

Guide to the Diagnosis and Treatment of Tourette Syndrome

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Introduction

This publication is the third edition of A Physician's Guide to the Diagnosis and Treatment of Tourette Syndrome. It has been revised substantially to include up-to-date clinical information for physicians treating patients with this complex, frequently misunderstood neurobehavioral (neurobiological) disorder.

Definitions of Tic Disorders

Tics are involuntary, rapid, repetitive and stereotyped movements of individual muscle groups. They are more easily recognized than precisely defined. Tic disorders are generally categorized according to age of onset, duration of symptoms, severity of symptoms and the presence of vocal and/or motor tics.

Transient tic disorders often begin during the early school years and can occur in up to 18% of all children. Common tics include eye blinking, nose puckering, grimacing and squinting. Transient vocalizations are less common and include various throat sounds, humming or other noises. Childhood tics may be bizarre - palm licking, poking and/or pinching the genitals are examples. Transient tics last only a few weeks or months and are usually not associated with specific behavioral or school problems. They are especially noticeable during times of heightened excitement or fatigue. As with all tic syndromes, boys are three to four times more often affected than girls. While transient tics by definition do not persist for more than a year, it is not uncommon for a child to have recurrent episodes of transient tics over the course of several years.

Chronic tic disorders are differentiated from transient tic disorders not only by their duration over many years, but by their relatively unchanging character. While transient tics come and go (sniffing may be replaced by forehead frowning and the frowning may become finger snapping), chronic tics - such as facial contortions or blinking - may persist unchanged for years.

Chronic multiple tics suggest that an individual has several chronic motor tics (or, in rare cases, several chronic vocal tics). Often it is not an easy task to draw distinctions between transient tics, chronic tics, and chronic multiple tics.

Tourette Syndrome (TS), first described by Gilles de la Tourette, can be the most debilitating tic disorder and is characterized by multiform, frequently changing motor and phonic tics. The current diagnostic criteria, as defined by the Diagnostic and Statistical Manual of Mental Disorders IV are as follows:

1. Both multiple motor and one or more vocal tics have been present at some time during the illness, although not necessarily concurrently.
2. The tics occur many times a day (usually in bouts) nearly every day or intermittently throughout a period of more than 1 year, and during this period there was never a tic-free period of more than 3

consecutive months.

3. The disturbance causes marked distress or significant impairment in social, occupational, or other important areas of functioning.
4. The onset is before age 18.
5. The disturbance is not due to the direct physiological effects of a substance (e.g. stimulants) or a general medical condition (e.g. Huntington's disease or postviral encephalitis).

While the criteria appear basically valid, they are not absolute. First, there have been rare cases of TS which have emerged later than age 18. Second, the concept of "involuntary" may be hard to define operationally, since many patients experience their tics as having a volitional component - either a capitulation to an internal sensory urge for motor discharge, or a more generalized psychological tension and anxiety, or both. Finally, the diagnostic criteria do not adequately portray the full range of behavioral difficulties that are commonly observed in patients with TS, such as attentional problems, compulsions, and obsessions.

Differential Diagnosis

Today, the full-blown case of TS is unlikely to be confused with any other disorder. In the past, however, TS as frequently misdiagnosed or undiagnosed.

The differentiation of TS from other tic syndromes may be no more than semantic, especially since recent genetic evidence links TS with multiple and transient tics of childhood and can only be defined in retrospect.

At times it may be difficult to distinguish children with extreme attention deficit hyperactivity disorder (ADHD) from those with TS. On close examination, many ADHD children have a few phonic or motor tics, grimace, or produce noises similar to those with TS. Since at least half of patients with TS also have had attention deficits and hyperactivity as children, a physician may well be confused. However, the treating doctor should be aware of the potential complications of treating a possible case of TS with stimulant medication.

On rare occasions, the differentiation between TS and a seizure disorder may be difficult. The symptoms of TS sometimes occur in a rather sharply separated paroxysmal manner and may resemble automatisms. Patients with TS, however, retain a clear consciousness during such paroxysms. If the diagnosis is in doubt, an EEG may be useful.

We have seen TS in association with a number of developmental and other neurological disorders. It is possible that central nervous system injury from trauma or disease may cause a child to be vulnerable to the expression of the disorder, particularly if there is a genetic predisposition. Autistic and retarded children may display the entire gamut of TS symptoms. Whether an autistic or retarded individual requires the additional diagnosis of TS may remain an open question until testing (biological or otherwise) is available for definitive diagnosis of TS.

In older patients, conditions such as Wilson's disease, tardive dyskinesia, Meige's syndrome, chronic amphetamine abuse and the stereotypic movements of schizophrenia must be considered in the differential diagnosis. The distinction can usually be made by taking a good history or by blood tests.

Since more physicians are now aware of TS, there is a growing danger of over-diagnosis or over-treatment.

It is up to the clinician to consider the effect that the symptoms have on the patient's ability to function (as well as the severity of associated symptoms) before deciding to treat with medication or other approaches.

Symptomatology

The varied symptoms of TS can be divided into motor, vocal, and behavioral manifestations ([Table 1](#)). Simple motor tics are fast, darting, meaningless muscular events. They can be embarrassing or even painful (such as jaw snapping). They are easily distinguished from simple muscular twitches or rapid fasciculations, e.g., of the eyelid or lip. Complex motor tics may be slower or more purposeful in appearance and more easily described by terms used for deliberate actions ([Table 2](#)).

Complex motor tics can be virtually any type of movement that the body can produce including gyrating, hopping, clapping, tensing arm or neck muscles, touching people or things and obscene gesturing.

At some point in the continuum of complex motor tics, the term "compulsion" seems appropriate for capturing the organized, ritualistic character of the actions. The need to do and then redo or undo the same action a certain number of times (e.g., to stretch out an arm ten times before writing, to "even up," or to stand up and push a chair into "just the right position") is compulsive in quality and accompanied by considerable internal discomfort. Complex motor tics may greatly impair school work, e.g., when a child must stab at a workbook with a pencil or must go over the same letter so many times that the paper is worn thin. Self-destructive behaviors, such as head banging, eye poking and lip biting also may occur. The distinction between complex tics and compulsions may be a difficult one for the physician to make and some "complex tics" may be alleviated by medications used for obsessive-compulsive disorder.

Vocal tics extend over a similar spectrum of complexity and disruption as do motor tics ([Table 3](#)). With simple vocal tics, patients emit linguistically meaningless sounds or noises, such as hissing, coughing or barking. Complex vocal tics involve linguistically meaningful words, phrases or sentences, e.g., "wow," "Oh boy, now you've said it," "Yup, that's it," "but, but..." Vocal symptoms may interfere with the smooth flow of speech and resemble a stammer, stutter or other speech irregularity. Often, but not always, vocal symptoms occur at points of linguistic transition, such as at the beginning of a sentence where there may be speech blocking at the initiation of speech or at phrase transitions. Patients suddenly may alter speech volume, slur a phrase, emphasize a word or assume an accent.

The most socially distressing complex vocal symptom is coprolalia, the explosive utterance of foul or "dirty" words or more elaborate sexual, aggressive or insulting statements (e.g., racial slurs). Coprolalia is not simply obscene speech spoken in anger or to offend. Rather it is often sudden speech (typically just the first syllable of an inappropriate word) that interrupts an otherwise appropriate flow of words. While coprolalia occurs in only a minority of patients with TS (from 5-30%, depending on the clinical series), it remains the most well known TS symptom. A diagnosis of TS does not require that coprolalia be present and the majority of patients do not ever exhibit this symptom.

Some patients with TS may have a tendency to imitate what they have just seen (echopraxia), heard (echolalia), or said (palilalia). For example, the patient may feel an impulse to imitate another's body movements, to speak with an odd inflection or to accent a syllable in just the same manner as another person. Such modeling or repetition may lead to the onset of new specific symptoms that will wax and wane in the same way as other TS symptoms. Some patients also describe "triggers" that almost invariably prompt a tic, e.g., another person coughing in a certain way.

The symptoms of TS can be characterized as mild, moderate or severe by their frequency, their complexity

and the degree to which they cause impairment or disruption of the patient's ongoing activities and daily life. For example, extremely frequent tics that occur 20-30 times a minute, such as blinking, nodding or arm flexion, may be less disruptive than an infrequent tic that occurs several times an hour, such as loud barking, coprolalic utterance or touching tics. The premonitory sensory urges tend to be present by 9 to 10 years of age. They are most commonly reported in the shoulder girdle, hands, throat and abdomen.

There may be tremendous variability over short and long periods of time in symptomatology, frequency and severity. Tics typically occur in "bouts" with many tics over a short interval of time. Patients may be able to inhibit or not feel a great need to emit their symptoms while at school or work. When they arrive home, however, the tics may erupt with violence and remain at a distressing level throughout the remainder of the day.

It is not unusual for patients to "lose" their tics as they enter the doctor's office. Parents may plead with a child to "show the doctor what you do at home," only to be told that the youngster "just doesn't feel like doing them" or "can't do them" on command. Adults will say "I only wish you could see me outside your office," and family members will heartily agree.

Often a patient with minimal symptoms may display more severe tics when the examination is over. Thus, for example, the doctor may often see a nearly symptom-free patient who then leaves the office and begins to hop, flail, or bark as soon as he or she reaches the street.

In addition to the moment-to-moment or short-term changes in symptom intensity, many patients have oscillations in severity over the course of weeks and months. The waxing and waning of severity may be triggered by changes in the patient's life; for example, around holidays, children may develop exacerbations that take weeks to subside. Other patients report that their symptoms show seasonal fluctuation. However, there are no rigorous data on whether life events, stresses or seasons do, in fact, influence the onset or offset of a period of exacerbation. Once a patient enters a phase of waxing symptomatology, a process seems to be triggered that will run its course for weeks or months.

In its most severe forms, patients may have uncountable motor and vocal tics during all their waking hours with paroxysms of full body movements, shouting or self-mutilation. At times the tics seem organized in orchestrated patterns that are characteristic of that individual. Despite this, many patients with severe tics manage to achieve adequate social adjustment in adult life, although usually with considerable emotional pain. More than the severity of motor and vocal tics, the factors that appear to be of importance with regard to social adaptation include the seriousness of attentional problems, obsessive-compulsive symptoms, the degree of family acceptance and support, intelligence and ego strength.

In adolescence and early adulthood, patients with TS frequently come to feel that their social isolation, vocational or academic failure and embarrassing symptoms are more than they can bear. At times, a small number may consider and attempt suicide. Conversely, some patients with the most bizarre and disruptive symptomatology may achieve excellent social, academic and vocational adjustment. Fortunately, in many cases, tics diminish during the course of adolescence. However, in other cases (<10%), the tic symptoms can become even more severe in adulthood.

[Return to the TSA Home Page](#)