Abstracts

87 COMPARISON OF FUNCTIONAL ABILITY IN SIBLINGS WITH DUCHENNE MUSCULAR DYSTrophy
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Results Between Feb 2011-Aug 2020, 75 (M=43) patients were treated. Mean age at follow-up was 87.5 months (range 18–157 m). 42 cases were bilateral and thus 117 feet were treated. 45 (60%) were idiopathic and 30 (40%) non-idiopathic.

Of the 75 patients, 50 (67%) underwent tenotomy under local anaesthetic in clinic. Tenotomy rates were higher in non-idiopathic vs idiopathic patients (83% vs 56%)(P<0.002)

Mean dorsiflexion (knee extended) in affected feet was 14°. In unilateral cases mean dorsiflexion (knee extended) in unaffected foot vs affected foot was 17° vs 12°. All idiopathic feet were plantigrade.

Abnormal evertor function was seen in 3 of 66 (5%) idiopathic feet versus 22 of 51 non-idiopathic feet (43%) (p<0.0001).

Calf circumference discrepancy was higher in unilateral cases versus bilateral (mean 1.3 cm versus 0.6 cm respectively). Unilateral cases undergoing tenotomy had a greater mean circumference difference (1.5 cm) versus those managed without tenotomy (0.7 cm).

43 idiopathic patients were suitable for OXFORD scoring at time of last review (i.e. older than 5 years old). The overall median OxFAQ was 96% with points lost only in the physical scale. Function in non-idiopathic cases was determined by their overall condition.

Conclusion The idiopathic club foot is well treated by a physiotherapy-delivered Ponseti technique with excellent outcomes at 5 yr follow-up both subjectively and objectively.

88 THE RELATIONSHIP BETWEEN UPPER AND LOWER LIMB FUNCTION IN A COHORT OF CHILDREN WITH CHARCOT-MARIE-TOOTH DISEASE
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Charcot-Marie-Tooth (CMT) is a progressive disease with clinical signs presenting first in the distal lower extremities. Upper limb function in this population is also affected at a later stage of life but it is poorly researched and little is known about hand function limitations and loss of manual dexterity. The purpose of this study is to investigate the possible relationship between upper and lower limb function in a group of children and adolescents. The CMT natural history study at Great Ormond Street Hospital in London has been collecting longitudinal data of more than 120 children and adolescents with CMT (age range 4 to 21 years). Eighty one children with CMT type 1 (53% CMT1A), 22 with type 2 and 21 with other types of CMT have been assessed up to date. To evaluate upper limb function we used myometry, functional dexterity test and 9 hole peg test; to measure lower limb function the 6 minute walk test, long jump and plantar and dorsiflexion strength tests. In this study we will use correlation analysis to explore the concordance between the upper and the lower limb function. A comparison will be made between children and adolescents with demyelinating CMT and those with axonal CMT, and also individual genotypes, to look if variation in genetic subtype affects individuals in different ways. In our analysis we will assess longitudinally the correlation between upper and lower limb function in children with different subtypes of CMT. If a strong relationship between the two is found in individual conditions, we will assess further the predictive model of loss of function between upper and lower limbs in different genetically defined conditions. This study is a part of a MSc project being undertaken at UCL and a full report and results will be presented later in the year.

89 CHRONIC DISSEMINATED CANDIDIASIS TREATED WITH ADJUVANT CORTICOSTEROID TREATMENT
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Early diagnosis is important to optimise management and prolong function in patients with Duchenne Muscular Dystrophy (DMD). There is little research comparing functional outcomes of siblings diagnosed at different ages.

This study aims to analyse the effect of age at diagnosis and prescription of steroids on function in siblings with the same genetic diagnosis of DMD. Functional ability was measured using the North Star Ambulatory Assessment (NSAA) which is a validated outcome measure in ambulant children with DMD.

NSAA data from 24 siblings (48 patients) was collected from their first NSAA assessment at Great Ormond Street Hospital, including all scores until July 2020.

Age matched NSAA scores were collated at 6 monthly intervals (± 3 months) between 5.5–10.5 years in siblings, where one or more NSAA scores could be compared. 17 siblings (34 patients) met the inclusion criteria and were analysed.

Results show that younger siblings were diagnosed on average 2.7 years earlier and started steroids 0.8 years earlier. The median peak NSAA scores were higher in older siblings at ages 5.26–6.25 and 7.26–8.25 years. Between 6.26–7.25 and after 8.26 years, the younger siblings median NSAA scores were consistently higher, although there were small numbers in each sub-group. Wilcoxon Signed Rank test showed no significant differences between groups.

Analysis of the graph suggests that despite the earlier age at diagnosis, younger siblings did not diverge from the older sibling in the first phase of the disease. Visual inspection of the NSAA score shows that after 8 years old, the younger siblings consistently scored higher values. This suggests that earlier initiation of steroids is likely to have played a role in this outcome, as previously demonstrated in the DMD population. Further research will assess long-term effects of these trends, regarding age at loss of ambulation, and of respiratory insufficiency.