Psychopathological and emotional deficits in myotonic dystrophy

C Bungener, R Jouvent, C Delaporte

Abstract

Objective—To evaluate psychopathological disturbances in patients with myotonic dystrophy (MD) and compare patients with MD to both patients with facioscapulohumeral dystrophy (FSHD) and healthy control subjects.

Methods—A semistructured interview was used to determine DSM III–R criteria for major depressive episodes, dystymic episodes, and generalised anxiety. The Montgomery and Asberg and the Hamilton depressive scales, the Covi and Tyrre anxiety scales, the Abrams and Taylor scale for emotional blunting, and the depressive mood scale were all used in the study. Subjects were also asked to complete questionnaires for physical and social anhedonia.

Results—Fifteen patients with MD, 11 patients with FSHD, and 14 healthy subjects were studied. Patients with MD were not more depressed or anxious than healthy controls. Patients with FSHD were the most depressed and most anxious. However, patients with MD had significantly lower scores for expressiveness and significantly higher scores for anhedonia than the other two groups.

Conclusion—Patients with MD did not present significant depressive or anxious symptomatology but rather an emotional deficit. This emotional deficit may be an adaptive reaction to the threatening implications of the disease, or the effect of the CNS lesions which occur with MD, or both.

(J Neurol Neurosurg Psychiatry 1998;65:353–356)

Keywords: depression; emotional deficit; myotonic dystrophy

Myotonic dystrophy (MD), an autosomal dominant disorder, is the most common adult form of muscular dystrophy. The main features of the disease are muscular weakness, atrophy, and myotonia. The disease is also a multisystem disorder; the pathogenesis is varied and includes cataracts, and endocrine, cardiovascular, and neurological abnormalities with both cognitive and affective changes.

The first description of behavioural abnormalities and marked emotionality in patients with MD was made by Steinert in 1909. Other authors later found a range of mental disturbances: moodiness, suspiciousness, dullness, apathy, excessive somnolence, lack of motivation, and diminished mental capacity.\(^1\)\(^2\) Ritteimoser,\(^3\) who investigated the psychologi-
### Table 1 Demographic characteristics

<table>
<thead>
<tr>
<th></th>
<th>MD (n=15)</th>
<th>FSHD (n=11)</th>
<th>Controls (n=14)</th>
<th>ANOVA p Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age (y):</td>
<td>36.8 (11.3)</td>
<td>33.2 (8.3)</td>
<td>35.7 (13.3)</td>
<td>0.72</td>
</tr>
<tr>
<td>Range 6–17</td>
<td>20–54</td>
<td>21–48</td>
<td>20–55</td>
<td></td>
</tr>
<tr>
<td>Sex (n (%)):</td>
<td>67%</td>
<td>64%</td>
<td>64%</td>
<td>0.99</td>
</tr>
<tr>
<td>Women</td>
<td>5 (33)</td>
<td>4 (36)</td>
<td>5 (36)</td>
<td></td>
</tr>
<tr>
<td>Education (y):</td>
<td>11.6 (3.5)</td>
<td>11.9 (3.7)</td>
<td>13.9 (2.7)</td>
<td>0.16</td>
</tr>
<tr>
<td>Range</td>
<td>6–17</td>
<td>6–17</td>
<td>9–18</td>
<td></td>
</tr>
</tbody>
</table>

### Psychopathology

Major depression, dysthymia, and anxiety disorders were diagnosed according to DSM III-R criteria. The Montgomery and Asberg depression rating scale (MADRS) (range 0–60), the Hamilton depressive rating scale, 17 items (HDRS) (range 0–51), the Covi brief anxiety scale (range 0–4), the Taylor scale for emotional blunting (AT) (range 0–30), and the depressive mood scale (EHD) were used in the study.

The depressive mood scale (EHD) is a 20 item scale with 10 items assessing emotional changes expressed by the patient and 10 items assessing the emotional state as perceived by the investigator observing the patient’s facial reactions, speech, and motor expressiveness. Each item is rated from 0 (absent) to 4 (severe). The scale has a coherent factorial structure with five components: irritability, anhedonia, expressiveness, sadness, and affective hypoaesthesia. These five factors define two main dimensions: emotional deficit or blunted affect (combining anhedonia and hypoexpressiveness) and loss of control (combining irritability and hypoeffectiveness).

The emotional deficit combines a lack of emotional initiation and reactivity, affective monotony, and anhedonia. The EHD scale has been validated for other neurological diseases.

### Anhedonia

Recent studies have shown that anhedonia is part of depressive symptomatology. Chapman et al. made the distinction between physical and social anhedonia. For the purposes of the present study, anhedonia was considered, from the patient’s point of view, as an individual subjective appreciation of whether or not he or she was experiencing pleasure.

Anhedonia was assessed using two self rated questionnaires: the physical anhedonia scale (PAS) and the social anhedonia scale (SAS).

Both questionnaires have been translated and validated in French.

### STATISTICAL ANALYSIS

Statistically significant differences (p<0.05) were identified using analysis of variance (ANOVA). Fisher’s test was used for one to one comparisons of the three groups.

### Results

Demographic data on the three groups studied are detailed in table 1; 67% of patients with MD, 64% of patients with FSHD, and 64% of controls were women. Age, sex, and education levels were not significantly different for any of the three groups.

Table 2 gives the results of the psychopathological scales and anhedonia questionnaires. The muscle weakness of patients with MD showed three patients in stage 0, 10 patients in stage 1, and two patients in stage 2.

### DEPRESSION

One patient with MD met DSM III-R criteria for a major depressive episode, and two
Table 2  Psychopathological scales

<table>
<thead>
<tr>
<th></th>
<th>MD mean</th>
<th>FSHD mean</th>
<th>Controls mean</th>
<th>p ANOVA</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>(SD) n=15</td>
<td>(SD) n=11</td>
<td>(SD) n=14</td>
<td></td>
</tr>
<tr>
<td><strong>Depression:</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>HDRS</td>
<td>4.7 (4.8)</td>
<td>7.2 (7.1)‡</td>
<td>1.9 (1.6)‡</td>
<td>0.04</td>
</tr>
<tr>
<td>MADRS</td>
<td>6.0 (6.5)</td>
<td>8.3 (8.4)‡</td>
<td>1.9 (1.7)‡</td>
<td>0.04</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Anxiety:</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Tyrer</td>
<td>6.1 (5.6)</td>
<td>9.4 (8.9)‡</td>
<td>3.5 (2.4)‡</td>
<td>0.06</td>
</tr>
<tr>
<td>Covi</td>
<td>1.6 (1.9)</td>
<td>2.5 (3.1)‡</td>
<td>1.1 (1.1)</td>
<td>0.31</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Blunting:</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mood scale:</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Emotional deficit</td>
<td>6.7 (5.4)‡</td>
<td>2.5 (3.4)†</td>
<td>1.6 (2.5)†</td>
<td>0.004</td>
</tr>
<tr>
<td>Loss of control</td>
<td>5.2 (4.7)</td>
<td>5.9 (6.5)</td>
<td>2.5 (2.6)</td>
<td>0.16</td>
</tr>
<tr>
<td>Irritability</td>
<td>4.6 (3.7)</td>
<td>4.8 (5.0)</td>
<td>2.0 (2.2)</td>
<td>0.11</td>
</tr>
<tr>
<td>Anhedonia</td>
<td>2.3 (2.8)†</td>
<td>1.4 (2.3)</td>
<td>0.8 (0.9)†</td>
<td>0.07</td>
</tr>
<tr>
<td>Expressiveness</td>
<td>−3.9 (4.3)†</td>
<td>0.1 (2.5)*</td>
<td>−0.8 (2.1)†</td>
<td>0.007</td>
</tr>
<tr>
<td>Sadness</td>
<td>2.1 (1.5)†</td>
<td>1.9 (2.2)</td>
<td>0.9 (0.9)†</td>
<td>0.11</td>
</tr>
<tr>
<td>Anxious hyperaesthesia</td>
<td>0.7 (1.0)</td>
<td>0.8 (1.7)</td>
<td>0.1 (0.4)</td>
<td>0.27</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>ANHEDONIA</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Physical</td>
<td>20.5 (11.4)†</td>
<td>17.8 (8.8)‡</td>
<td>9.3 (6.5)‡</td>
<td>0.008</td>
</tr>
<tr>
<td>Social</td>
<td>12.3 (7.2)</td>
<td>12.8 (7.5)</td>
<td>8.1 (3.8)</td>
<td>0.12</td>
</tr>
</tbody>
</table>

*p<0.05 Fisher’s test MD v FSHD.
‡p<0.05 Fisher’s test MD v controls.
†p<0.05 Fisher’s test FSHD v controls.

Patients with FSHD for a dysthymic episode. No subject in the control group met DSM III-R criteria.

Ratings of depression (HDRS and MADRS) were significantly different for the three groups (MADRS (F(2,39)=3.65; p=0.04) and HDRS (F(2,39)=3.66; p=0.04)). Patients with FSHD scored the highest but only differed significantly from control subjects (Fisher’s test, p<0.05).

ANXIETY

No subjects met DSM III-R criteria for generalised anxiety. The Covi and Tyer anxiety scales showed no significant difference between the three groups, although results for the Tyer scale fell just short of significance (F(2,39)=2.38; p=0.11), although patients with MD were slightly but significantly sadder than controls (Fisher’s test, p<0.05). The anxious hyperaesthesia factor did not differ significantly between the three groups studied.

ANHEDONIA

Self questionnaires on anhedonia showed a significant difference in physical anhedonia for the three groups studied (F(2,39)=5.65; p=0.008) but not in social anhedonia (F(2,39)=2.29; p=0.12). In the physical anhedonia questionnaire, patients with MD and patients with FSHD scored significantly higher than control subjects (Fisher’s test, p<0.05).

Discussion

Patients with MD in the present study were not severely depressed according to DSM III-R criteria, although they did present symptoms of mild depression. Patients with MD on the other hand had no symptoms of anxiety, whereas patients with FSHD presented significantly greater symptoms of depression and anxiety than control subjects. These results concur with those of Cuthill et al who found that although symptoms of depression were common in patients with MD, few met the standard criteria for depression. Conversely, Brumback et al found a high incidence of major depressive episodes in the MD patient population studied.

In the literature there is a close association between depression and progressive diseases. Duveneck et al noted that this depressive symptomatology may arise through an emotional reaction to the disease. Patients with MD or other chronic diseases have to cope not only with physical restrictions and disabilities, but also with the financial and emotional ramifications of the disease; these may include loss of employment and income, lowered self esteem, and the possibility of a shortened life span. It is often difficult to clarify the origin of certain symptoms. Disturbances of sleep pattern (mostly hypersomnia), loss of appetite, and impaired memory and concentration are common in patients with neurological disease and whereas they may be symptoms of depression, they may also be related to the neurological disease itself.

Duveneck et al found that stress related symptoms and depressive reactions are common in multisystem diseases. Emotional disturbances—in particular emotional deficit—seem more common than typical depressive episodes. Patients with MD did, however, have a different emotional profile compared with FSHD and control subjects, with higher scores for emotional deficit. This deficit manifested as anhedonia and lack of expressiveness as evidenced by a monotonous mood, apathy, and an inability to anticipate pleasure. The emotional pattern of negative symptoms as noted here is...
similar to the apathy and lack of motivation previously found in MD.1 2 9 In the present study, these symptoms were not related to the presence of depression or anxiety. In fact, patients with MD with emotional deficit were neither depressed nor anxious, whereas the patients with FSHD who did not have emotional deficit were more depressed and anxious. A link may be seen between these results and the findings of the personality assessment. Four patients with MD displayed an avoidant personality disorder as opposed to none in the FSHD and control groups.31 Physical anhedonia was perceived in both patients with MD and patients with FSHD, but social anhedonia was the same for all three groups. The anticipation of future physical disability in the two patient groups may account for the differences in the ratings of physical, but not social anhedonia. This suggests that it is important to make the distinction between physical and social anhedonia.

There are different hypotheses for the cause of these emotional disturbances (other than the process of psychological adaptation to the disease). There may be a genetic cause involved in this particular emotional process. Ambrosini et al16 considered these psychiatric phenomena as a primary aspect of the disease and as the direct pathogenesis of the neuromuscular condition. These authors present clinical evidence that altered mental functioning is a basic feature of MD, rather than a reactive or secondary phenomenon; they described MD as an extensive “multiple dystrophy”. The CNS is a target, with specific neuropathology and psychopathology. In the present study, patients with MD were all in the early stages of the disease and, despite that, the CNS showed MRI abnormalities in nine patients.32 All patients with MD presented emotional deficit, whereas patients with FSHD, in whom the CNS is not affected, presented no deficit at all.

Conclusion

It seemed that patients with MD presented a characteristic emotional profile of emotional deficit, with the deficit appearing early in the disease and which could be interpreted as an adaptive psychological process or as a direct consequence of the CNS lesions caused by the genetic mutation. Close attention should be focused on these symptoms and their development and they should be taken into consideration in the care of chronic patients, because of the influence they have on their the physical and psychological wellbeing.

The present study has received support through a research grant from the French Myopathy Association (AFM). We thank Professor Bruno Eymard for referring the patients.


16 Tyrer P, Owen RT, Cicchetti DV. The brief scale for anxiety a subdivision of the comprehensive psychopathological rating scale. J Neurol Neurosurg Psychiatry 1984;47:970–5.


