significance was reached for hemoglobin import \( (p = 0.000001) \) and calcium ion binding GO \( (p = 0.00007) \). These GO groups were also significantly enriched with genes caring a mutation annotated as damaging according to the PolyPhen-2 score. Further validation of the identified SNVs by Sanger sequencing and also by genotyping in a larger DMD cohort is currently being carried out in order to confirm their possible role as biomarkers for DMD.

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**ASSESSING DUCHENNE MUSCULAR DYSTROPHY – POSTER PRESENTATIONS**

**S.P.49 Cautions regarding subcapital whole body DXA scan interpretation among boys with Duchenne muscular dystrophy (DMD)**

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Lumbar and subcapital whole body Dual-energy X-ray absorptiometry (DXA) scans are recommended for monitoring bone mass (BMD) in boys with DMD. Complications of DMD and corticosteroids include: obesity, contractures, loss of ambulation and fractures contributing to errors in subcapital DXA accuracy and reproducibility. DXA scans in DMD boys frequently contain technical artifacts influencing BMD interpretation. We report the frequency of positioning and acquisition issues influencing subcapital DXA scan interpretation of DMD boys. Subcapital DXA scans \( (n = 189) \) of 64 DMD boys on Deflazacort were reviewed for motion artifact, contractures, obesity, suboptimal positioning, missing body segments, and orthopaedic hardware. Artifact-free scans with optimal positioning were obtained in the 64 boys when ambulatory, age 8.1 ± 2.2, BMI 16.7 ± 2.3 and 18.9 ± 3.9% subcapital body fat. 25% of the boys (16/64) when non-ambulatory, age 16.1 ± 2.1 had scans with motion artifact. 47% of the boys (30/64) developed contractures precluding optimal positioning. 14% of the boys (9/64) age 17.0 ± 2.5 when non-ambulatory, BMI 28.3 ± 4.0 and 52.8 ± 5.9% subcapital body fat had scans missing body segments; 6/9 missing digits, 1/9 both elbows, 1/9 ulna and 1/9 foot. 8% of the boys (5/64) age 17.1 ± 2.8 had hardware which increased BMD (g/cm²) from 8% to 124% \( (52.3 ± 50.6\%) \) from their previous hardware-free scan. In general there are no technical issues with subcapital DXA scans in DMD boys with DMD age, subcapital DXA scans contain a higher frequency of scan acquisition and interpretation errors from multiple sources. Attention to positioning for scan acquisition and analysis of scans in array mode may limit the potential for reproducibility and accuracy errors. Despite technical limitations DXA can identify boys with high fat mass and fracture risk. Future studies should compare the diagnostic yield of serial pQCT to DXA, to avoid the technical pitfalls of subcapital DXA.

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**S.P.50**

Physiotherapy assessment of Duchenne and Becker muscular dystrophy in the Neuromuscular Clinic: Standardising the process

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The Neuromuscular Clinic at the Royal Children’s Hospital (RCH), Melbourne, manages the care of boys with Duchenne and Becker muscular dystrophies (DMD and BMD) in the Australian states of Victoria and Tasmania. The multidisciplinary team includes two physiotherapists who share patients and often need to communicate with their community colleagues regarding ongoing physical management for these boys. Since the clinic’s inception in 2007, the physiotherapists have developed and refined standard assessment methods and documentation for ambulant and non-ambulant boys with DMD/BMD. The assessments include subjective measures: (including current mobility, falls, wearing of splints, performance, performance of a home exercise programme) and objective measures (including goniometry, timed function tests, North Star Ambulatory Assessment and EK Scale). This process has been found to have many advantages including:

- Facilitation of patient data sharing between clinicians within the RCH and community centres,
- Consistent collection of the most relevant data,
- Familiarisation of the boys with standard assessment, optimising their performance and enhancing their readiness to participate in clinical trials,
- Quality clinical record keeping that makes it easy to track boys’ progress or extract data for clinical audits.

Having standard forms prompts the physiotherapists to constantly question the usefulness of the information collected and evolve the process accordingly to meet clinical needs. The current forms will be presented and represent a potential means of collaborative clinical research between units providing specialised physiotherapy care of children with neuromuscular disorders.

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**S.P.51**

A study of below knee serial casting for calf contracture in ambulant boys with Duchenne muscular dystrophy

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The progressive muscle weakness that occurs in Duchenne muscular dystrophy (DMD) is known to be associated with secondary muscle tightness and contracture. The Achilles tendon (TA) and calf muscle complex (gastrocnemius and soleus) is typically one of the earliest and most severely affected muscle groups. It has been noticed clinically that there is a subset of boys who develop surprisingly early shortening in the presence of well-preserved lower limb muscle strength. A small prospective study has commenced to assess the effects of short term below knee serial casting on muscle length as well as strength, physical function and endurance. Tight inclusion criteria ensure that only boys with good anti-gravity muscle strength and range of motion at hip and knee are included. Outcome measures include goniometry, myometry, timed function tests, the North Star Ambulatory Assessment, Six Minute Walk Test, falls frequency and measurement of gait parameters using the GAITRite system. Early data will be presented.

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**S.P.52**

Troponin T adventures in Duchenne muscular dystrophy

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Troponins are the biomarkers of choice to diagnose acute cardiac injury. We present two cases of DMD with elevated serum Troponin. Case 1 is a 14 year old non-ambulatory male who presented to the ER with chest pain and diaphoresis. There were q waves in leads II, III and aVF with no ST elevation. Troponin I level was elevated at baseline and at 12 h. He was then transferred to a pediatric hospital. The chest pain resolved at 14 h. An echocardiogram was normal with good biventricular function. No perfusion mismatch was detected on a lung ventilation perfusion scan. A CT of the coronary arteries was normal. The Troponin T remained elevated at 24 h (0.34 μg/L, N < 0.1) and 36 h (0.13 μg/L). The conclusion was that the chest pain was not cardiac in origin and the elevated troponin was thought to be secondary to DMD. The ECG findings remained unchanged for the past 6 years. Case 2 is a 4 year old boy who presented with abdominal pain and tachycardia (140–160 bpm). The CK was elevated (1068 U/L, N = 75–230 U/L) and Troponin T - high sensitivity was elevated (66 ng/L, N < 14 ng/L). He was diagnosed with ruptured appendicitis and query myocarditis. The Troponin T – high sensitivity continued to be elevated (59–177 ng/L) as was the CK (3824–11,456 U/L) over 1.5 months. He had a normal 24 h Holter monitor. He had 4 echocardiograms during this period showing normal left ventricular function (ejection fraction 68% and fractional shortening 37%). The first echocardiogram revealed a small pericardial effusion that resolved on subsequent echocardiograms. In both cases there were no acute changes noted on cardiac testing despite an elevated Troponin T. The second case may have had myocarditis however, there were no changes on the ECG and typically the elevated troponin will return to normal within a few days. If cardiac injury is suspected, a cardiac MRI could be considered to further evaluate the myocardium for injury.

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S.P.54
The effects of ankle position to function in Duchenne muscular dystrophy
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The progressive muscle weakness in muscular dystrophy causes muscle shortening and joint limitations that effects the functions of children. The study was planned to investigate the effects of ankle limitation on functions in children with Duchenne Muscular Dystrophy. Forty-three DMD patients (mean age 96.84 ± 28.08 months) who are in first and second level according to Brooke Scale were included to the study. Weight, height, hip, knee and ankle joint limitations, 10 m walking time, total lower extremity muscle strength (hip flexors, extensors, abductors and adductors, knee flexors and extensors, ankle dorsi and planatar flexors) and functional independence level according to WeeFIM (114.21 ± 13.97) of children were recorded. There were negative correlations between ankle limitation and lower extremity muscle strength (r = −0.381) and WeeFIM locomotion subtitle (r = −0.358) (p = 0.005). There were positive correlations between ankle limitation and height (r = 0.575, p < 0.05), weight (r = 0.577, p < 0.05), limitation of knee joint (r = 0.475, p < 0.01), 10 m walking time (r = 0.368) and Brooke functional level (r = 0.397, p < 0.05). Equinus position in ankle joint is an important postural adaptation which maintains children’s functional activities although increased joint limitations and decreased muscle strength while growing up in DMD children.

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S.P.55
Transition and outcomes for young men with Duchenne muscular dystrophy in New South Wales
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Duchenne muscular dystrophy (DMD) is a progressive disorder that affects 1 in 3500 males and that typically results in death between 20 and 35 years of age from respiratory or cardiac failure. International standards of care have been devised to guide the management of DMD patients. Due to its high incidence in all populations of the world and relatively homogeneous natural history, health outcomes in DMD provide a useful yardstick by which to assess neuromuscular services between regions. There is currently little data available on how males with DMD in NSW, Australia, fare in terms of health outcomes. Such data would provide health service providers important feedback about the effectiveness of neuromuscular services in NSW compared to other populations in Australia and overseas. This study was designed to conduct an audit of all paediatric and adult neuromuscular services in NSW to document the morbidity and mortality of patients with Duchenne muscular dystrophy in NSW in the last 10 years, and evaluate their transition experience to adult services. In addition we describe the current range of paediatric...