



Karolinska Institutet

**Institutionen för neurobiologi, vårdvetenskap och samhälle,
sektionen för sjukgymnastik**

Disability and Physical Exercise in adults with Myotonic Dystrophy type 1

AKADEMISK AVHANDLING

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ABSTRACT

Background: Myotonic dystrophy type 1 (DM1) is an inherited, slowly progressive, multi-system disease. There is no overall picture of its effect in individuals, and there is a lack of scientific evidence to support recommendations on physical exercise.

Aim: The aims of the work described in this thesis were to explore aspects of functioning, disability and contextual factors in adults with DM1 with regard to different stages of the disease; to describe the reliability and feasibility of the six-minute-walk test (6MWT), and to evaluate the feasibility and effects of a physical exercise programme.

Methods: Seventy adults with DM1 were assessed using various methods and measures, including a modified ICF checklist, tests and questionnaires. The reliability of the 6MWT was evaluated in 12 persons with DM1, and its feasibility in another 64. A comprehensive group exercise training programme, the Friskis&Svettis® Open Doors programme, was evaluated in 35 adults with DM1. They were assigned by lot to either a training group (18 persons) or a control group (17 persons). The training group participated in the exercise programme for 60 minutes twice a week during 14 weeks. The 6MWT was the primary outcome measure. Stages of disease progression were in all the studies based on the disease-specific muscular impairment rating scale.

Results: Excessive daytime sleepiness, muscle weakness and fatigue were common body-function impairments. Activity limitations were most frequently found in physically-demanding mobility activities. Few reported participation restrictions. Support from the immediate family was the most important facilitator for functioning. The individual's total number of impairments, limitations and restrictions was high and persons with severe muscular impairment had more impairments and limitations/restrictions than did those with mild. The 6MWT was reliable and feasible. The better of two possible trials was identified for use as test result. A difference of 33 metres or 6% for an individual with DM1 for a change not to be ascribed to measurement error is suggested. The Open Doors programme was well tolerated and no detrimental effects were found. Intention-to-treat analyses revealed no significant between-group differences in the primary outcome measure. Six persons in the training group and two controls increased their 6MWT distance by $\geq 6\%$. Many participants in the training group experienced subjective improvements and could recommend this form of physical exercise to others with the same disease.

Conclusion: The finding of a wide variety of body-function impairments, activity limitations and participation restrictions underlines the multi-systemic nature of the disease and the vast impact it has on health. It further implies that a multi-professional approach is needed for optimal care. The information can be used for developing clinical practice and for health promotion for people with DM1. The 6MWT was reliable and feasible, and can be used as an outcome measure in adults with DM1. The Friskis&Svettis® Open Doors programme was feasible for adults with DM1 who had been screened for cardiac involvement, had distal or mild-to-moderate proximal muscle impairment and no severe cognitive impairments.

Keywords: classification, disability, functioning, ICF, myotonic dystrophy, physiotherapy, physical exercise, rehabilitation, reliability