

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

B. Wong, S.Y. Hu, A. King, P. Horn

Cincinnati Children's Hospital Medical Center, Cincinnati, United States

**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.

doi:10.1016/j.nmd.2010.07.208

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

J.J. Collins, M.M. Rutter, J. Woo, S. Rose, H. Sawani, L. Cripe, K. Kinnert, K. Hor, B. Wong

Cincinnati Children's Hospital Medical Center, Cincinnati, United States

**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  $5 \pm 2.2$  years. There

was an increase in mean height growth velocity by  $4 \pm 0.5$  cm/year in the 1st year post GH treatment initiation compared to pre-GH treatment ( $p < 0.0001$ ), and by  $5 \pm 1.7$  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.

doi:10.1016/j.nmd.2010.07.209

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

A.C. Pardo, T. Do, T. Ryder, C. Schaeffer, M. McMahon, B. Wong

Cincinnati Children's Hospital Medical Center, Cincinnati, United States

**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.

doi:10.1016/j.nmd.2010.07.210

**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<sup>a</sup>, J. Veloni<sup>b</sup>, Y. Almeida<sup>a</sup>, B. Delfino<sup>a</sup>, R. Gatti<sup>a</sup><sup>a</sup>University City of Sao Paulo, Sao Paulo, Brazil, <sup>b</sup>University of Ribeirao Preto, Ribeirao Preto, Brazil