Tourette syndrome; a childhood disorder

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The syndrome of multiple motor tics and involuntary vocalizations was delineated by Georges Gilles de la Tourette[1] in 1885 when he described nine cases. One of these cases had previously been described by Itard[2] in 1825. Only a few additional cases were published over the next 75 years, but an increasing number have been reported since the 1960s. Reasons for this increased reporting include (1) the discovery of an effective medical therapy; (2) the activities of a special interest group, the Tourette Syndrome Association; and (3) descriptions of this entity in the popular press which have led to self-referral of patients who have recognized the syndrome in themselves or in family members.[3, 4] Along with the increasing number of patients with Tourette syndrome is the recognition that this is a disorder which invariably begins in childhood.[5] This was true in Tourette's initial series and has been confirmed in all subsequent reports.

We are reporting 12 cases of Tourette syndrome. Eleven of these patients were examined in a 1-year period at the Cleveland Clinic; ten were children and two were young adults whose symptoms dated back to childhood. The correct diagnosis had been made previously in only two

cases, but three other patients sought medical attention because of self-diagnosis.

The 12 patients were between 9 and 24 years of age when first examined; 10 were between 8 and 15 years. Symptoms began in all patients before 10 years of age. The delay between onset of symptoms and correct diagnosis ranged from 2 to 17 years and averaged 6 years (*Table 1*). The types of motor tics are listed in *Table 2*. Vocal tics usually begin later than motor tics and examples are listed in *Table 3*. Coprolalia was part of the history in only four patients and had spontaneously stopped prior to the examination in one patient. Birth and medical history were unremarkable in all patients, and all had experienced normal early childhood development. Seven patients had behavioral difficulties or learning problems and, in general, the patients who were the youngest when seen for evaluation were those with the greatest difficulties in this area. Three children were attending special education classes.

Eight patients had been treated un-

Table 1. Clinical findings

Patient	Sex	Age at onset (yr)	Age at diagnosis (yr)	Previously diagnosed	Coprolalia	Medical history	School situation; behavior	Family history
1	M	8	24	No	Yes	N1	College graduate	Brother with occasional motor tics
2	M	7	9	No	No	N1	Hyperactive, disruptive	Adopted
3	M	6	8	No	No	N1	Fidgety, restless	Maternal aunt had motor tics as child
4	F	8	10	Yes	No	N1	No problems	Father had motor tics for 7 years
5	M	5	15	No	No	N1	No problems	None
6	F	9	13	Yes	Yes	N1	Fearful	Brother and maternal cousins with eye blinking
7	M	4	12	No	No	N1	Hyperactive, disruptive	Paternal first cousin with motor and vocal tics (related to patient 10)
8	M	6	10	No	Yes	N1	Hyperactive, special class	Adopted
9	F	6	23	No	No	N1	Completed 1 yr college	None
10	M	4	9	No	No	N1	Hyperactive, special class	Father had motor and vocal tics as child; paternal cousin has motor and vocal tics (related to patient 7)
11	M	7	10	No	Yes	N1	Fidgety, restless	Half brother with facial tics
12	M	9	14	No	No	N1	No problems	Paternal cousin with motor and vocal tics.

Table 2. Types of motor tics

Eye blinking
Eye rolling
Nose twitching
Lip biting
Facial grimacing
Head shrugging
Head twisting
Shoulder shrugging
Arm extension
Arm twisting
Finger tapping
Body shuddering
Chest beating
In and out movements of chest and abdomen
Extension of legs
Foot stamping
Toe twitching
Hopping
Jumping
Smelling
Touching genitalia

Table 3. Vocal tics

Grunting
Humming
Swallowing
Snorting
Gutteral noise
Throat clearing
Coughing
Gurgling
Hooting
Formed nonobscene words
Echolalia
Coprolalia

successfully with various pyschotropic drugs including phenobarbital, methylphenidate, dextroamphetamine, pemoline, chlorpromazine, thioridazine, imipramine, diazepam, and hydroxyzine.

Haloperidol was used to treat ten patients. Of these, eight received 3 mg or less a day; one received 4 mg/day; and one, 8 mg/day. Vocal tics were controlled in all patients except one whose dosage is still being increased. Control of motor tics was improved by more than 50% in all patients, although the motor tics persisted to some degree in all patients. Stress and anxiety typically increased the tics even when the patient was receiving medication.

Case report

The patient was 10 years old when first seen for neurologic evaluation. He was an adopted child who had enjoyed good health and normal early development. Hyperactivity had been a concern from the time he started school and he was transferred to a learning disability class. Motor tics began at 6 years of age. Methylphenidate was prescribed for his hyperactivity and led to a marked increase in motor tics as well as to the onset of vocal tics and coprolalia. All tics decreased when the methylphenidate therapy was temporarily discontinued and increased when the medication was resumed. When examined, he was no longer receiving methylphenidate and coprolalia was no longer present.

Examination was normal except for multiple motor and vocal tics. These included eye blinking, head throwing, shoulder shrugging, touching of genitalia, snorting, hiccuping, and grunting. Haloperidol was begun and maintained at 1 mg/day, since higher dosage led to an undesirable amount of sedation. On this dosage, all vocal tics disappeared and a 50% reduction in motor tics was achieved.

Discussion

In the absence of a biochemical or electrophysiologic marker, the diagnosis of Tourette syndrome remains a clinical one. Essential clinical features include involuntary motor and vocal tics. Motor tics are usually the first to appear and often begin as simple tics involving the face or head. Complex or stereotyped movements may also occur and include jumping,

hopping, squatting, smelling, or touching especially the genitalia. Vocal tics may consist of sounds or words. Sounds include coughing, hissing, barking, and throat-clearing. Stammering, stuttering, or echolalia may occur, or there may be repetitive utterings of partial or whole words. The most dramatic vocal tic is termed coprolalia, the involuntary use of obscene language. It must be emphasized, however, that the presence of coprolalia is not necessary for the diagnosis of Tourette syndrome. Coprolalia occurs in only approximately 60% of the cases and may not appear for many years after the onset of motor or other vocal tics.

As in other series, the onset of symptoms in our patients was between 2 and 15 years of age, and more boys were affected than girls. None of our patients were Jewish, although a relatively high incidence in Ashkenazi Jews has been reported by Shapiro et al.[6,7] It has been suggested that the high incidence in Ashkenazi Jews reflects a selection bias due to Shapiro's referral base.[8]

The background medical history was normal in all cases, and no problems were encountered in early development. Interestingly, five of our patients were hyperactive or restless and had been diagnosed as having minimal brain dysfunction (MBD). Whether MBD and Tourette syndrome coexist in the same patient or whether both are due to a common mechanism is unknown, but such an association has only recently been emphasized.[5,8] This area deserves further study, especially since medication frequently prescribed for MBD may exacerbate the symptoms of Tourette syndrome.

The genetic aspect of this disorder is being increasingly appreciated.[9] There have been reports on many families with more than one involved family member, although no consistent pattern of inheritance is yet apparent. In our series, a history of tics was reported in the families of eight of the ten patients whose family backgrounds were known (the remaining two patients had been adopted). The families of three patients included others with both vocal and motor tics. The remaining five patients were from families where other members had histories of only motor tics. This suggests that Tourette syndrome is not an isolated entity, but represents one point along a spectrum of disorders characterized by tics.[10] Two of our patients were paternal first cousins, and the father of one of them (the uncle of the other) had exhibited motor and vocal tics during childhood.

Neither the neurologic examination nor known laboratory tests have been helpful in establishing a diagnosis. An electroencephalogram was performed in eight patients and was normal in six. The remaining two had nonspecific changes such as poor voltage regulation and excessive posterior slowing. A CT brain scan was normal in four patients.

Even though the symptoms of Tourette syndrome are dramatic, a long delay occurred between the onset of symptoms and the time when the correct diagnosis was made. Most patients had been previously diagnosed as having an emotional or behavioral disorder, and many had received a variety of psychotropic drugs in an unsuccessful attempt to relieve the symptoms. Although the differential diagnosis includes Huntington's chorea, dystonia musculo-

rum deformans, Sydenham's chorea, Lesch-Nyhan syndrome, and Wilson's disease, the motor and vocal tics of Tourette syndrome are distinct and easily distinguished from these disorders.[11] The only entity which may be difficult to distinguish from Tourette syndrome is simple or multiple tics of childhood and adolescence. These motor tics are due to psychological adjustment reactions and are transiently present in many normal children during periods of anxiety or stress. They are not long-lasting and are rarely associated with vocal tics. The persistence and variability in motor tics along with significant vocal component differentiates Tourette syndrome from this benign entity.

The discovery that haloperidol, a known dopamine blocking agent, ameliorates the symptoms of this syndrome, has led to the current concept of an organic etiology for Tourette syndrome. In previous years, Tourette syndrome had been considered a psychiatric disorder. Shapiro et al[12] have refuted this and have shown that patients with this syndrome are not characterized by common psychopathologic factors. Another misconception which has recently been discarded is that such patients suffer from progressive mental and physical deterioration. This belief is unfounded, and patients with Tourette syndrome have normal intelligence and lead long and productive lives, a situation modified only by possible social stigma and the potential reactive emotional sequela which may then result.

A wide variety of treatment has been used in the past without major success. Included are minor and major tranquilizers, antidepressants, stimulant drugs, hypnotics, levodopa, thalamotomy, psychotherapy, and behavior therapy. Methylphenidate, a stimulant drug, has been found to precipitate or accentuate tics of Tourette syndrome in some patients.[13] This is presumably due to its pharmacologic action of increasing the amount of dopamine in the synaptic cleft. Four of our patients had previously received methylphenidate, and as a result symptoms increased in two of them.

The current treatment of choice is haloperidol. This drug has been helpful in 90% of cases and was successful in varying degrees in all of our patients who received this medication.[14] It is not known why other dopamine blocking agents such as the phenothiazines do not provide relief as successfully as does haloperidol. Up to 180 mg/day has been necessary in controlling symptoms in adults,[14] but most children respond to much lower dosages.[5] Eight of our nine treated children improved with less than 4 mg/day. The major side effect at lower dosages is sedation, and this is usually the factor which limits the ultimate daily dosage. All our patients who were receiving haloperidol complained of some degree of sedation, and it was necessary to reach a balance between the symptomatic relief of the tics and the sedative effects of the therapy. At higher dosages, extrapyramidal symptoms develop, and patients require additional treatment with antiparkinsonian drugs. Both patients receiving 4 mg or more per day of haloperidol required the addition of such a drug.

The natural history of Tourette syndrome consists of waxing and waning symptoms interspersed, at times, with symptom-free periods. It

is possible that some patients spontaneously become permanently symptom-free, although it has been argued that, by definition, Tourette syndrome is a lifelong disorder.[8] Because of this fluctuation, treatment with haloperidol must be closely monitored. Dose requirement will vary not only due to the intensity of the side effects, but also to the natural cyclical variation in the intensity of the symptoms.

Summary

Tourette syndrome is characterized by multiple motor and vocal tics with or without coprolalia. Although the disorder was once considered rare, increasing numbers of patients with Tourette syndrome are being recognized. Twelve cases seen at The Cleveland Clinic Foundation are summarized. Tourette syndrome always begins in childhood, and there is often a long delay before the correct diagnosis is made. Haloperidol is usually effective in ameliorating the symptoms of this dramatic disorder.

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