13-year-old Duchenne muscular dystrophy boy was admitted to our hospital because of cyanosis, dyspnea, and difficulty in expectoration of sputum. His oxygen saturation was 85% and pCO₂ was 122 mmHg under 10 L/min oxygen mask. He was immediately intubated. Chest X-ray revealed no cause of his dyspnea. He was stable under mechanical ventilation but sometimes suddenly deteriorated and DOPE evaluation (tube displacement, tube obstruction, pneumothorax, equipment failure) detected no problems. He was extubated but re-intubated several hours later because of dyspnea. He received tracheotomy but dyspnea and asphyxia attack with no DOPE problem repeated. Further evaluation by bronchofiberscopy and contrast computed tomography