P.2.2
North Star ambulatory assessment in young DMD boys
R. De Sanctis, F. Bianco, E. Mazzone, C. Palermo, S. Sivo, L. Fanelli, A. Graziano, M. Pane, E. Mercuri
Catholic University, Rome, Italy

Very few studies have investigated early neurodevelopmental and motor aspects in preschool boys affected by Duchenne muscular dystrophy. This is mainly due to the age at diagnosis which is still on average above the age of four years. Although the first signs of concern can often be backdated to the second year of age, when DMD children show some signs of developmental delay and inability to develop new motor abilities the diagnosis is on average performed much later. The recent development of therapeutic approaches for DMD has highlighted the need to identify clinical outcome measures for planned therapeutic clinical trials that could be used as early as possible, ideally soon after diagnosis when the disease is still in the early phases.

There have been suggestions that the North Star Ambulatory Assessment, a functional scale already used in DMD children older than 4 or 5 years may be used in younger children.

The aim of this study was to assess the suitability of this scale in young children by performing the scale in a cohort of typically developing and DMD children all assessed before the age of 5 years. More specifically we aimed to identify if all the items were appropriate for children from the age of 3 years onwards.

Results: in the typically developing children the scale was easily administered in all. All the children above the age of 5 were able to pass all the items with a full score while children below the age 5 often failed some items, such as hopping on one leg. Children between the age of 3 and 4 also failed additional items such as standing on one leg, getting up from the floor without upper limb aid, lifting head from supine and standing on heels. DMD boys had similar findings but in a proportion of them other items were also failed.

Our results suggest that the NSAA can be easily administered in typically developing children and DMD young patients but the results should be interpreted taking into account age related difficulties due to the age appropriate development of motor coordination skills.

http://dx.doi:10.1016/j.nmd.2013.06.407

P.2.3
Assessment of Upper Limb function in DMD patients: Comparison with normative data
E. Mazzone 1, M. Pane 1, L. Fanelli 1, R. De Sanctis 1, F. Bianco 1, S. Sivo 1, A. D’Amico 2, S. Messina 1, L. Polito 1, P. D. Battini 3, S. Frosoni 3, M. Pedemonte 3, P. Boffi 1, D. Pegoraro 1, Berardinelli 2, G. D’Angelo 2, A. Pini 1, E. Iotti 1, G. Baranello 1, L. Morandi 1, E. Mercuri 1
1 Catholic University, Rome, Italy; 2 Ospedale Bambino Gesù, Rome, Italy; 3 Messina University, Messina, Italy; 4 Second University of Naples, Naples, Italy; 5 Stella Maris Institute, Pisa, Italy; 6 Istituto G. Gaslini, Genova, Italy; 7 Turin University, Turin, Italy; 8 Padua University, Padua, Italy; 9 Istituto Mondino, Pavia, Italy; 10 IRCCS Bosisio Parini, Lecco, Italy; 11 Maggiore Hospital, Bologna, Italy; 12 Istituto Besta, Milano, Italy

While there have been considerable advances for ambulant children with Duchenne Muscular Dystrophy (DMD), no prospective study has so far been devoted to outcome measures in non ambulant patients, with increasing complaints from families and patients. This information appears to be relevant not only for a better understanding of the disease progression but also for possible enrolment of patients in future trials.

As a result of an international effort, a new tool, the Performance of Upper Limb (PUL) was specifically designed to assess upper limb function in DMD boys. The purpose of the PUL is to assess changes that occurs in motor performance of the upper limb over time from when a boy is still ambulant to the time he loses all arm function when non-ambulant.

The aim of the present study was to use the PUL in:

(1) a cohort of typically developing children from the age of 3 years onwards in order to identify the age when the activities assessed in the individual items are consistently achieved.

(2) a cohort of DMD children and young adults to assess the range of findings at different ages.

We collected normative data for the scale validation on 258 typically developing children from 3 to 14 years old. A full score was consistently achieved (>85%) by the age of 3.5 years. Below the age of 3.5 years there was a significant number (>15%) who had difficulties in the items involving stacking cans, opening a Ziploc container, tearing a piece of paper and in lifting the heavier weights. After the age of 3.5 years difficulties were only occasionally found in tearing a sheet of paper.

When the PUL was performed in the 211 DMD patients (age range 2.8-23 years), we observed a progressive deterioration of scores with age, with early involvement of the proximal muscles that was more obvious after the age of 10 years. Even the oldest and weakest DMD patients were still able to perform some of the distal items, suggesting that the scale is capable of measuring small.

http://dx.doi:10.1016/j.nmd.2013.06.408

P.2.4
Upper extremity reachable workspace evaluation in DMD using Kinect
G. Kurillo 1, L.J. Han 2, A. Nicorici 2, L.B. Johnson 2, R.T. Abresch 2, E.K. Henricson 2, C.M. McDonald 2, R. Bujovc 1
1 UC Berkeley, College of Engineering, Berkeley, United States; 2 University of California Davis, Physical Medicine and Rehabilitation, Sacramento, United States

There is a lack of outcome measures evaluating upper extremity function in Duchenne Muscular Dystrophy (DMD) and a need to develop a quantitative low-cost method to evaluate upper extremity function in clinical trials.

We developed a 3D reachable workspace tool to quantify joint mobility within the patient’s functional space. Reachable workspace is defined as all points relative to the torso an individual can reach by moving their hands, and its envelope is defined by the encompassing surface area. Reachable workspace is associated with functional upper limb status and is applicable in evaluating patients with DMD, where weakness and dysfunction stems from early involvement of shoulder girdle muscles.

We developed a system based on the Microsoft Kinect to capture upper-limb movement during a specially devised workspace protocol. We are able to fit spherical surface data to the hand trajectory. The resulting envelope is quantified as a surface area using 3D surface plots projected to anatomical body planes. Surface area is normalized to the unit of the hemisphere of hand movement to facilitate comparison between individuals.

We collected data in 8 pediatric subjects with DMD aged 6–13 years (mean: 9.4 ± 2.5 years). All subjects were able to follow the movement protocol for assessment of the reachable workspace. Average normalized surface area for the dominant side was 0.749 (SD: 0.063) with a minimal area of 0.627 obtained in an 11-year-old. Maximal area of 0.883 was obtained in another 11-year-old subject with milder phenotype. Performance of the same movement protocol in a healthy control boy showed normalized surface area of 0.884, and in the adult control group (n = 10), the average normalized surface area reached 0.679 (SD: 0.090).

To increase the sensitivity to detect subtle weakness we measured the workspace with loading condition (500 g wrist weight). Further studies