

Assessments in myotonic dystrophy (DM). A disease with many faces

By Diderichsen, Jørgen

People with DM represent huge challenges for diagnostic work up, medical follow up and multi-professional co-working. How can we meet these challenges?

FRAMBU is a National Center and meeting place for families with rare diseases, among these DM.

We have developed and introduced a preliminary interview guide to catch up with the many aspects of this multi faced disease and uncovered a vast amount of problems in the families we met in the last 2 years which had not been noticed in the care system. We developed this guide with a background on involved body-functions, activities and participation (IC-F) and how they influence the transitional phases from childhood during adolescence to adult life. This is what the planned Scandinavian reference program on DM tries to meet.

We tried to compare the results of our guide with the Myotonic Dystrophy Care Pathway, which we did find too narrow in its medical approach.

The most important issues we conclude are needed are:

Identification of resources and weaknesses, worries and subjective needs in a comprehensive but systematic way. Identify tracks in the disease process on an individual basis and based on groups of individuals with the same variant of the disease to make more helpful prognostications.

To develop means of evaluating Q-o-L for comparing and developing optimal care programs.

Tools for better management and care.

To attain these means we propose the following headlines in future care plans:

When (age) were the first symptoms or signs noticed and which were they.

When was the diagnose DM settled and which investigations were done.

Age, when important body functions and activities were disabled.

1. Gross motor (to run, walk staircases, carry school luggage, dress and undress
2. Eat and drink with no trouble. Pains in the stomach region
3. Digestion and stools
4. Respiratory functions
5. Pains in muscles and/or joints
6. Speech and other language functions
7. Attention and learning capacity/cognitive problems
8. Visual functions
8. Psychological or psychiatric difficulties
9. Endocrinological disturbances.

*(Guide manual for children and adults in Norwegian language is placed beside the poster).

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