208P

Are people living with neuromuscular disorders in the north of England satisfied with National Health Service wheelchair service provision?

J. Michell-Sodhi, D. Moat, T. Dias, J. Mason, E. Robinson, M. Schiava, E. Barr, C. Bolano, S. Doaa, K. Wong, E. Maha, G. Emma, G. Michela, M. Michelle, G. Tasca, J. Diaz-Manera, M. James, V. Straub, C. Marini-Bettolo, R. Muni-Lofra
John Walton Muscular Dystrophy Research Centre, Newcastle upon Tyne, UK

People living with neuromuscular disorders (NMD) often describe weakness, reduced mobility, falls, pain and fatigue as factors requiring mobility support with a wheelchair (WC). The timely and appropriate provision of mobility aids, including WC can help manage symptoms and improve quality of life for both patient and care givers. Optimising WC provision is essential in facilitating independence in functional ability, participation and activities of daily living. This is particularly relevant for those who are reliant on powered wheelchair (PWC) provision during waking hours to enable independent mobility. The specific WC prescription needs of persons with NMD are not well documented. Within the UK, regional variations in WC provision exist leading to disparity and dissatisfaction with the provided service. The aim of this project was to understand National Health Service WC provision and level of satisfaction with WC prescription in our clinical cohort. We surveyed 149 wheelchair users with NMD living in the North of England between 2019-2023. Respondents were aged 3-81 years, with a mean age 33 years. 42 respondents were paediatric. Diagnoses included Duchenne muscular dystrophy (45), spinal muscular atrophy (22), myotonic dystrophy (14), limb girdle muscular dystrophy (12), and Becker muscular dystrophy (13). 82 respondents used PWC, 49 a manual WC (MWC) and 17 power-assisted MWC. A third of respondents felt that their needs were not met by their current WC prescription. 15% of respondents reported delay in WC provision. This survey highlights concern at current practises in WC provision in the North of England and the need to determine evidence-based best practice guidelines for WC prescription for persons with NMD more generally.

http://dx.doi.org/10.1016/j.nmd.2024.07.059

209P

Custom orthosis improves mobility and caregiver experience in an adolescent with congenital myasthenic syndrome and myofibrillar myopathy

V. Varghese [1], I. Rajadurai [1], M. Paul [1], S. Banavara Shyamprasad [2], A.A. Mathew [1,2]
[1] *Bangalore Baptist Hospital, Bengaluru, India;* [2] *Synapse Neuro and Child Development Centre, Bengaluru, India*

Weakness and their attendant contractures are the norm in neuromuscular disorders. Each disease entity follows its natural history in how contractures form consequent to the pattern of muscle weakness. Both Congenital Myasthenic Syndrome (CMS) and Myofibrillar Myopathy (MFM) are rare neuromuscular disorders, characterized by muscle weakness and fatigue and challenging on their own. Here we present a child who had a dual genetically proven pathology, with corresponding phenotype of both CMS and MFM. This presented some unique hurdles faced by this adolescent who had these conditions. This necessitated a novel custom orthotic intervention design from our team, which had a beneficial impact on his mobility and caregiver experience. The Assessment of Caregiver Experience with neuromuscular Disease (ACEND) score assesses the impact for a caregiver of tending to a child with a neuromuscular disease1. It is a 41-item questionnaire covering seven domains. An eleven-year-old boy diagnosed with CMS and MFM presented with progressive proximal muscle weakness, facial, bulbar, and extra-ocular muscle weakness as well as ptosis. He had anatomical variations in his posture, including an exaggerated cervical extension, thoracic kyphosis and a pelvic tilt. This consequently exacerbated his motor difficulties, resulting in frequent falls and a reluctance to ambulate. A custom-made cervical-thoracic orthosis was prescribed, comprising of a cervical extension of fifteen degrees along with a thoracic extension to support the spine. This orthosis aimed to improve posture, enhance stability, and reduce falls during ambulation. Following this orthotic intervention, the visual 'access' of the child improved. He demonstrated significant improvement in mobility, walking indoors and walking without support; it further enabled him to ambulate independently in unfamiliar environments with reduced assistance. Assessment using the ACEND score revealed a notable enhancement in caregiver perception of physical impact, particularly in the domain of mobility. These advancements empowered the child to independently carry out school and home self-care activities. The newfound ability instilled a sense of confidence in both the subject and caregiver, marking a significant milestone in their journey towards independence. A Histogram revealed that the data did not follow normal distribution. Hence, we applied Wilcoxon Signed rank test to assess pre and post intervention scores. The alpha was set at 0.05. We found a statistically significant difference between pre-and post- intervention scores. We found the critical value less than the test statistic value ($+/-4.3 < 5$), thus validating the efficacy of the orthotic intervention. This case emphasizes the importance of tailored orthotic interventions in neuromuscular conditions such as CMS and MFM. Custom orthotic intervention can significantly improve mobility and caregiver experience. Further research is warranted to elucidate the long-term efficacy and benefits of orthotic interventions in this patient population.

http://dx.doi.org/10.1016/j.nmd.2024.07.060

210P

Advancing upper limb motor function evaluation in Duchenne muscular dystrophy and spinal muscular atrophy via kinematic parameterization with the wearable device "ArmTracker"

D. Natera De Benito [1], A. Favata [2], J. Expósito-Escudero [1], R. Gallart [3], O. Moya [1], S. Roca [1], A. Marzabal Gatell [2], L. van Noort [2], C. Ortez [1], C. Torras [3], A. Nascimento [1], J. Medina-Cantillo [1], R. Pàmies Vilà [2], J. Font-Llagunes [2]
[1] *Neuromuscular Unit, Barcelona, Spain;* [2] *Biomechanical Engineering Lab, Department of Mechanical Engineering and Research Center for Biomedical Engineering, Universitat Politècnica de Catalunya, Barcelona, Spain;* [3] *Institut de Robòtica i Informàtica Industrial, CSIC-UPC, Barcelona, Spain*

Assessing upper limb motor function in Duchenne Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA) traditionally relies on the Performance of Upper Limb (PUL) and Revised Upper Limb Module (RULM) scales, respectively. While considered gold standards, using these scales in isolation presents some challenges, notably in capturing subtle changes in motor function over time or in response to treatments. Inertial Measurement Units (IMUs) are inertial sensors that provide objective and quantifiable movement data. We hypothesize that integrating IMU measurements into these scales could provide complementary data for a more comprehensive assessment of motor function in individuals with neuromuscular diseases. Ten children with DMD (aged 12-17, Brooke score 2-5), 10 children with SMA (aged 6-13, Brooke score 2-5), and 6 healthy control children (aged 5-17, Brooke score 1) performed the PUL and RULM scales while wearing the ArmTracker device, equipped with 7 IMUs (Xsens Dot, Xsens Technologies) placed on the back of the hands, forearms, arms, and torso. Each IMU provided quaternion data. A sensor-to-segment calibration process was conducted with subjects seated in a chair with forearms resting on a table. Photographs were taken in frontal and lateral planes during calibration, and this visual information was integrated with IMU data to enhance calibration accuracy. Euler angles YZ'Y'' for the shoulder, and ZX'Y'' for the elbow and wrist were utilized. Maximum reachable area of hands, workspace area, and range of motion of shoulder, elbow, and wrist were evaluated and correlated with motor function scale scores using the Spearman correlation coefficient. Workspace area was normalized by the maximum achievable area by individuals (in %). We found significant correlations between the workspace area of upper limbs, notably on the frontal plane, and the scores of both the PUL and RULM. Additionally, significant correlations were observed between the scores of both motor function scales and the range of motion (ROM) of the shoulder. Furthermore, a correlation matrix analyzing the angles of the three primary upper limb joints revealed compensation patterns, which proved particularly valuable in identifying compensatory movements during the shoulder abduction task within the scales. Employing inertial sensors during the administration of functional motor scales in individuals with neuromuscular diseases yields valuable variables for assessing motor function, with particular interest in workspace area of the upper limbs and ROM of the shoulder. These and other variables are currently under further investigation within the same cohort of individuals who have undergone evaluations at home and school spanning four days utilizing the ArmTracker. This ongoing research aims to ascertain the system's potential for conducting assessments at home and assessing real-life movements in everyday scenarios.

http://dx.doi.org/10.1016/j.nmd.2024.07.061

211P

Following patient mobility in daily life: the EJP-DT4RD project

J. Hogrel [1], R. Muni-Lofra [2], P. Santmarty [1], V. Decostre [1], T. Marques [1], E. Haf Davies [3], V. Straub [2], the DT4RD Project Group
[1] *Institute of Myology, Paris, France;* [2] *John Walton Muscular Dystrophy Research Centre, Newcastle, UK;* [3] *Aparito, Wrexham, UK*

The DT4RD (Digital Tools 4 Rare Diseases) project is dedicated to the development of a non-invasive toolbox that measures patient mobility in daily life. Although tremendous progress has been made in this regard in the last few years, there is still a strong need to obtain disease-related information that generates fundamental clinical and patient-relevant outcomes accepted by regulators. Such data can be harnessed through digitised means using tools such as wearable devices, sensors, videos and smartphone apps. For example, movement sensors in the patient's home, worn on the body, or mobility aid can assess general movements whilst distinguishing between the patient's voluntary and involuntary actions. The development of novel clinical outcome assessments (COA) has the potential to facilitate the drug development process across rare diseases and