Matters arising

Cognitive and personality function in myotonic muscular dystrophy

Sir: The recent article by Bird et al. presents a much-needed detailed neuropsychological and personality assessment of myotonic dystrophy patients. The authors should be complimented for undertaking study of this patient group in which cooperation is often difficult to obtain. We believe, however, that several comments upon their report are necessary. As noted by Bird et al., myotonic dystrophy is an autosomal dominantly-inherited multisystem disorder. Patients frequently present with non-neuromuscular symptoms such as cataracts, glucose intolerance, defective cardiac conduction and contraction, poor gall bladder function, and abnormal oesophageal and gastrointestinal motility, and the diagnosis of myotonic dystrophy is only entertained much later. In addition, some obligate heterozygotes for the myotonic dystrophy gene will have no detectable signs of myotonia or weakness. We have found that, independent of their degree of neuromuscular handicap, myotonic dystrophy patients show somatotomology of chronic depression. Over 50% of our myotonic dystrophy patients showed clinical elevation of the MMPI Depression (D) scale, and the averaged MMPI profile of sixteen of our patients (fig) showed elevations of the Depression (D) and Schizophrenia (S) scales (the "2-8 profile" code). Bird et al. found these same scales elevated in their patients. The cardinal features of patients showing the 2-8 MMPI code are pervasive unhappiness, depression, apprehension, introversion, unrealistic negative self-valuations, and blunted expressions of affect; thought processes are disturbed, but schizophrenic breaks seldom occur. We used our previously reported MMPI profiles of rheumatoid arthritis, multiple sclerosis, and neuromuscular disease patients as control groups with which to compare the profiles of our myotonic dystrophy patients. Although the physical handicaps of the control patients were equal to or more severe than that of the myotonic dystrophy patients, the control MMPI profiles showed significantly lower levels of depression. However, both the myotonic dystrophy patients and the patients in the control groups had elevations of the Hypochondriasis (Hs) and Hysteria (Hy) scales indicating physical concerns; Bird et al. also noted elevation of the Hs scale.

Our myotonic dystrophy patients also fulfilled the DSM-III criteria for major depressive disorder (occasionally, this diagnosis was made retrospectively when, after treatment, the patient admitted to chronic symptoms which he or she had considered "normal" until the symptoms disappeared with treatment), and had high scores on the Hamilton Rating Scale for Depression. With tricyclic antidepressant treatment of our myotonic dystrophy patients, we were able to demonstrate a marked reduction in the Hamilton Rating Scale scores and remission of the DSM-III depressive symptoms.

Bird et al. found that their patients scored lowest on the Digit Span, Object Assembly, and Block Design subtests of the Wechsler Adult Intelligence Scale (WAIS). Our patients had low scores on the Digit Span, Arithmetic, and Digit Symbol subtests, while in another recent report Woodward et al. found that their myotonic dystrophy patients had low scores on all five of these WAIS subtests. As a part of our ongoing study of cognitive changes associated with affective illness, we have shown a relationship between depression and impaired right hemisphere functioning, manifested by performance deficits on the Wechsler Block Design, Object Assembly, and Coding (Digit Symbol) subtests. We believe that chronic depression explains many of the "personality" problems attributed to patients with myotonic dystrophy. We also suggest that the presence of this chronic depression is relatively independent of the neuromuscular handicap. If, as we have hypothesised previously, myotonic dystrophy is a disease of abnormal membrane amnergic and peptidergic receptors, the depression in myotonic dystrophy may be the result of abnormal post-synaptic central nervous system receptors for these biogenic amines; this would make the depression in myotonic dystrophy pathophysiologically different from the more common major depressive disorder which is presumably the result of pre-synaptic reduction in formation and release of biogenic amines.

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References

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We agree that depression can be an important component of the clinical symptomatology in myotonic dystrophy. As indicated in table 6 of our paper depression was the major personality characteristic of two of our subjects, and a third patient also developed significant clinical depression following completion of our study. Seven of our 25 subjects (28%) tested with the MMPI had a T-score greater than 70 on the Depression scale. Six of these seven subjects had a physical disability score of Grade III or V. This small sample suggests some positive correlation between depression and degree of physical disability. The one subject with an elevated MMPI Depression scale and a physical disability Grade of II was mildly retarded (WAIS Full Scale score of 75), had obvious facial myopathy and a prominent personality disorder. The mean WAIS Full Scale score of the seven subjects with elevated MMPI Depression scales was 90, with a range of 64 to 106.

Although we agree that depression can be a very significant finding in some individuals with myotonic dystrophy, we suggest that ascertainment bias explains the smaller number of depressed subjects in our study compared with the patients of Brumback and Wilson. We designed our study to examine the full range of subjects with the myotonic dystrophy gene and, therefore, actively sought out other affected family members who had never sought medical attention and were often relatively asymptomatic. Forty-one per cent of our subjects had little or no physical handicap. Brumback and Wilson seemed to have primarily studied symptomatic patients who had already come to medical attention, a population likely to be skewed toward greater physical and mental handicap.

It is most difficult to find an appropriate control population to use in comparison with psychological study of myotonic dystrophy patients. Ideally the control group should have an autosomal dominant disorder with great variability in expression that sometimes, but not always, includes mental retardation, progressive physical handicap and abnormal facial appearance. Rheumatoid arthritis, multiple sclerosis, Duchenne muscular dystrophy and hereditary neuropathy all lack many of these characteristics. We have no simple solution to this methodological problem. However, neurofibromatosis meets many of the characteristics of the ideal comparison group, and we suspect that depression is relatively common in that disorder.

Neglected conditions producing preauricular and referred pain

Sir: We would like to comment on the recent paper "Neglected conditions producing preauricular and referred pain." (Friedman MH, Agus B, Weissberg J. J Neurol Neurosurg Psychiatry 1983;46:1067-72.) There is little evidence to support their conclusion that patients with atypical facial pain require specialised dental treatment. The incidence of malocclusion has not been shown to be any higher in patients with such facial pain in the general population, and attempts made to relieve symptoms with various forms of mechanical treatment, including occlusal therapy have not confirmed that their efficacy is greater than placebo. The American Dental Association has recently convened a conference on the management of such pain and concluded that only conservative reversible forms of therapy, including drug treatment such as analgesics or antidepressants could be recommended. The role of emotional stress has been increasingly recognised in these patients, and recently all varieties of facial pain have been shown to be associated with high incidence of previous adverse life events.

Facial pain is not exclusive to these patients. Eighty percent of them also complain of other recurrent symptoms, such as tension, headache, neckache, migraine, chronic low back pain, pelvic and spastic colon, suggesting the existence of a "pain person" with a vulnerable neurochemistry. A recent comparison of the demographic and social characteristics and previous history of various facial pain syndromes such as atypical facial pain and facial arthropalmygia (temporomandibular joint dysfunction syndrome) failed to differentiate the disorders. In addition, it was found that the facial symptom complexes recurred sequentially or simultaneously in the same patients along with other somatic disturbances as in response to stress. The superiority of the tricyclic antidepressant dothiepin (Prothiaden) compared to placebo in the relief of various facial pain disorders was also demonstrated in pain relief and found to be independent of any antidepressant effect of the medication.

Pain recurrence was associated with withdrawal from medication and in some cases the tricyclic antidepressants had to be maintained for at least a year to prevent relapse. In other words, facial pain should probably be considered to be a chronic disturbance, such as migraine or trigeminal...

Bird and Follett reply:

Brumback and Wilson emphasise the importance of depression in myotonic dystrophy. They found that more than 50% of their myotonic dystrophy patients had elevations of the MMPI Depression scale, that these patients were more depressed than subjects with rheumatoid arthritis, multiple sclerosis, or other neuromuscular diseases, that they responded to tricyclic antidepressant treatment, that their depression was relatively independent of their neuromuscular handicap and that the depression in myotonic dystrophy may be the result of abnormal post-synaptic central nervous system receptors for biogenic amines.


We agree that depression can be an important component of the clinical symptomatology in myotonic dystrophy. As indicated in table 6 of our paper depression was the major personality characteristic of two of our subjects, and a third patient also developed significant clinical depression following completion of our study. Seven of our 25 subjects (28%) tested with the MMPI had a T-score greater than 70 on the Depression scale. Six of these seven subjects had a physical disability score of Grade III or V. This small sample suggests some positive correlation between depression and degree of physical disability. The one subject with an elevated MMPI Depression scale and a physical disability Grade of II was mildly retarded (WAIS Full Scale score of 75), had obvious facial myopathy and a prominent personality disorder. The mean WAIS Full Scale score of the seven subjects with elevated MMPI Depression scales was 90, with a range of 64 to 106.

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