Paroxysmal myoclonic dystonia with vocalisations

Sir: Feinberg et al.1 described four patients whom they distinguished from the large majority of patients with Gilles de la Tourette syndrome on the basis of four “atypical” characteristics: (1) paroxysmal bursts of regular, repetitive, rhythmic, stereotypic, coordinated, simultaneous and bilateral myoclonus and vocalisations often with tonic symptoms; (2) presence of hyperactivity and attentional and learning disorders; (3) interference of symptoms with voluntary functioning; and (4) ineffectiveness of haloperidol. I have examined Case 1 and have considered him not to have the lightning-like jerks of myoclonus. Rather, he has virtually all the features of Gilles de la Tourette syndrome. I presented videotapes of his paroxysmal bursts of abnormal movements at the Unusual Movement Disorder Seminar held 29 May 1986, at the meeting of the American Academy of Neurology. The audience of 160 neurologists was in complete agreement that this young man suffered from the tic syndrome, commonly known as Gilles de la Tourette syndrome.

Let me address the four features of the disorder that the authors considered atypical. The first are the “paroxysmal bursts of regular, repetitive, rhythmic, stereotypic, coordinated, simultaneous and bilateral abnormal myoclonus (sic) and vocalisations often with tonic symptoms.” Paroxysmal bursts of stereotypic and coordinated movements are the hallmark of tics,2 and, in fact, are not encountered in other movement disorders. Paroxysmal dyskinesias of dystonia and chorea are well recognised,3 and paroxysmal tremor has been reported,4 but none of these are coordinated sequences of complex movements that are so typical of tics. Vocalisations, also, are a classical feature of the Gilles de la Tourette syndrome, and are only encountered elsewhere in Meige syndrome and as a feature of akathisia. The vocalisations of Case 1 included coprolalia, which is almost diagnostic for Gilles de la Tourette syndrome. Bursts of repetitive, rhythmic, bilateral movements are not commonly seen as part of the motor tic spectrum, but I see no reason why this phenomenology cannot be included within the realm of motor tics. Indeed, a minority of patients seen by me with otherwise typical features of tics have this feature. Tonic symptoms have long been recognised as a feature of tics,5 6 and today are commonly referred to as dystonic tics.

It is not clear why Feinberg et al listed “presence of hyperactivity and attentional and learning disorders” as atypical for patients with tics. Several investigators report that attention deficit disorder occurs in approximately 50% of patients with Gilles de la Tourette syndrome.7-9

Interference of voluntary functioning by symptoms does occur in Case 1. When he has a burst of the repetitive, rhythmic flexion movements of his arms, he stops speaking, other than occasional vocalisations. There is no loss of contact with the environment; rather, it appears as if his mind is actively and compulsively engaged in other activity, which is what he and other patients with tics who have this symptom inform me. Like bursts of repetitive movements, these simultaneously mind-occupying states should be considered within the spectrum of tics.

The final point raised by Feinberg et al is the ineffectiveness of haloperidol to suppress these paroxysmal motor bursts. But responsibility to medication is not an acceptable criterion for the diagnosis of tics! As an aside, and for a point of information, based on a telephone conversation I had with the patient on 8 May 1986, he considers himself 80% improved on fluphenazine and clonidine. Thus, the former drug, which blocks dopamine, similar to haloperidol, has benefited the patient.

I would like to propose that authors who wish to describe new or variants of movement disorders should also submit a videotape showing the abnormal movements. The demonstration of the videotape can most easily be accomplished in the newly founded journal Movement Disorders which includes a video format in addition to the classical written format. Those interested in this new journal can contact its publisher, Raven Press. I have obtained written permission from Case 1 to publish his videotape, and I will append it as part of the review of tics by Jankovic and Fahn.6

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References

Feinberg et al reply

We are pleased to respond to Dr Fahn’s comments on our paper. The fact that Case No 1 was presented at the Academy of Neurology meetings highlights the atypical nature of the symptoms. We also have presented Case No 1, in addition to the three other patients described in our report, including videotapes, to neurologists interested in movement disorders at the University of Michigan Medical School, Mount Sinai School of Medicine and the Basal Ganglia Club in New York City. None of over 150 physicians at any of these meetings opined that the symptoms represented Gilles
de la Tourette syndrome, except Dr Fahn and his colleague, Dr Burke.

Our paper addressed the important and generally acknowledged problem of potential heterogeneity in tic and Gilles de la Tourette disorders, and proposed that these patients might represent an atypical form of Gilles de la Tourette syndrome. How one views these cases will in large part be determined by whether one is a "lumper" and groups these patients with Gilles de la Tourette syndrome (or some other entity) or a "splitter" and separates them out from more typical cases. When one is confronted with a group of 1,600 patients that we have studied over the past 21 years, the majority categorised as typical Gilles de la Tourette syndrome, and when, from among that group, a small subgroup demonstrates an unusual, relatively consistent symptom pattern, one is led to hypothesise that these patients may represent a distinct entity. It is true that tics may occur in paroxysmal bursts and may have dystonic or tonic features. However, typical tics in Gilles de la Tourette syndrome are brief, irregular, nonrepetitive, non-rhythmic, usually unilateral, and, if they occur in paroxysms, they are usually of brief duration. Our patients' movements were regular, repetitive, rhythmic, and bilateral, often with coordinated vocalisations, and occurred upon occasion in prolonged bursts lasting as long as thirty minutes. In addition, these movements interfered with ongoing activity, a feature highly uncharacteristic of Gilles de la Tourette syndrome. There is no reason why these symptoms could not be included within the realm of motor tics, but we were struck with the fact that a particular subgroup demonstrated this cluster of unusual symptoms.

We also noted that all four cases (and two others not reported in detail), had hyperactivity, attentional and learning disabilities (ADD). It is also true that patients with Gilles de la Tourette syndrome may have hyperactivity and attentional and learning disorders. However, it is likely that the diagnosis of ADD in Gilles de la Tourette syndrome patients had been overestimated in the past because of ascertainment bias (greater likelihood of either diagnosis in patients with two illnesses). In fact, only 17% of 340 Gilles de la Tourette patients studied by us from 1981 to 1984 have had a concomitant diagnosis of ADD, a percentage which is close to the estimated population prevalence.

Whether the report of response to fluphenazine and clonidine in Case No 1 represents spontaneous improvement or drug response is difficult to ascertain, especially by telephone. Cases 2, 3, 4, and two others in our series, also demonstrated significant refractoriness to pharmacotherapy. Although poor response to medication is an inadequate diagnostic criterion, it may clinically point to the possibility that we are dealing with a particular subtype of Gilles de la Tourette syndrome, or, an as yet uncharacterised novel form of movement disorder.

In the final analysis, it is not critical whether we view this symptom complex as a new entity, a subentity, or typical Gilles de la Tourette syndrome. Our analysis is proposed as a heuristic hypothesis warranting further clinical observation and study. We hope our paper and Dr Fahn's comments, and the future availability of the videotape, will stimulate further study of atypical tic disorders. This hopefully will lead to greater diagnostic acumen, and definition of subtypes which might predict treatment response. If our paper stimulates work in this direction, we feel it will have served its purpose.

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**Book reviews**


Metal ions appear to exert fundamental actions on all parts of the nervous system. A deficiency or an excess of many elements may distort normal neuronal function. The present volume examines in detail the effects of metal ions relevant to both neurology and psychiatry.

A series of 22 chapters divided into five sections outlines in detail the advantages and disadvantages of metal ions on the nervous system. Some topics are predictable such as the actions of lithium, the possible involvement of aluminium in Alzheimer's disease, the role of copper in Wilson disease and lead toxicity. However, each of these is covered efficiently and in a manner summarising the current state of the art. In some chapters stimulating global hypotheses for the actions of metals are put forward. In particular, I would recommend the chapter by J Cramer which attempts to integrate the facets of the actions of lithium. Similarly, the chapter by JCL Lai and colleagues clearly portrays the essential role of a variety of metal ions in the normal functioning of neurotransmitter systems and the aberrations that alterations in concentration may cause.

Earlier chapters cover the importance of metal ions on the development of the nervous system and on the aging process. In particular, the involvement of metals in the rapid aging of patients with Down's syndrome is discussed in detail. Other interesting contributions include the role of vanadium as a cause of affective disorders and the potential of rubidium for treating psychiatric disorders. The complexity of the action of metal ions is illustrated in an excellent chapter on manganese by J Donaldson and the late Andre Barbeau. Clearly, this ion can induce a complex neurological syndrome when present in excess but at the same time is essential for the regulation of enzymes protecting the nervous system against attack by free radical mechanisms.

Finally, there is a collection of chapters on essential reading dealing with the critical issue of measurement of metal content in nervous tissue. The problems and pitfalls of obtaining accurate determinations are clearly stated. Also, there is a detailed description of the sophisticated techniques allowing localisation of metal ions within neuronal structures.

I found this volume well balanced and stimulating. It is a book of interest to many in the field of neurology, neuropathology, neurotoxicity, psychiatry and the basic sciences and provides a modern treatise on the role of metal ions that will be widely read and cited.


This is one of the best of the current glut of small books devoted to migraine and headache. The author is a senior registrar in Leeds who has had extensive experience in