Personality Patterns in Patients With Myotonic Dystrophy

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Background: Myotonic dystrophy (DM) is a multisystemic disease. The central nervous system is affected by cognitive, affective, and personality disturbances. A characteristic behavior was noted from the first clinical descriptions, but no definitive conclusions have been drawn despite extensive debate. As DM is a genetic disease of well-defined abnormality, it may be a good model for understanding the relative contributions of nature and nurture in the building of personality traits.

Objective: To investigate the possibility that there is a personality pattern that is characteristic of patients with DM.

Subjects and Methods: The personalities of 15 adult (age range, 20-53 years) patients with DM with no, or minimal, muscle weakness were studied by means of the International Personality Disorder Examination. The results were compared with those from 14 matched healthy subjects (age range, 20-54 years) and 12 patients (age range, 22-50 years) with a mild form of facioscapulohumeral dystrophy.

Setting: The Department of Neuromuscular Diseases, Hôpital de la Salpêtrière, Paris, France.

Results: Patients with DM exhibited a homogeneous personality profile, with statistically significant differences (P < .005) for avoidant, obsessive-compulsive, passive-aggressive, and schizotypic traits. In both groups of controls, the personality profiles were extremely heterogeneous. Personality disorders (avoidant personality) were found in 4 of 15 patients with DM.

Conclusions: The findings of this study suggest that the personality pattern of patients with DM is related to their disease. Their personality disorders are not attributable to their adjustment to a disabling condition. By contrast, among the patients with DM, the high incidence of avoidant personality, a phenotype poorly represented in the general population, supports the idea that it is the expression of a primary phenomenon related to a genetic mutation.

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Myotonic dystrophy (DM) is an autosomal dominant disorder that is the most common muscular dystrophy affecting adults (mean incidence, 1/20 000). Frequently, the primary symptoms are myotonia and progressive muscle weakness, but it is clear that DM is a multisystemic disorder, since its pathogenesis is varied, involving cataracts, endocrine deficiencies, cardiovascular manifestations, and the central nervous system, the known abnormalities of which concern cognitive processes, affective status, and personality. In terms of personality, behavioral disturbances were noted in the first clinical descriptions, but until the beginning of the 1980s, behavioral oddities were only mentioned in the clinical workup and were generally related to the severity of the muscular disability. The main character disorders described essentially involved suspicious attitudes, egocentricity, disagreeableness, or indifference.

At the end of the 1970s, the question of personality dysfunction was raised in an attempt to establish whether DM could be a primary disease of the central nervous system, as reported in a review of the literature on the psychopathologic manifestations of DM. Since this review, only 3 studies, to my knowledge, have been partly devoted to personality in patients with DM. One study that was focused exclusively on cognition and personality found that one third of the patients exhibited personality abnormalities that were prominent but without a characteristic pattern representative of the entire group. The results of a later study were in agreement with the described personality disorders but claimed that they were secondary to chronic and severe depression. A more recent study assessing cognitive deficits and personality pattern in 2 groups of patients with DM of maternal or paternal inheritance revealed no severe personality disorders, but did identify dependent traits among more than half of all the patients.

Interest in the study of personality dysfunction is increasing for 2 reasons. First, psychiatric physicians interested in how personality develops wish to establish the relative contributions of nature and nurture in...
SUBJECTS AND METHODS

SUBJECTS

Fifteen outpatients with DM (6 men, 9 women; mean age, 33 years; age range, 20-53 years) were included in the study. They were recruited from the Department of Neuromuscular Diseases, Hôpital de la Salpétrière, Paris, France, according to 2 criteria: (1) the absence of past and present psychiatric disease and (2) the extent of muscle deficiencies (the patients included exhibited a very mild form with no, or minimal, muscle weakness, rated as class I or II on the Muscular Disability Rating Scale. The diagnosis of DM was made on the basis of clinical examination findings, family history, electromyographic findings, and results of genetic analysis. Congenital forms of DM were excluded from this study, and the diagnosis was made after the age of 17 years in all cases. The interval between diagnosis and examination was not more than 5 years. Of those selected, 8 had so much minimal disturbing muscle weakness that their condition was diagnosed during a family survey. Their diagnosis was made on the basis of clinical data or electromyographic myotonic discharges. In 2 patients, a cataract was first diagnosed on the basis of colored lens densities at slit-lamp examination, and in 1 woman, the diagnosis was made at the birth of her child, who had a congenital form of the disease.

Twelve patients were the offspring of affected men, and 2 were the offspring of affected women. The sex of the potentially affected parent was unknown in 1 patient, but the pedigree indicated that the father was probably the transmitter. No differences in clinical symptoms were found between patients with paternal or maternal inheritance. In particular, maternal inheritance did not result in earlier onset or greater severity.

For all patients, the mutation on chromosome 19 was analyzed by Southern blot experiments and polymerase chain reaction. The results showed abnormally numerous repeats of the CTG fragment (from 260 to 1000). This high expansion was found in 2 patients with a clinical onset after the age of 25 years. One male patient exhibiting all the clinical and electromyographic signs of DM was nonetheless excluded from the study because fewer than 50 CTG repeats were found on DNA analysis.

The patients' educational level was almost representative of the general population: 6 had a university education, and 2 of the 6 had obtained a PhD. Only 1 displayed educational retardation. The 8 others had a high-school education (between the 9th and 12th grades). Four participants were unemployed. Three others had found employment below their qualification level. The characteristics of the 15 patients with DM are listed in Table 1. Two groups were used as controls: healthy subjects and patients whose disease exclusively involved the muscle apparatus ie, facioscapulohumeral dystrophy (FSHD). Fourteen healthy volunteers who were matched with the patients with DM for age, sex, educational level, and profession were prospectively recruited through an advertisement and were included after verification that they had no psychiatric history or disease involving the central nervous system. Their mean age was 35.7 years (age range, 20-54 years). No adequate control could be recruited for 1 patient, and no sibling without DM agreed to participate in this study.

Five men and 7 women with FSHD aged 33 ± 8 years (mean ± SD) were chosen because they exhibited minimal muscle deficiencies comparable in intensity to those of patients with DM, being classified as class I or II on the Muscular Disability Rating Scale. Although FSHD is the second most frequent myopathy in adults, there were too few appropriate cases to allow a paired association with the patients with DM. Nevertheless, persons in the same age range (20-50 years) were selected only if they were unaffected by psychiatric and/or central nervous system diseases.

The 3 groups of subjects were studied in accordance with the approved consent of the Salpêtrière Hospital's Ethics Committee for Human Research. The informed consent of the participants was obtained after the purpose of the study had been fully explained to them.

METHODS

A French translation of the International Personality Disorder Examination was used for personality assessment of the patients with DM and the controls. This questionnaire was developed by the World Health Organization for international use13 from the initial version, the Personality Disorder Examination, which was published in 1988.14

This test was chosen for 2 reasons. First, it allows diagnosis of personality disorders according to the criteria defined in the Diagnostic and Statistical Manual of Mental Disorders, Revised Third Edition,15 and the International Statistical Classification of Diseases, 10th Revision.16 Second, it is not a self-administered questionnaire but a standardized clinical interview.

The 1993 revised version of the International Personality Disorder Examination, containing 131 items, was used. The 144 questions are grouped in 6 areas as follows: work, self, interpersonal relations, affect, reality testing, and impulsive control. Through these items, the International Personality Disorder Examination systematically surveys the criteria of the life experiences and the behaviors relevant to a diagnosis of the personality disorders. The remaining 7 items pertain to subject behavior during the interview. Thus, by means of this semistructured interview, assessment is possible not only of personality disorders but also of personality profiles.

This test was also chosen because the methodology is extremely cautious: negative answers are scored 0, many behavioral examples, present for at least 5 years, are required for each positive answer to be scored as a probable (1) or definite (2) personality disorder. The advantage of this in-depth inquiry, with some similar questions, is that retesting is not necessary for the credibility of the results. However, the full examination is lengthy: around 3 hours for the patients with DM and a little less for the controls, who acknowledged only a few behaviors.

STATISTICAL ANALYSIS

The data obtained in the 3 groups of subjects were analyzed using a 1-way analysis of variance. Pairwise comparisons were performed between the patients with DM, the healthy subjects, and the patients with FSHD using a Student t test with the Bonferroni correction, and the significant threshold was chosen at a value of α = .003.
the building of personality traits. Personality can be regarded as the effect of life events on the temperament, which may be genetically transmitted. Consequently, the study of personality may reflect inherited traits if these are continuously found in a subject. The second reason is that DM, which has long been thought to be associated with a characteristic personality, is a genetic disease whose abnormality is now well identified. To summarize, DM is due to a mutation on chromosome 19, which has been identified as an unstable DNA sequence containing an abnormally high number of CTG repeats in the DM1 region.9 The protein product of the DM1 gene is believed to be a 72-kd “myotonin protein kinase.”10 The extent of expansion may roughly correlate with the severity of the disease and increases through generations. Such a disease may serve as a model in studies of the role of genetics in the personality development.

The present study was thus designed to investigate the possibility that there is a personality pattern that is characteristic of patients with DM. If so, it would be interesting to assess whether personality disturbances occur secondarily to the general course of the disease or can be considered as part of the disease. A group of adult patients without muscular disabilities was therefore investigated by means of a new standardized assessment specifically designed to detect personality disorders.

### Results

The International Personality Disorder Examination data were analyzed according to the Diagnostic and Statistical Manual of Mental Disorders, Revised Third Edition.15 Data analysis indicated a very homogeneous profile among the patients with DM, since only 5 patients differed in their answers. Each patient answered in the same way, agreeing or disagreeing more or less strongly with the proposal for all but 5 questions. For these 5 questions, some patients with DM were in agreement with the question, while others disagreed completely. It may have been more judicious to score these answers with a negative value rather than with 0.

The analysis of the personality profiles showed a prevalence of traits as part of both the anxious and the odd clusters, 2 of the 3 clusters organized by the Diagnostic and Statistical Manual of Mental Disorders, Revised Third Edition, with the 11 primary disorders. The scores allowed construction of a personality profile for the patients with DM, independent of any supposition regarding the existence of actual disorders.

The scores, whose individual values are reported in Table 2, indicated a characteristic profile for the 15 patients with DM, and analysis of each questionnaire item covering all personality traits showed the following: Among the traits of the anxious cluster, which comprises obsessive-compulsive, passive-aggressive, avoidant, and dependent characteristics, no criteria of the dependent traits were found, but the others were very marked. The most frequent characteristics noted in half or more of the patients with DM exhibiting obsessive-compulsive behavior were a high rigidity and stubbornness in the thoughts and attitudes of 14 of the 15 patients who acknowledged that these personality characteristics possibly caused problems in their relationships. Two thirds of the patients confessed that they were extremely scrupulous and even inflexible about matters of morality, or right and wrong. Most patients (12) demonstrated an inability to discard any objects, even those worn-out or worthless. The last noticeable trait was the extrovert-compulsive register, experienced by all patients but one, was difficulty in expressing warm emotions, a feeling, in fact, near the schizoid-schizotypic register.

The passive-aggressive traits that were present concerned the way the patients behaved at work. The most frequently encountered type of behavior (found in 9 of the 15 patients with DM) was a reluctance to do an irritating job, which was performed at a much slower rate, although all the patients asserted that their job was nev-
ertheless finished on time. Similarly, more than half recognized that they became sullen and argumentative when asked to do something they did not want to do. Also, 8 of the 15 patients experienced marked resentment when they were given useful suggestions to improve their work productivity.

The most frequent avoidant criterion was a difficulty in communicating with others. Thirteen patients said they often remained silent when in company because of a fear of having to answer a question and of saying something inappropriate or foolish. Similarly, 10 patients acknowledged a hypersensitivity to criticism. Another prevalent avoidant trait (10 patients) was excessive worry about being rejected by others.

In the odd cluster, which includes schizoid, schizotypic, and paranoid traits, the main schizotypic disturbance felt by the patients was a frequent uncomfortable feeling in the presence of other people. In the same cluster, 11 of the 15 patients with DM said that, besides their first-degree relatives, they did not have close friends. Moreover, some patients occasionally demonstrated odd beliefs, which clearly influenced their lives.

More than two thirds of the 15 patients with DM showed a positive response for several paranoid items: almost all (n = 13) said that they upheld the defense of their personal rights with combativeness and great tenacity. For example, 14 of the patients admitted that they would bear a deep grudge for a long time if they were convinced that they were given something inappropriate or foolish. Similarly, 10 patients said they often remained silent when in company because of a fear of having to answer a question and of saying something inappropriate or foolish. In particular, they had a tendency to feel pathologic jealousy regarding their partner. This suspicious attitude engendered unfounded fear of being exploited by others. Nine patients acknowledged that they felt easily slighted.

In contrast to the above results showing many positive criteria among the anxious and the odd clusters, only 1 narcissistic trait was found with great frequency in the dramatic cluster of narcissistic, histrionic, and sadistic traits. This trait was present in 13 of the 15 patients with DM and concerned their reaction to criticism, which provoked rage, shame, and humiliation. This trait aside, the other criteria of the dramatic cluster were totally absent in 2 of 3 patients who did not recognize themselves in the proposed criteria. In some cases, they even claimed that their feeling was the exact opposite of the one proposed. In the remaining patient, rare criteria were found, without predominance of one or another.

Four patients fulfilled a sufficient number of criteria that were scored 2 in a personality category, thereby reaching the pathologic level and allowing diagnosis of a personality disorder. In these 4 cases, the criteria met were positive for avoidant personality traits. This represents 26.6% of the sample of patients with DM in this study. The diagnosis of personality disorder was only probable in 3 patients (20% of the group), and the fourth patient (representing 6.6% of the group) had a score indicating a well-determined disorder. The latter subject also exhibited enough criteria that were positively scored for the schizotypic traits to allow diagnosis of probable schizotypic disorder in addition to the determined avoidant disorder.

No relationship was found between the presence of a probable or definite personality disorder and the increase in the number of CTG repeats, since the subjects with a personality disorder were in the midrange, rather than the upper range, of CTG elongation. Similarly, the duration of the disease among these patients was not longer than that among the other study subjects.

The homogeneity of the personality profiles and the presence of the same disorder in nearly one third of the patients with DM contrasted with the findings in the 2 control groups. Analysis of variance of the mean values for each trait revealed statistically significant differences between the DM group and both control groups for the obsessive-compulsive, passive-aggressive, avoidant, and schizotypic traits and between the DM group and the control group of healthy subjects for the schizoid and paranoid traits (Table 3).

In contrast to the patients with DM, the healthy subjects and the patients with FSHD displayed neither a characteristic type of personality pattern nor a personality dis-

Table 2. Individual Values of Dimensional Scores in Patients With Myotonic Dystrophy

<table>
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<tr>
<th>Patient No.</th>
<th>1</th>
<th>2</th>
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<td>Obsessive-compulsive</td>
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<td>21</td>
<td>14</td>
<td>04</td>
<td>03</td>
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<td>14</td>
<td>48</td>
<td>02</td>
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<td>42</td>
<td>001</td>
<td>32</td>
<td>30</td>
<td>03</td>
<td>88</td>
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<td>Schizotypic</td>
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<td>25</td>
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<td>24</td>
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<tr>
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<tr>
<td>Histrionic</td>
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</tr>
</tbody>
</table>

*For an explanation of the score values, see the “Results” section.

Table 3. Mean Values of Personality Traits

<table>
<thead>
<tr>
<th></th>
<th>Patients With DM (n = 15)</th>
<th>Patients With FSHD (n = 12)</th>
<th>HS (n = 14)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Obsessive-compulsive</td>
<td>4.60 ± 1.60†</td>
<td>2.18 ± 1.40</td>
<td>1.79 ± 1.42</td>
</tr>
<tr>
<td>Passive-aggressive</td>
<td>3.33 ± 2.66†</td>
<td>0.91 ± 1.14</td>
<td>1.14 ± 1.10</td>
</tr>
<tr>
<td>Avoidant</td>
<td>4.33 ± 3.04†</td>
<td>1.82 ± 1.94</td>
<td>1.43 ± 0.94</td>
</tr>
<tr>
<td>Dependent</td>
<td>1.67 ± 1.45</td>
<td>1.36 ± 1.50</td>
<td>1.57 ± 1.16</td>
</tr>
<tr>
<td>Schizoid</td>
<td>2.27 ± 2.12‡</td>
<td>1.27 ± 1.79</td>
<td>0.93 ± 0.99</td>
</tr>
<tr>
<td>Schizotypic</td>
<td>4.47 ± 1.92‡</td>
<td>1.36 ± 1.57</td>
<td>0.93 ± 1.27</td>
</tr>
<tr>
<td>Paranoid</td>
<td>4.13 ± 1.81‡</td>
<td>2.64 ± 2.46</td>
<td>1.21 ± 1.63</td>
</tr>
<tr>
<td>Antisocial</td>
<td>1.00 ± 1.19</td>
<td>0.64 ± 1.80</td>
<td>1.14 ± 1.46</td>
</tr>
<tr>
<td>Borderline</td>
<td>1.00 ± 1.00</td>
<td>1.00 ± 1.55</td>
<td>0.71 ± 0.91</td>
</tr>
<tr>
<td>Histrionic</td>
<td>0.67 ± 0.98</td>
<td>1.36 ± 1.57</td>
<td>1.50 ± 1.56</td>
</tr>
<tr>
<td>Narcissistic</td>
<td>1.07 ± 0.88</td>
<td>1.00 ± 1.26</td>
<td>0.93 ± 1.33</td>
</tr>
</tbody>
</table>

*DM indicates myotonic dystrophy; FSHD, facioscapulohumeral dystrophy; and HS, healthy subjects.
†Value statistically different from those of the HS and FSHD groups (P<.005).
‡Value statistically different from that of the HS group (P<.005).
order. None of the healthy subjects closely matching the patients, especially in terms of education and occupation, showed avoidant personality. When a personality profile stood out, it appeared within the dramatic cluster and, in 3 cases, within the odd cluster, but never within the anxious cluster.

**COMMENT**

The findings of this study indicate that the personality of patients with DM differs strikingly from that of individuals who do not have this disease. This assertion is substantiated by 2 main findings: (1) the strong homogeneity in the personality profiles among the patients with DM, in contrast to the large variety in the personality profiles among the 2 control groups; and (2) the noteworthiness of the observed personality. The special profile concerned the odd and anxious clusters, and when a disorder appeared, it related in each case to the avoidant personality.

The first question is in regard to whether these results are representative of individuals with DM. This question can be addressed by considering patient recruitment. Fifteen patients with DM were selected on the basis of their muscle disturbances and the absence of psychiatric disease. All but 2 had nothing in common, with very diversified sociocultural surroundings, geographical separation, and, for 13, different families. Two patients were relatives (uncle and nephew), but displayed only 13 of 145 similar positive answers in their questionnaires, a number not higher than that found when their answers were compared with those of some other patients. Although the uncle was positive for a probable avoidant personality disorder, the nephew was beyond the pathologic scores, with scores between 0 and 3 for all personality traits, except for passive-aggressive behavior, which was scored 6. Consequently, the only relationship between the patients was their disease, and the homogeneous data gathered from a small sample of patients with DM show that the typical personality pattern that was found is actually representative of individuals with this disease. This finding strongly suggests that the personality pattern of patients with DM should be regarded as related to the disease.

The results of this study agree with those of previous investigators, but only in part. One area of agreement concerns the acknowledgement of the existence of a typical personality. Thus, Steiner et al described his patients with DM as demonstrating abnormal behavior with a marked emotionality. Curschmann described his patients as exhibiting moodiness, suspiciousness, and dullness. Later, Rohrer and Naegeli made the same observations and depicted their subjects as mellow, somnolent, and disagreeable. Maas and Paterson characterized their patients as cheerful and somewhat condescending. Rittmeister characterized his patients as mainly withholding and reluctant. These reports predate World War II, and, to my knowledge, no postwar research has been devoted to the personality of individuals with DM, perhaps for fear of a eugenic connotation. Until the 1980s, then, research on mental disturbances focused on cognitive functions and affect disorders. In 1983, Bird et al assessed personality with the Minnesota Multiphasic Personality Inventory and found that 32% of patients with DM had prominent personality abnormalities, the most elevated scales being for schizophrenia, hypochondriasis, and depression. Brumback and Wilson reported similar results. In 1994, Palmer et al compared the cognitive deficits and personality pattern in cases of paternally vs maternally inherited DM with the Millon Clinical Multiaxial Inventory. They found 1 personality disorder (paranoia) in 13 subjects, and 62% of their patients displayed elevated dependency scores without difference between maternal and paternal inheritance. Thus, when the presence of a type of personality and the possibility of a disorder are considered, our results are in agreement with those of previous authors.

There are, however, 2 differences. The first concerns the types of personality disorders that vary from one study to another. For example, I did not find dependent traits, whereas Palmer and colleagues observed a high incidence of dependent tendencies. This divergence is not as great as it would appear: all reported traits belong to the anxious and the odd clusters, and the differences may be attributable to the various personality tests used. More important are the dissensions about the origin of the personality disorders. There are longstanding controversies as to whether mental disturbances are primary or secondary to the other handicaps. Before discussing this point, it should be emphasized that this question was never formulated concerning the other disorders encountered in DM, such as endocrine, ocular, or cardiac disease, perhaps because precise anatomical findings or laboratory data were always recorded for these. The same is not true for mental disorders, the pathogenesis of which is subject to prolonged debate. Curschmann first suggested that DM was a multisystemic disorder comprising mental disturbances in addition to the other abnormalities. Rittmeister considered that lack of motivation was seemingly a primary symptom in the development of the disease. In a literature review, Ambosini and Nurnberg concluded that psychopathologic manifestations are a basic feature of DM and even hypothesized that these manifestations could be the result of a dysfunction of the thalamic nuclei. Nevertheless, most investigators consider that the mental abnormalities are secondary aspects of the disease. This hypothesis is especially mentioned for affects and personality. Bird et al believed that many personality problems of patients with DM were caused by their limitations in coping with their physically deforming and debilitating muscle disorders. Palmer et al concluded that personality disorders in patients with DM might, for the most part, be a reflection of problems brought about by the patients having to adjust to the disabling condition. The data from this study do not support this explanation. Indeed, the patients were carefully chosen as having no or very little muscle disturbances, and their ability to cope with the disease was excellent; in some cases, the clinicians noted that the patients were even able to achieve a certain distance from their condition. The subjects were also free from psychiatric diseases. Despite these recruitment precautions, a typical personality profile and disorders were still found, thereby supporting the assertion that personality disorders are primary manifestations in DM. An additional argument against the idea that DM personality traits and disorders are secondary to muscle disability is that the FSHD control group did not display any characteristic personality, despite an identical level of muscle damage.
When a personality disorder was found in a patient with DM, it was always the avoidant personality. This finding could raise a question. Indeed, in the general population, avoidant personality seems not to be widespread. In a study using a self-administered questionnaire, the Personality Diagnostic Questionnaire, no case of avoidant personality was found among a cohort of 235 subjects. In other publications, the rates vary with the survey methods from 0.4% to 1.3% among close to 1000 persons. By contrast, if the 15 cases included in this study are considered representative, the prevalence of avoidant personality appears to be 20 times higher in the DM population than in the general population. The Diagnostic and Statistical Manual of Mental Disorders, Revised Third Edition, specifies that the avoidant personality may be found among dysmorphic persons. In advanced DM, facial dysmorphism due to marked muscle wasting produces a dazed and idiotic appearance, thus possibly inducing social avoidance behavior that leads to an avoidant personality. However, none of the study patients displayed this state. The existence in DM of a behavioral phenotype that is poorly represented in the general population lends credence to the idea that it is the expression of a primary phenomenon directly related to the genetic mutation. Myotonic dystrophy could therefore be a candidate to join the group of genetic diseases with behavioral phenotypes, such as fragile X syndrome and Williams syndrome. In a case of this syndrome with a partial mutation, it has recently been demonstrated that the cognitive and behavioral impairments were precisely related to this mutation. Likewise, in DM, the abnormal elongation of the CTG triplets may cause cerebral dysfunctions.

Having an avoidant personality, or simply an odd-anxious personality profile, must affect behavior across a variety of domains, such as work, family, and recreation. This can lead patients with DM to withdraw from society. In terms of work and matrimonial status, this withdrawal was obvious among our patients: 5 held a job for which they were overqualified but which was more solitary than their initial occupation, and 11 lived alone after divorce or are still unmarried. Thus, beyond the theoretical findings, in practice, knowledge of the personal-ity of the patient with DM could allow a follow-up designed to optimize avoidance of behavior related to excessive social anxiety, leading to isolation.

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