Understanding factors hampering activities of daily living performance in childhood-onset myotonic dystrophy phenotypes

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Myotonic dystrophy type 1 (DM1) is the most prevalent neuromuscular disorder among adults, with three phenotypes presenting symptoms during childhood. It is a multisystemic disease, with a complex presentation of system involvement, severity, and progression of signs and symptoms. Recent studies have clearly suggested the need for phenotype-specific clinical profiles to help with prognostic and long-term follow-up related to health and independent living for this population, among other issues.[1] The study by Eriksson et al.[2] provides key information that will facilitate better support for affected individuals and caregivers towards optimizing independent living. The authors have documented, for the first time, the level of motor and process skills that hinder the performance of activities of daily living among individuals with the congenital or childhood phenotype of DM1.

One key finding is the identification of markedly deficient process-skills, including the ability to initiate, which is strongly associated with the concept of apathy. Despite an existing positive relationship between apathy and executive dysfunction levels, the specific difficulty in initiating goal-directed behaviours, leading to a reduction in everyday productivity, may be due to the behavioural and cognitive dimensions of apathy in DM1.[3]

Although present in the original 1916 definition by Rohrer, apathy was rarely assessed formally and its association with independent living was not investigated until recently.[4] A recent systematic review showed that a clinically significant level of apathy is present in 55% of patients with DM1. However, apathy has never been studied in young patients with DM1, nor in patients with the childhood and congenital phenotypes (children or adults).

If we consider the results from Eriksson et al.'s[2] study, we could hypothesize that apathy may occur in young patients with the congenital and childhood phenotypes of DM1. Further studies should consider assessing process skills, apathy, and activities of daily living concurrently, and as early as possible in relation to age and disease stage. If apathy is identified during childhood and at the first stage of the disease, this could shed new light on apathy, which has been perceived as a feature of neurodegenerative diseases rather than neurodevelopmental ones. The better understanding of apathy in congenital or childhood phenotypes may be highly relevant clinically, since non-pharmacological management of apathy has shown promising effectiveness and should be explored in the population with DM1.

This study is also important in regard to potential modification of service provision to this population. All participants were living either with their parents or in a group home. This is different from the study by Gagnon et al., where 44% of adults were living with a spouse or alone.[5] Eriksson et al.[2] found that 62% of their population had a predicted need for help to live independently in the community. This is lower than that reported in the retrospective study of Gagnon et al. where 91% of participants needed help to accomplish house-related tasks independently.[5] These diverging results could be related again to the concept of apathy, where participants are able to perform an activity in a standardized situation, but will not pursue this activity at home because of lack of interest or will. Therefore, service provisions could help decrease the burden experienced by parents or spouses, or improve the conditions of participants living alone. In addition, the study clearly highlights the need to base the evaluation for service provision not only on motor skills, but also on process skills, including initiation and problem-solving ability.

This study highlights the need to further assess participation in this population and the role of occupational therapists as key players in providing guidance for patients and their families.

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