Development of an upper limb functional scale in dystrophinopathies

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Development of appropriate outcome measures that are able to measure both deterioration and therapeutic effectiveness across the spectrum of severity in dystrophinopathies is complex. A multi-disciplinary international Clinical Outcomes Group consisting of clinicians, scientists, and patient advocacy groups specifically identified a need for a scale to measure motor performance of the upper limb. Out of this larger group, a Physiotherapy Focus Group was formed. Two in person meetings were held where existing upper extremity scales were reviewed for common elements and functional relevance in DMD. Individuals with DMD contributed in these meetings by participating in the proposed assessments and offering their expert opinion on task difficulty and relevance. In parallel, patient focus groups identified patient related outcome measures. The aim of the two parallel groups was to ensure that the scale clearly related to meaningful functional activities. A new scale, PUL (Performance of the Upper Limb), was then designed according to a specific contextual framework, of upper limb function in both ambulant and non-ambulant individuals with dystrophinopathy. The methodology included establishment of the scale framework considering functional muscle strength, growth and contractures on motor performance. Item generation was based on expert review of the assessments and test implementation. Item suitability was based on clinical progression patterns, patient testing and observation, scoring detail, and video review of movement patterns with item performance. Operational definitions and manuals were generated to standardize PUL assessments and piloted across seven international sites. The results from a preliminary Rasch analysis of 70 patients were then interpreted for clinical sensibility and the scale appropriately modified.

S.P.9

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S.P.10

Upper limb assessment in DMD: An exploratory study and critical review of the existing scales

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In the last few years there has been increasing evidence of the lack of outcome measures in non ambulant patients affected by Duchenne muscular dystrophy. We report an exploratory study on upper limb function in 61 DMD patients using three measures previously used in DMD and an upper limb module previously developed for spinal muscular atrophy. This study, rather than being a formal suitability study, explored whether the existing scales were appropriate for DMD patients at different ages and spectrum of abilities. The Brooke scale proved to be easily and quickly administered and useful to subdivide patients into motor performance domains but comprises only few categories providing ordinal data and does not specifically assess hand function. The Jebsen Hand Function test is a standardized timed test. Most items are composite tasks that do not take into account compensatory strategies in the scoring system and are biased by testing positions and contractures. Some items proved not to be suitable due to age and IQ dependency. The MFM is a functional scale developed in NMD to assess motor performance. It includes items subdivided in functional domains and also assesses distal function. A floor and ceiling effect were observed in some items across the whole spectrum of abilities and age range. The ULM was developed for non-ambulant SMA patients and evaluates upper limb activities on all planes of workspace. When used in DMD the scoring system and weight bearing did not sufficiently assess patients across spectrum of abilities with a ceiling effect in the stronger ones. Each measure had pros and cons and several shortcomings, related to posture, contractures and pattern of weakness requiring compensatory strategies. None of the existing scales proved to be suitable alone, this probably reflecting the fact that most of them had not been specifically designed for use in DMD. Studies are in progress to overcome some of these difficulties.

S.P.11

Development of a virtual upper extremity assessment tool for individuals with DMD across the lifespan

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Clinical trials are being conducted for DMD using timed walking as a primary outcome measure due to the ease of administration, ability to quantify distance, and established validity. Unfortunately, participation in clinical trials is not equally accessible to all individuals due to the lack of a reliable, valid and sensitive upper extremity measure for the non-ambulatory population. It is essential that this gap in knowledge be filled.
as upper body movement is required for many activities of daily living impacting quality of life across the lifespan and abilities in individuals with DMD. The purpose of this project was to develop a reliable and engaging upper extremity measure to quantify workspace volume, reaching velocity, and rate of arm fatigue using the Kinect system to capture data during a video virtual reality game across the spectrum of abilities. The Microsoft Kinect Windows 7 used for this project is a controller-free gaming device that projects an infrared reference matrix that is reflected off of the person and generates a high-resolution depth map of each pixel allowing for tracking of movement. Software is available to track 20 joints and provide positional data which can be used to calculate measurements such as workspace volume, reaching velocity, and rate of arm fatigue. Virtual gaming was developed as a motivational tool to be used during assessment. Work space volume gaming uses bubble popping game while velocity and fatigue use a disappearing image to encourage rapid movement. A prototype assessment system is being validated against a marker based motion capture system and traditional upper extremity assessments such as the Jepsen Hand Function Test, 9-hole peg test, and the Brooke Upper Extremity Rating Scale. Preliminary data suggests that this may be an engaging, reliable and valid upper extremity assessment measure for individuals with DMD across the lifespan.

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S.P.13
Exploring arm function in FSHD by means of questionnaires
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Facioscapulohumeral muscular dystrophy (FSHD) is a progressive muscular dystrophy. One of the major problems is limitations in performing daily activities. The natural course of the disease has been described before, and research is done to investigate functional ability of walking. However, little is known about functional abilities of the arms, which are even more important in performing many daily activities. Clear understanding is essential to improve rehabilitation therapies and for development of new arm supports. The aim of this study is to investigate arm function of people with FSHD on activity level, by using questionnaires. A web-based questionnaire to assess arm function was developed. We used questions from the Brooke scale (grading scale, 1–6), the Capabilities of Upper Extremity questionnaire (CUE) (basic arm functions, 30 items, max score 210, higher score means better function) and Abilhand questionnaire (22 ADL items, max score 44, higher score meaning lower function). Also questions about disease duration, age, gender were asked. The Dutch patient organization (VSN) supported with distribution of the questionnaire. Seventy-six people participated (age 13–74 y) in the period August 2011 till February 2012. Preliminary results show that from this group, 88% have Brooke 2–4. The CUE total median score was 155 (range 53–210), corresponding with 73.8% of the maximum score. The total Abilhand median score was 38 (range 13–44) corresponding with a 86.4% of the maximum score. Especially activities above shoulder level are problematic. Patterns of limitations in arm function are further analyzed as are correlations with disease course, age, and gender. The project McArm is a national Dutch ‘Pieken in de Delta’ project. It is partially funded by: Agentschap.nl, an agency of the Dutch Ministry of Economic Affairs, Agriculture and Innovation, the Province of Noord Brabant and the Province of Limburg.

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S.P.14
Exploring arm function in persons with LGMD by means of questionnaires
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Limb-Girdle Muscular Dystrophies (LGMD) mainly involve the pelvic and/or shoulder girdle muscles. One of the major problems is limitations in performing daily activities. The natural course of the disease has been described before, and research is done to investigate functional ability of walking. However, little is known about functional abilities of the arms, which is even more important in performing many daily activities. Clear understanding is essential to improve rehabilitation therapies and for development of new arm supports. The aim of this study is to investigate arm function of people with LGMD on activity level, by using questionnaires. A web-based questionnaire to assess arm function was developed. We used questions from the Brooke scale (grading scale, 1–6), the Capabilities of Upper Extremity questionnaire (CUE) (basic arm functions, 30 items, max score 210, higher score means better function) and Abilhand questionnaire (22 ADL items, max score 44, higher score meaning lower function). Also questions about disease duration, age, gender were asked. The Dutch patient organization (VSN) distributed the questionnaire. Fifty-one people (age 6–72 y) participated in the period August 2011 till February 2012. Preliminary results show that in this group, 33% has Brooke scale 1 and 51% Brooke 2–4. The CUE total median score was 159 (range 60–210), corresponding with 75.7% of the maximum score. The total Abilhand median score was 39 (range 8–44) corresponding with