THE IDIOPATHIC DYSTONIAS

A NOTE ON THEIR ORTHOPAEDIC PRESENTATION

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Eight patients suffering from various forms of idiopathic dystonia are described whose initial referral was for an orthopaedic opinion. The diagnoses of these patients, who were seen over a two-year period, comprised dystonia musculorum deformans, dystonia of the foot, spasmodic torticollis and occupational cramps. Although various musculoskeletal sequelae often occur, the primary underlying neurological cause of these unusual conditions is emphasised.

The dystonias are a group of motor disorders characterised by abnormal involuntary movements and postures produced by muscle spasms. Although attributable to disturbance of the extrapyramidal system, the underlying mechanisms remain unknown and no structural abnormalities have, as yet, been demonstrated (Marsden and Harrison 1974). These disorders may sometimes be symptomatic, for instance occurring in association with cerebral anoxia, birth trauma, Wilson's disease, encephalitis and especially drugs such as phenothiazines and butyrophenones. However, the disorders may often be idiopathic. They may either occur as a more generalised and sometimes inherited condition, such as dystonia musculorum deformans, or as a focal condition, such as spasmodic torticollis, and writer's and other occupational cramps.

The disturbance of movement and impaired function resulting from these involuntary movements result in disability that ranges from the catastrophic to a mild nuisance. However, there is neither weakness nor wasting and, since the condition is confined to the motor system, sensory, sphincter and reflex alterations do not occur, although the involuntary movements may cause sometimes major secondary effects on bone, joint and muscle. It is not surprising, therefore, that the patient with an idiopathic dystonia may be referred to the orthopaedic surgeon with what appears to be a musculoskeletal condition.

The purpose of the present report is to draw attention to the comparative frequency of such orthopaedic presentations.

CASE REPORTS

Over two years, eight patients with idiopathic dystonic disorders were referred to the neurological clinic of an orthopaedic hospital. By

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comparison, over the same period 10 patients were referred with Parkinson's disease, one patient with benign essential tremor and three patients with symptomatic dystonia—two with athetoid cerebral palsy and one with phenothiazine-induced dystonia and spasmodic torticollis.

Table I. Summary of clinical diagnoses

Case	Sex	Age (years)	Condition
1	F	30	Dystonia musculorum deformans
2	M	46	Dystonia musculorum deformans (predominantly axial)
3	F	19	Foot dystonia
4	F	41	Spasmodic torticollis
5	M	29	Harpischordist's cramp
6	M	60	Violinist's cramp
7	M	51	Cellist's cramp
8	M	31	Writer's cramp

The details of the eight patients are summarised in Table I. The investigations they underwent included electromyography, serum copper and caeruloplasmin estimation, radiography of the appropriate area of the spine and, where relevant, an electroencephalogram, examination of the cerebrospinal fluid, computerised axial tomography of the head and ophthalmological assessment to exclude Kayser-Fleischer rings; these investigations were normal in all patients except where indicated in the case reports below.

Case 1. A woman, aged 30 years, had had left-sided sciatica since the age of 13 years. Initially this had resolved with conservative treatment. A severe recurrence at the age of 24 years led to removal of a sequestered lumbosacral disc through an interlaminary fenestration; good symptomatic relief resulted. She then began to lean to the right when walking and at one time walked "almost bent double". She received physiotherapy without benefit and a year later right-sided sciatica developed, for which decompression of the right fifth lumbar root was undertaken; again, good symptomatic relief resulted. However, she continued to have impairment of her gait with aching in the right buttock and difficulty in walking and standing because of a tendency to tilt to the right which limited walking to 200 yards.

On direct questioning, it emerged that she had developed an intermittent facial asymmetry for four years with the right side at times moving less well; at the age of 13 years she had had difficulty in controlling her right hand, particularly when writing, and had changed

to left-handed writing. There had also been an impression of slightly impaired function of the left hand for a few years. Her family history was unremarkable, except that her father had had a disturbance affecting the right hand for 30 years (see below).

Examination revealed that she was now predominantly left handed. There was a fluctuating right facial asymmetry that appeared on talking and smiling. In the limbs, there was no weakness but fine finger movements, particularly of the right hand, were remarkably impaired, with the fingers curling into the palm and the wrist involuntarily twisting into a dorsiflexed posture. Occasional lateral dystonic movements of one or other foot occurred, and her gait was accompanied by a bizarre twisting tilt that fluctuated and was often accompanied by variable dystonic posturing of the arms.

There was no improvement with diazepam or tetrabenazine.

Her father, aged 70 years, was subsequently seen. He had had involuntary movements of large amplitude affecting the right hand for at least 30 years, the movements only occurring with activity and consisting of tremulousness, particularly when writing and with other dexterous movements such as carrying cups of tea. He had written with his left hand for many years, and in recent years a similar but milder tremor had affected that hand as well. Examination revealed a coarse irregular tremor of the whole of the right arm including the shoulder; this was of such a degree as to shake the body, but only occurred on maintained posture and when manual activities were carried out, particularly with the right hand.

A diagnosis of dystonia musculorum deformans with an autosomal dominant mode of inheritance was made in both patients.

Case 2. A man, aged 46 years, had had persistent, mainly posterior pain in the neck for two years. This tended to ease when he was lying down and relaxed. At the onset there had been some rapid involuntary side to side shaking movements of the head but these subsided after some months. Over the two years, he had noticed a progressive flexion deformity of the upper back and neck and had used a stick when walking. The spinal deformity caused difficulty in climbing stairs but strength in the legs remained unimpaired. He had had to give up work as a hatter over the same period. He had had rheumatic fever at the age of 27 years but his previous health had otherwise been good. There was no history of exposure to toxins such as mercury. A diagnosis of degenerative spinal disease had been considered and he had been treated with a collar and bed rest. His family history was unremarkable.

Examination revealed impassive facies, occasional involuntary snarling movements of the mouth of which the patient was unaware, slight titubation and strikingly increased tone in the arms. The trunk appeared markedly stiff. He stood and walked with a pronounced stoop but on request was able to stand perfectly upright for a brief period.

A diagnosis was made of some form of generalised dystonia with prominent truncal involvement and extrapyramidal features.

Case 3. This educationally-subnormal woman, aged 19 years, was born six weeks premature. She walked at 10 months and until the age of 18 years had experienced no motor problems, and was able to walk unlimited distances and danced for pleasure. One year previously she had fallen, twisting her right ankle which then became swollen. The swelling subsided but since this accident her foot had remained firmly plantar flexed. Physiotherapy, analgesics and a period of immobilisation in plaster with the foot forced into the neutral position did not improve her symptoms. She had been thought hysterical.

Examination was limited by poor co-operation. The only abnormality in the nervous system was of forceful plantar flexion and slight inversion of the right foot with apparent overactivity of the posterior tibial compartment muscles; the toes, however, were normally positioned and capable of dorsiflexion. The abnormal posture of the foot persisted during sleep. Her gait was curious, being ungainly and stiff and with the right foot plantar flexed. The reflexes were normal as was sensation, and there was no pain.

A diagnosis of a focal dystonia affecting one foot was made.

Case 4. A woman, aged 41 years, had noticed for two years that her head involuntarily tended to turn to the right; there was initially just a pulling sensation but gradually this was followed by actual movement

of the head. This occurred on many occasions during the day—usually for periods of some seconds. Sometimes the movements were forceful, especially when writing, and she complained that she “kept on looking over her right shoulder”. She had had to abandon driving her car and had also given up using lipstick which she could not apply accurately. Seven years previously she had sustained a whiplash injury of the neck in a road traffic accident, and she had had intermittent pain and stiffness of the neck since.

Examination proved normal except for intermittent brief lateral movements of the head to the right, which were sometimes forceful and were associated with painless overactivity of the left sternomastoid muscle.

She undoubtedly suffered from spasmodic torticollis. Treatment with physiotherapy, biofeedback, diazepam, Sinemet, tetrabenazine and pimozide has been ineffective.

Case 5. A male right-handed harpsichordist, aged 29 years, noticed two years previously that, on resuming playing after an interval of four months, the little and ring fingers of his right hand began to tire easily. He also noticed that his grip tended to “collapse”. Gradually this became more pronounced and the fingers involuntarily curled into the palm of his hand; he found that if he tried to extend these fingers he developed a sense of strain in the forearm muscles. These difficulties only occurred when he played either the organ or harpsichord and not when writing; he had had to abandon playing musical instruments entirely for the past year. A diagnosis of possible ulnar neuritis had been considered, also carpal tunnel syndrome and cervical spondylosis; he had been treated both by manipulation of the neck and by ultrasound. He had had a long history of depression and been prescribed phenothiazines and antidepressants, as well as electroconvulsive therapy. For 10 years he had been on trimipramine and tranlycypromine.

On examination this highly intelligent man was found to be normal except when mimicking or actually playing a piano when his two ulnar fingers involuntarily adopted a flexed posture. Exhaustive investigations revealed an isolated low serum copper level (8.2 micromoles per litre) and a slightly reduced serum caeruloplasmin level (1.2 micromoles per litre); his normal brother was found to have similar abnormalities of copper and caeruloplasmin levels. Detailed metabolic studies showed normal handling of copper and an ophthalmological assessment excluded the presence of Kayser-Fleischer rings on slit-lamp examination, eliminating the diagnosis of Wilson's disease.

It is probable that the low serum copper and caeruloplasmin levels were an unrelated finding as they were abnormal in the unaffected brother. This indicated that the history of drug exposure was fortuitous. A diagnosis of a musician's cramp seemed likely.

Case 6. A male professional violinist, aged 60 years, had noted an aching around his right shoulder for seven months. He subsequently noticed when playing his violin that his right little finger could not be placed properly on the bow, and he then developed difficulty in discrete and fine finger movements of that hand but only when playing the violin. Later there developed a slight tremor of the index finger and thumb, and a tendency for the index finger to “wrap itself around the bow” and the thumb appeared incapable of controlling the weight of the bow. All other movements including writing, doing up buttons and manipulating cutlery were normal. He had a long-standing history of cervical spondylosis and 12 years previously had had diazepam and later chlorthalidopoxide for a short-lived anxiety state.

Examination revealed slightly reduced movements of the right side of the face, dystonic posturing of the right thumb and a low-amplitude rest tremor of that thumb with impaired swing of the right arm while walking. Radiographs of the cervical spine revealed minor degenerative changes.

A diagnosis was made of a musician's cramp with mild Parkinson's disease and cervical spondylosis.

Case 7. A male professional cellist, aged 51 years, had for three years experienced episodes in which the middle and ring fingers of his left hand suddenly and involuntarily extended when he gripped the bow of his cello; a diagnosis of some form of peripheral nerve disorder had

been considered. These involuntary movements would last one or two minutes and then subside spontaneously. Subsequently he had similar brief episodes on other occasions—playing the piano, putting on his coat and when driving. Only during the most recent occasion had other fingers been involved; all four fingers had suddenly extended.

Examination revealed no abnormality. A diagnosis of a musician's cramp was made.

Case 8. A male right-handed teacher, aged 31 years, found that from the age of 18, whenever he started to write, his hand would pronate and a sense of strain would develop in his forearm muscles. At times the right arm tended to pronate spontaneously. The symptoms gradually became more obtrusive and impaired certain other activities, such as manipulating nuts and bolts and playing the piano. He also tended to clench excessively objects he was using, such as a screwdriver. Other symptoms had been of mild and occasional neckache with radiation down the lateral aspect of the right arm. At the age of 22 years he had had an exploratory operation on the right supraclavicular fossa with negative findings.

Examination revealed slightly impaired fine finger movements of the right hand, variable impairment of pinprick appreciation over the lateral aspect of the right forearm and a diminished right supinator reflex. His fingers adopted a peculiar tight posture when he wrote and his arm pronated.

A diagnosis of writer's cramp with mild compression of the C6 root was made. The results of neurophysiological studies were normal, except that electromyographical studies on the forearm muscles showed only brief periods of relaxation during writing—consistent with a diagnosis of writer's cramp. Biofeedback therapy resulted in mild improvement.

DISCUSSION

These cases demonstrate that a number of patients seen by orthopaedic surgeons suffer from some form of dystonia. In contrast with other extrapyramidal disorders, such as Parkinson's disease, patients with dystonia may occasionally escape detection for many years, and noteworthy with some of the focal dystonias is the strict relationship to some narrowly defined activity. While it is probable that delay in diagnosis of a dystonic condition will not adversely affect the patient's management, it is clearly beneficial that the cause of the symptoms is recognised.

The obscure nature of the dystonias has been referred to earlier, as has a classification into generalised and focal dystonia (Marsden and Harrison 1974). The generalised dystonias in which involuntary movements affect much of the body, even if asymmetrically and patchily, often occur in childhood, with the legs involved early with later spread to the arms and trunk. These generalised dystonias of early onset may occur as a hereditary disorder, most commonly as an autosomal dominant condition, but an autosomal recessive mode of inheritance, seen particularly among Ashkenazi Jews, is well recognised. Case 1 is of particular interest in that the patient's father suffered from the marked tremulous condition that almost certainly represents a *forme fruste* of dystonia (Larsson and Sjögren 1966), and hence an autosomal dominant mode of inheritance seems likely. Atypical clinical features such as tremor imply that caution should be exercised before a patient's dystonia can be considered sporadic, which in fact many cases with onset in adulthood appear to be.

Cases 3 to 8 are examples of focal dystonic conditions. The curious relationship of the involuntary movements to a specific manual task seen in Cases 5 to 8 cannot satisfactorily be explained at present. The commonest activity to be affected is writing, but typists (and in former times writers of copperplate and Morse-key operators) may have similar problems; apart from the musicians' cramps described above, trumpeters, drummers and harpists may also suffer (Critchley 1977). These cramps may sometimes be a fragment of a more widespread dystonia as Cases 7 and 8 illustrate; not only are other manual activities affected but there may also be spread to involve nearby muscles. This latter phenomenon has given rise to the term "segmental" dystonia; such cases usually occur after childhood and there is often no further development to the more serious generalised torsion dystonia. Focal involvement of the neck muscles gives rise to spasmodic torticollis as seen in Case 4, and Case 3 is probably an example of the rare but well-recognised dystonic involvement of the foot (Messina *et al.* 1979).

It is of interest that neck symptoms and minor features of cervical spondylosis were reported in three of the eight patients. Although the role of trauma as an initiating factor in these and other involuntary movements is unclear (Schott 1981), previous trauma does seem to occur with relative frequency at least in patients with spasmodic torticollis (Sheehy and Marsden 1980), and the injury preceding the development of the dystonic foot disorder in Case 3 may be relevant. The history of prematurity and mental subnormality in Case 3 is also noteworthy; while these features might preclude categorisation as an idiopathic dystonia, the lack of other previous or progressive neurological abnormality may indicate that the mental subnormality is an unrelated feature, although the issue remains controversial (see Burke, Fahn and Gold 1980).

Just as scoliosis may occur in association with Parkinson's disease (Duvoisin and Marsden 1975), so the involuntary movements in dystonia may cause considerable musculoskeletal problems, and it is these problems which often result in referral for an orthopaedic opinion. Spasmodic torticollis in particular gives rise to degenerative changes in the cervical spine, and pain, sometimes severe, is a frequent accompaniment due not only to spasm of the affected muscles but also to the bony changes and nerve entrapment that may result. Treatment of these sequelae is unsatisfactory, and conservative measures such as physiotherapy and use of a collar in spasmodic torticollis usually fail in the face of the relentless involuntary movements. In the focal dystonias affecting the arm, it is not uncommon for the patient to undergo decompression of the median or ulnar nerves, invariably with failure; peripheral nerve surgery is as illogical as it is valueless.

By definition, underlying primary conditions will have been excluded before a diagnosis of idiopathic

dystonia is made. Nevertheless, the fact that certain diseases and drugs can provoke similar conditions makes unsatisfactory the common view that such disorders have a psychological basis (see Matthews *et al.* 1978), as do the abnormal electromyographical findings seen in writer's cramp (von Reis 1954) and other neurophysiological observations seen in generalised dystonia (Yanagisawa and Goto 1971). That specific factors can induce dystonia may eventually provide a clue to the mechanisms involved. They have already to some extent indicated a possible rational approach to therapy.

There is no doubt that the treatment of these involuntary movements, whether focal or generalised, is extremely unsatisfactory (Marsden 1981). At least 27 drugs have been tried ranging from diazepam to marihuana. As L-dopa and other dopaminergic agonists may produce dystonic movements, it appeared rational to use dopamine antagonists to alleviate the involuntary movements but this and other apparently logical approaches have generally been fruitless. Occasional patients unpredictably seem to respond to one or other drug, but the author's preference is to use in turn three or four relatively non-toxic drugs—a benzodiazepine such as diazepam, an anticholinergic drug such as benzhexol, and an anticonvulsant such as carbamazepine—in the hope that one will prove useful and of sufficiently low toxicity that long-term use would be justified. Other therapeutic techniques tried with limited benefit have

included biofeedback and psychotherapy, but many other psychiatric techniques have been attempted with singular lack of success. Finally, surgery has a limited place in management but controversy persists concerning both the indications and the procedures. In severe uncontrollable spasmodic torticollis, rhizotomy of anterior or posterior roots or both from C1 to C4 may be undertaken, alone or in combination with denervation of the sternomastoid muscle, and with or without thalamotomy. In the severe generalised dystonias, thalamotomy and other stereotactic procedures may be considered. The use of drugs and surgical procedures that may carry major risks requires careful consideration in diseases which themselves carry a comparatively low mortality. Despite this low mortality, however, morbidity can be severe and the prognosis is poor; prolonged remission in idiopathic dystonia occurs in under 20 per cent of patients, and only one patient with spasmodic torticollis in a series of 30 recovered completely (Matthews *et al.* 1978).

Thus it can be seen that the dystonias, in which there is often a secondary disturbance of function of the musculoskeletal system, comprise sometimes disabling but at other times serious disorders which are fairly frequently referred to the orthopaedic surgeon. An appreciation of the true nature of the patient's condition will prevent inappropriate surgery, and may well lead to more satisfactory forms of treatment in the future.

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