Background

- Myotonic dystrophy is the most common inherited muscle disorder characterized by myotonia with dystrophic involvement of muscles and other multisystem manifestations. Myotonic Dystrophy is associated with cognitive symptoms, cardiologic manifestations such as conduction defects, tachycardia, and lack of initiative and cognitive symptoms.
- The Scandinavian guidelines on Myotonic Dystrophy were developed to standardize treatment and physical examinations in different areas of health conditions related to Myotonic Dystrophy. The program is planned to be finally presented in 2008.

Aim

- The focus of this study was to investigate to what extent people with Myotonic Dystrophy in Denmark are followed with regular physical examinations by a medical doctor (neurologist, cardiologist, ophthalmologist, and respiratory expert). Furthermore the patients were asked whether they were trained by a physiotherapist, whether they felt fatigued during the day and whether they suffered from impairment of memory.

Methods

- A questionnaire, based on recommendations in The Scandinavian Guidelines on Myotonic Dystrophy, was used in this pilot study.
- The questionnaire answered mainly with yes or no was given to a study group of 25 adults with Myotonic Dystrophy Type 1 who attended a seminar for adults diagnosed with Myotonic Dystrophy within the previous 5 years.
- The questionnaire was answered by 22 of the 25 participants.

Results

Table 1
Percentage of patients’ answers:

<table>
<thead>
<tr>
<th>Question</th>
<th>Yes (%)</th>
<th>No (%)</th>
<th>Unknown (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Myotonia</td>
<td>55</td>
<td>36</td>
<td>9</td>
</tr>
<tr>
<td>Cardiologist</td>
<td>23</td>
<td>48</td>
<td>14</td>
</tr>
<tr>
<td>Respiratory specialist</td>
<td>58</td>
<td>41</td>
<td>9</td>
</tr>
<tr>
<td>Ophthalmologist</td>
<td>18</td>
<td>77</td>
<td>5</td>
</tr>
<tr>
<td>Physiotherapist</td>
<td>58</td>
<td>41</td>
<td>9</td>
</tr>
</tbody>
</table>

Table 2
- Shows that 95% suffered from daytime fatigue and 82% fell asleep in front of the television every night. 73% woke up non-refreshed in the morning and 50% had trouble remembering.
- Asked whether they knew which medicine to avoid when diagnosed with Myotonic Dystrophy, 45% did not know.
- Asked how many hours of sleep they get in 24 hours, 68% answered more than 9 hours (up to 14 hours).

Discussion

- This pilot-study only included a rather small group of study subjects and should therefore be considered guiding on the subject.
- Furthermore, it is uncertain whether everybody with Myotonic Dystrophy is able to answer a questionnaire. That is the reason why the relatives were asked to help answering the questionnaire.
- The fact that 50% had trouble remembering, that 73% of the group were not well awake in the morning and that 95% were tired during the day indicate that regular medical check-ups are crucial.
- There are various reasons for not having the necessary medical check-ups in Denmark:
  1. Knowing the nature of the disease (lack of initiative and cognitive symptoms), the person does not stay in contact with the controlling doctors. It may not remember the dates for controls.
  2. Patients may have difficulties reading and understanding a call for control, for instance if they live alone.
  3. The medical doctors who see the patients, for instance the General Practitioner, do not know enough about the disease in order to refer him/her to the relevant specialist departments.

Plan for pedagogic effort

- A pamphlet which in very short terms, accompanied by line-drawings, describes the necessary medical check-ups, why they are important and how to get in contact with the relevant medical departments. The pamphlet should be distributed to people with Myotonic Dystrophy registered in Denmark and in contact with the relevant medical departments. The pamphlet should be distributed to General Practitioners, neurological departments at hospitals and other relevant departments.

- A pamphlet written for medical professionals who informs about the most important recommendations as suggested in The Scandinavian Guidelines for Myotonic Dystrophy – to make them aware of its existence. This is scheduled for late 2008 and should be distributed to General Practitioners, neurological departments at hospitals and other relevant departments.