Ariaudo, G. [1,*]; Orcesi, S. [1]; Gorni, K. [2]; Rossi [1]; Berardinelli, A. [2]; Beghi, E. [3]; Lanzarotti, C. [3]; Angelini, C. [4]; Bertini, E. [5]; Pini, A. [6]; Palmucci [7]; Mercuri, E. [8]; Fazzi, E. [1]; Lanzi, G. [1]
[1] *Neurological Institute "C. Mondino", Child Neurology and Psychiatry, Pavia, Italy;* [2] *Neurological Institute "C. Mondino", Regional Neuromuscular Center for Children, Pavia, Italy;* [3] *Mario Negri Institute, Milan, Italy;* [4] *Neuroscience, University of Padua, Padua, Italy;* [5] *Bambin Gesù Children's Hospital, Rome, Italy;* [6] *Maggiore Hospital, Child Neurology and Psychiatry, Bologna, Italy;* [7] *University of Turin, Neuroscience Department, Turin, Italy;* [8] *Gemelli Hospital, Child Neurology and Psychiatry, Rome, Italy*

Self-report questionnaires are regarded as the primary method of measuring HRQoL, but there are very few instruments with a self report form for children under 8 years old, so usually HRQoL is evaluated by proxy report. The SOLE questionnaire is a new and original self-administered questionnaire for the study of HRQoL in children with neuromuscular disorders (NMDs) and allowed us to compare parent's and children's answers and evaluate the agreement between them. Evaluate the agreement between child and parent reports on children's health-related quality of life (HRQoL) using a new instrument: The Strips Of Life with Emoticons (SOLE) questionnaire. The data were collected from a group of 78 NMD's patients aged 5–12 years and a group of 81 healthy children of the same age. Both children and parents answered to a specific form of the SOLE questionnaire and children's answers were compared with those of their parents using Kappa agreement analysis. In our sample all Kappa values were under a 0.6 significance level. For most of the questions the Kappa value was from −0.2 to 0.2 that means disagreement or "casual" agreement; only for few questions, mainly related to medical aspects and physical health/independence, there was a trend towards agreement (Kappa ~ 0,25). In our group Kappa values were not sufficiently high for any of the questions demonstrating a substantial disagreement between children and their parents. These results follow those highlighted in literature: parents are more able to rate the child's HRQoL in relation to physical functioning compared with less concrete areas such as social or emotional aspects. We therefore underline how self-report is fundamental to assess HRQoL also under 8 years of age and we promote the use of questionnaires that can provide this kind of assessment. This project was supported by Telethon UILDM Grant GUP 030556.

doi:10.1016/j.nmd.2007.06.359

G.P.15.17

Activity limitations in patients with neuromuscular disorders: A study on the responsiveness of the ACTIVLIM questionnaire

Vandervelde, L. [1,*]; Van den Bergh, P. [2]; Thonnard, J. [1]; Goemans, N. [3]
[1] *Université Catholique de Louvain, Laboratory of Rehabilitation and Physical Medicine, Brussels, Belgium;* [2] *Cliniques Universitaires St-Luc, Neuromuscular Centre and Neurology Department, Brussels, Belgium;* [3] *Universitair Ziekenhuis Gasthuisberg, Department of Child Neurology, Leuven, Belgium*

ACTIVLIM, a self-report scale of activity limitations, was recently developed and validated in children and adults with neuromuscular disorders. The purpose of the present study was to investigate the responsiveness of this questionnaire. It is of interest when considering the progressive course of the neuromuscular disorders and the new therapeutic possibilities for these patients. The activity limitations of 127 NMD patients were assessed twice with a delay of 21 ± 4 months using the ACTIVLIM questionnaire. The responsiveness was evaluated by using a paired t-test between the two evaluations and by correlating the measure provided by ACTIVLIM and the subjective impression of the patient. A significant difference in the ACTIVLIM measures was observed between the first and the second evaluation ($t = 6.6$, $p < 0.001$). The patients tend to have a lower activity level at the second evaluation. The measure changes were significantly but moderately correlated with the patient's impressions ($r = 0.42$, $p < 0.001$). Moreover, the activity limitations were significantly higher in patients reporting a subjective impression of deterioration ($t = 3.1$, $p = 0.002$). These results suggest that the ACTIVLIM questionnaire presents a good sensitivity to changes indicating that it can be used to characterize the disease evolution and to objectively follow the NMD patients. Moreover, ACTIVLIM has a good potential to measure the effects of new therapeutic interventions.

doi:10.1016/j.nmd.2007.06.360

POSTERS 22 ALPHA-DYSTROGLYCANOPATHIES

C.P.3.01

LGMD2I in a North American population

Feener, C. [1]; Kang, P. [2]; Estrella, E. [1,*]; Darras, B. [2]; Amato, A. [3]; Kunkel, L. [1]
[1] *Children's Hospital, Boston, Program in Genomics, Boston, United States;* [2] *Children's Hospital, Boston, Neurology, Boston, United States;* [3] *Brigham & Women's Hospital, Neurology, Boston, United States*

There is a marked variation in clinical phenotypes that have been associated with mutations in the *FKRP* gene, including severe congenital muscular dystrophies, Walker–Warburg syndrome, muscle–eye–brain disease, a severe form of limb–girdle muscular dystrophy type 2I (LGMD2I), and a mild form of LMGD2I. The *FKRP* gene encodes the Fukutin-related protein (FKRP) and is believed to participate in the glycosylation of α-dystroglycan in the muscle fiber. There is also evidence which suggests that the FKRP protein localizes to both the Golgi apparatus and the endoplasmic reticulum, with some debate over the possibility that mislocalization of FKRP plays a role in the pathogenesis of disease. It has been shown in several populations (UK, Germany, Denmark, Brazil) that the *FKRP* mutation, c.826C > A, p.L276I, is by far the most common mutation causing LGMD2I, either in homozygote form or as part of a compound heterozygote genotype. In this study, we screened the *FKRP* gene in two cohorts totaling 97 patients with the LGMD phenotype by PCR analysis followed by direct sequencing on the 3730 DNA analyzer of the entire coding region of the *FKRP* gene. The common mutation, c.826C > A, p.L276I appeared to be homozygous in six patients; the seventh patient was a compound heterozygote. One patient who appeared to have the homozygous mutation had early onset at 3 years, while the other patients, including the compound heterozygote, had later onset. We have similarly found that LGMD2I is common among a sample of previously undiagnosed cases of LGMD from the United States. These findings raise the possibility that LGMD2I is as common in North America as in Europe. Therefore this diagnosis should be considered early in the evaluation of LGMD.

doi:10.1016/j.nmd.2007.06.361

C.P.3.02

Expanding the clinical spectrum of *POMT1* and *POMT2* phenotype: A multicentric study in the Italian population

Messina, S. [1,*]; Mora, M. [2]; Pegoraro, E. [3]; Pini, A. [4]; Mongini, T. [5]; D'Amico, A. [6]; Pane, M. [7]; Bruno, C. [8]; Biancheri, R. [8]; Berardinelli, A. [9]; Toscano, A. [10]; Morandi, L. [2]; Moroni, I. [2]; Farina, L. [2]; Uggetti, C. [9]; Pichiecchio [11]; Scuderi, C. [12]; Ruggieri, A. [13]; Muntoni, F. [14]; Santorelli, F. [6]; Bertini, E. [6]; Mercuri, E. [7]
[1] *Department of Paediatric Neurology, Catholic University, Rome and Department of Neuroscience, Psychiatry & Anesthesiology, University of Messina, Messina, Italy;* [2] *Myopathology and Immunology Unit, National Neurological Institute C. Besta, Milan, Italy;* [3] *Department of Neurosciences and Psychiatry and Anaesthesiology, University of Padua, Italy;* [4] *Child Neurology and Psychiatry Unit, Maggiore Hospital, Bologna,*