More than a century has passed since the French neurologist Georges Gilles de la Tourette first described the condition that bears his name, a name familiar to the lay public and health professionals alike. Once considered so rare that neurologists might expect to witness the disorder perhaps once in their professional lifetime, Tourette syndrome (TS) is, in fact, common enough that virtually all pediatricians will have several patients with this condition in their practice. Yet, despite its name recognition and the number of people it affects, TS often goes undiagnosed or misdiagnosed. Misconceptions about this tic disorder are customary, with the syndrome often perceived as characterized by bizarre, fitful behaviors or comical outbursts of uncontrollable profanity.

Colorful descriptions of motor and phonic tics in literary characters (such as “Mr. Pancks” in Charles Dickens’s *Little Dorrit* and “Captain Hardcastle” in Roald Dahl’s autobiographical works) and in historical figures (including the Roman Emperor Claudius, Wolfgang Amadeus Mozart, and 18th century English literary scholar Samuel Johnson) testify to the widespread awareness of TS across cultures and time, despite (or perhaps because of) its perceived rarity. So it seems that the medical community trailed the untrained community by centuries in recognizing the syndrome.

From the time of its initial medical description in 1885 until the 1960s, medical experts viewed TS as a psychological disorder, and treatment was customarily directed toward psychotherapy. In the 1960s, the observed benefit of psychotropic medications in alleviating tics reformed this perception, proving a clear neurologic basis to the phenomenon. That discovery has prompted a wide range of research-based interest in understanding and classifying this varied disorder.

The tics of TS range widely in their severity, form, frequency, and intensity. Moreover, although tics are the hallmark of diagnosis, TS is associated with a range of comorbid conditions. When present, these conditions are usually more serious or disabling than the tics themselves.

The pediatrician can be most helpful to a child with TS by recognizing signs and symptoms early in onset.
The diagnosis of TS (the focus of Part 1 of this two-part article) and its management (the focus of Part 2, which begins on page 38) depend on recognizing the unique clinical presentation of each child and directing an ongoing comprehensive and supportive care plan.

The tics of Tourette: There is no textbook presentation

Recent studies suggest that tics—mostly transient, lasting less than one year—occur in 20% or more of schoolchildren. Tourette syndrome, in which the tics are chronic, may occur in 1% or more of mainstream children, with a higher prevalence still among children with special education needs.

The clinical manifestations of TS are extremely diverse, and no two patients present identically. Most have mild tics; the minority is severely affected. Tics are repetitive, sudden, involuntary or semivoluntary productions. They are customarily classified as “simple” or “complex” and as “motor” (movement) or “phonic” (sound-producing). Tics may appear to be exaggerated fragments of ordinary motor or phonic behaviors that occur out of context.

TS is the most severe tic disorder, marked by both motor and phonic tics; Table 1 lists the diagnostic criteria. Chronic tic disorders other than TS are characterized by motor tics only or, less commonly, by phonic tics only. Chronic motor tic disorders occur more commonly than do TS, but follow a course similar to that of TS in terms of tic progression and comorbid affliction. The following discussion refers specifically to TS.

So-called simple tics involve a single muscle or functionally related group of muscles, and are generally brief, lasting less than a half second each. However, simple tics may occur in repeated bouts of rapid succession, each bout lasting seconds or more. What are known as complex tics involve more muscle groups, often have a more sustained duration or orchestrated pattern, and may appear purposeful. Complex phonic tics more clearly involve linguistically meaningful cession.

TABLE 1

Diagnoitic criteria for Tourette syndrome

- Both multiple motor and one or more vocal tics have been present at some time during the illness, although not necessarily concurrently.
- The tics occur many times a day (usually in bouts) nearly every day or intermittently throughout a period of more than one year, and during this period there was never a tic-free period of more than three consecutive months.
- The onset is before 18 years.
- 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).

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speech and language than do simple phonic tics. Table 2 lists examples of simple and complex motor and phonic tics.

The words “phonic” and “vocal” are often used interchangeably in the TS literature when describing oropharyngeal and nasopharyngeal noise-producing tics. Strictly speaking, a vocal tic is produced by the vocal cords; so, some phonic tics (such as sniffing) technically are not vocal tics. Though somewhat controversial, it is within accepted clinical practice to include phonic tics like sniffing in the vocal tic category.

Tics typically appear in early childhood (most often by 6 or 7 years), initially as simple motor tics, followed by phonic tics and more complex motor tics usually within the ensuing two years or so, although sometimes not until early or middle adolescence. At onset, motor tics usually are simple in nature, with fast, darting, and purposeless eye blinking or neck movements being the more common ones. Over months or years, motor tics may become increasingly complex, involving more areas of the body, generally progressing over time in a top-to-bottom and central-to-peripheral direction. In addition, the movements may become more elaborate or sustained and include combinations of movements, such as touching or tapping, hopping, or holding unusual postures, such as an outstretched neck, eyes deviated to one side, or other dystonic posturing. Motor tics may also be self-injurious to the skin (scratching oneself, frequently licking the lips resulting in severe chapping, or pulling out one’s hair), mucous membranes (biting the inside of the cheeks), spinal, muscular, or orthopedic structures (violent head-jerking, bending, or muscle straining), or vision (poking at the eyeballs).

Similarly, phonic tics usually are simple at onset, and are generally meaningless noises, such as coughing, grunting, or barking. Over time, more complex phonic tics may appear, and can include palilalia: repetition of one’s own words, such as “How are you? You? You? You?” sometimes with an exaggerated volume or pace to the repeated words. Other complex phonic tics include echolalia (repeating someone else’s words).

Although widely associated with TS, coprolalia (the utterance of socially unacceptable words or phrases, such as an obscenity or specific ethnic, racial, or religious slur) are not vocal tics. Though somewhat controversial, it is within accepted clinical practice to include phonic tics like sniffing in the vocal tic category.

Tics typically appear in early childhood (most often by 6 or 7 years), initially as simple motor tics, followed by phonic tics and more complex motor tics usually within the ensuing two years or so, although sometimes not until early
 Tourette Syndrome—Toward Diagnosis

Scenities or other socially inappropriate remarks) occurs in only a minority of patients. Because of its sensational potential, however, this feature of TS is routinely exploited in media portrayals. The common misconception that coprolalia must be present for a person to have TS may delay or obscure the diagnosis.

Note that the distinction between motor and phonic tics may be artificial and misleading. Technically, phonic tics are motor tics, just ones that produce sound, and classification of some tics as motor vs. phonic is not universally agreed on (e.g., a sniff or a tongue click). For now, at least, a phonic tic (present or past) must be present for the condition to meet the definition of TS.

Because a picture speaks a thousand words, you can better familiarize yourself with the diversity of tics by viewing examples online at www.mdvu.org/multimedia/slides/ts/default.htm.

Tics generally occur daily, but typically wax and wane. This quality of increasing and decreasing frequency of tics is evident both in short-term and long-term segments of time. As such, tics usually are seen to have a fractal quality; in other words, tics may occur in bouts over seconds or minutes separated by periods of tic-free behavior lasting for seconds or minutes. The bouts themselves may occur in bouts over hours, such that there are bouts of bouts. That pattern may continue for days, months, or longer (Figure 1).

Many people with TS describe a “premonitory urge” or sensory discomfort in a muscle or muscle group preceding the tic, which builds over time. This urge may be described as tension, pressure, a tickle, an itch, or another sensory experience. The urges are sometimes described less as a physical sensation than as a psychic phenomenon, such as anxiety or anger. External stimuli, such as a noise, a word, or an image, can pro-

![Figure 1](https://example.com/fig1.png)

**Figure 1**

**Bouts of tics take on a fractal character over time**

Tics occur in bouts (A), and these bouts occur in bouts (B – E), which may explain the waxing and waning nature of tics (E).

not tic. However, voluntary suppression comes at the cost of growing discomfort and distraction caused by the unrelenting, unfulfilled urge. Tic suppression is disruptive and can be more debilitating than the tic itself. The suppression may interfere with a child’s ability to focus and can increase anxiety. Tic suppression usually results in a rebound phenomenon at a later—safer, as it were—time and place (such as when the child returns home from school) during which the severity of the urges that had been suppressed compels more frequent and intense tics.

Stress, anxiety, fatigue, and other high-level emotional excitement exacerbate tics. Certain situations may reduce, or invoke suppression of, tics, such as settings or occasions where it may be embarrassing to tic—visits to the doctor’s office or speaking with friends,7 for example. Tics usually do not occur during sleep, but are more likely to happen at times and places of personal relaxation, such as while watching television at home.

Other conditions coexist more often than not

About 90% of patients with TS have one or more comorbid conditions8 (Table 3). These conditions are stand-alone diagnoses that occur more often in patients with TS than in the general population. Outward signs of comorbid conditions may not appear as dramatic as tics and may therefore be overlooked. Each of these conditions can follow its own natural course, both in onset and in progression or cessation (Figure 2).

More than half of TS patients will have comorbid attention deficit hyperactivity disorder (ADHD). The presenting clinical signs of ADHD usually precede the onset of tics. Psychostimulant medication, such as methylphenidate, is often used to treat ADHD. As a result, there has been a longstanding misperception that psychostimulant medication causes or exacerbates tics (I will address this misperception in Part 2).

Features of ADHD may include hyperactivity and restlessness, difficulty with sustained focus and attention, and distractibility. Loss of impulse control is another common feature. Among TS patients, some impulsive tendencies may, in fact, be responses to premonitory urges, a kind of a complex tic. Manifestations can include nonobscene, complex, socially inappropriate behaviors,9 such as pointing out an unpleasant body odor or a flaw in someone’s appearance. Children with comorbid ADHD are more likely to suffer greater psychosocial and academic consequences,10 and to have more disruptive or delinquent behavior, than are those without comorbid ADHD.11

Obsessive thoughts and compulsive behaviors occur in most children with TS. In about one quarter of these children, the thoughts or behaviors are so severe that they disrupt normal function or cause

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<thead>
<tr>
<th>TABLE 3</th>
<th>Comorbid conditions that complicate management of TS</th>
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</thead>
<tbody>
<tr>
<td>Attention deficit hyperactivity disorder</td>
<td></td>
</tr>
<tr>
<td>Obsessive-compulsive behaviors or disorder</td>
<td></td>
</tr>
<tr>
<td>Learning differences or disabilities</td>
<td></td>
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<tr>
<td>Anxiety disorders</td>
<td></td>
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<td>Mood disorders</td>
<td></td>
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<td>Sleep disorders</td>
<td></td>
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<td>Executive dysfunctions</td>
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<tr>
<td>Self-injurious behaviors</td>
<td></td>
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<td>Personality disorders</td>
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<td>Oppositional defiant disorder</td>
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anxiety, and constitute obsessive-compulsive disorder (OCD). It may be difficult to discriminate complex tics from compulsive behaviors. Some behaviors may appear more tic-like (such as evening things up, touching something over and over until it feels “just right,” or creating symmetry in surrounding objects) for which the unofficial term “compultics” (compulsions that look like tics) has been coined. Other behaviors may appear more like obsessions (such as a fear of contamination leading to repetitive or excessive hand washing) and typically cause significant emotional distress. Still other behaviors may appear to be a fusion of an impulsive behavior and a tic, such as the urge to touch a hot flame or stare at the sun, for which the unofficial term “impulsion” has been coined.

Learning disorders, especially in math and written language, are also common comorbid conditions. Intelligence is distributed normally among children with TS, but learning disorders prevent these children from functioning at their intellectual potential. One unifying feature of many learning disorders and of ADHD is executive dysfunctions. Executive functions describe goal-directed skills that allow us to use intelligence efficiently (organizing, planning, decision making, impulse control, social skills awareness, prioritizing). People with executive dysfunctions may appear scatterbrained or find themselves easily distracted despite attempts to concentrate. These people have difficulty with behavior that is governed by rules and do not easily learn to correct their behavior from past mistakes.

Tics and other comorbid conditions can impose learning difficulties above and beyond those caused by learning disorders. Examples include the interfering symptoms of ADHD; tics that hinder efficient thinking or functioning; strenuous energy spent suppressing tics; and obsessions focusing on irrelevant details or compulsions to perform functionless and time-consuming behaviors, such as counting to 10 every three sentences or rehearsing a song mentally before being able to answer each test question.

People with TS are prone to anxiety. Symptoms may include generalized anxiety, social phobias or other simple phobias, panic attacks, and sleep difficulties. Mood disorders are common and may include depression or dysthymia (chronic but low-grade depression), and, possibly, bipolar disorder. Episodic sudden and explosive rage attacks occur in a minority of patients with TS and may reflect an underlying neurologic disturbance or a severe effect of anxiety and mood disturbances. Both anxiety and depression may also be outcomes of stresses imposed by the emotional interference and stigmatizing of tics and comorbid conditions.

Sleep-related problems include increased arousal, prolonged wakefulness, and increased time to sleep latency, and are more likely in patients who have comorbid ADHD.

There is growing evidence that autism spectrum disorder may be a comorbid condition of TS, albeit an infrequent one.

One of the most regrettable outcomes in children with TS is poor self-esteem. These children are often the target of mockery and
What’s going on in the brain? Failed filters and overloaded circuits

If the brain were simple enough that we could understand it, we’d be so simple that we couldn’t.” So remarked Dr. Paul Greengard, winner of the 2000 Nobel Prize in Medicine or Physiology. No surprise, then, that the neurologic basis of TS is unclear. That uncertainty is reflected in inconsistent classifications given the condition: “Tourette syndrome” vs. “Tourette disorder.” A syndrome describes a set of signs and symptoms without an identified cause, whereas a disorder suggests an understood cause and, often, a predictable course. What seems clear is that the underlying mechanisms for pathology in TS and its comorbid conditions are linked.

According to our current model for understanding the pathophysiology of TS, the problem lies in the area of neurotransmission. Normally, dopamine and other neurotransmitters regulate messages transmitted along an important brain circuit that includes the basal ganglia, prefrontal and other cortex regions, and thalamus. One of the primary functions of this circuit is to process and filter information that refines controlled movement, thought, judgment, and the learning of behavior sequences. In TS, messages that should be filtered out instead slip through, in part because of faulty dopamine regulation at critical points in the circuit. The failure of appropriate filtering disinhibits thoughts, intellectual organization, sensory experiences, and behaviors.

What bridges tics and comorbid phenomena is a so-called urge-relief cycle. The overlapping circuitry may result in sensory urges relieved by tics, cognitive obsessive urges relieved by compulsive behaviors, or a sense of urgency relieved by an impulsive act (like blurt out an answer or interrupting someone) or by another socially inappropriate behavior (such as making a sudden off-color statement or observation). Other related manifestations that involve overlapping brain structures can result in depression, executive dysfunctions (such as poor organizational skills or inefficient intellectual processes), or sudden loss of emotional control that may be expressed as rage.

TS is a neurodevelopmental disorder so, as the brain develops, the form of the disorder evolves. The consequence is an array of colorfully unique features that vary not only from person to person, but also within any affected individual over time. To capture the range of potential symptoms, it has been suggested that an alternative name for TS and related disorders would be “developmental basal ganglia disorder,” implying a spectrum of disorders that may manifest as any or all of the behavioral phenotypes. Although it is essential that tics be present for a diagnosis, it is most reasonable to regard tics as one of several possible symptoms of this filtering disorder.

REFERENCES
TOURETTE SYNDROME—TOWARD DIAGNOSIS

TS- and/or OCD-affected parents, observed for up to five years, were three times as likely to develop tics than children with only one TS- and/or OCD-affected parent.15 Another study showed 43% of young children with a parent or sibling with TS developed some tic disorder during a four-year prospective study.16

An important point is made with an adage borrowed from the field of asthma: “All that tics is not Tourette.” Children with mental retardation, autism, or another neurologically-based developmental disability may have tics for reasons unrelated to the genes that cause TS.

PANDAS at play?
One controversial and intriguing theory posits an immunologic trigger for tics and obsessive-compulsive behaviors. PANDAS, or pediatric autoimmune neuropsychiatric disorders associated with streptococcal infection, was first defined in 1998.17 The PANDAS theory proposes that antibodies produced against group A β-hemolytic streptococcal infection cross-react with critical tissue in the brain and result in abrupt onset or exacerbation of symptoms. Proponents of the theory draw mechanistic similarity with Sydenham chorea, a manifestation of rheumatic fever. Beyond the scope of this review, PANDAS is the subject of an excellent commentary by Singer and Loiselle.18

Look to the history
Children and adolescents with tics alone, and those with comorbid difficulties, are likely to feel embarrassed or ashamed. Parents may feel a blend of personal failure, fear, and anger. To reduce these perceptions, a sensitive, empathic, and accepting interviewing approach is essential when taking a history. It is usually helpful to interview older children and adolescents privately, as well as in the company of their parents, to build a trusting alliance. Interviewing parents alone can allow an opportunity to discuss the impact that their child’s behaviors have on family or spousal relations. These approaches paint a picture of the broader, global experience of Tourette syndrome.

In taking the history and performing the physical examination, be mindful not only of tics but of clues to each of the comorbid conditions and considerations in the differential diagnosis (Table 4). The ability to suppress tics distinguishes the movements from other hyperkinetic movement disorders and is a useful diagnostic aid. Tics are seldom uncertain when observed directly. Exceptions include so-called compulsions and impulsions, as described. Another exception is stereotypies, such as those seen in children with pervasive developmental disorder. Such behaviors may include orchestrated behavioral sequences with touching, tapping, spinning, or other actions. Unlike tics, stereotypies tend to be completely voluntary, intentional, and self-soothing.

For the most part, the physical examination will be unrevealing. The money is in the history.19 The medical history should be comprehensive and include prenatal and perinatal factors, developmental history, neurologic history (e.g., seizure, coordination, trauma, sleep, atypical development), and confirmation of adequate hearing and vision. Ask about a family history of tics or comorbid conditions.

It is likely that several tics, past and present, will not be recognized, either because they were never noticed or because they have been forgotten. Reviewing the list of common tics (Table 2) can help the patient and family recall overlooked tics, and may be particularly helpful in diagnosing TS when there is any doubt about a history of phonic tics.

A social history can include information about parenting and discipline, family stressors, friendships, bullying, abuse, temperament, and self-esteem. Psychosocial function...
can be facilitated through use of the Pediatric Symptom Checklist (www.dbpeds.org/pdf/psc.pdf, with scoring instructions at www.dbpeds.org/pdf/jellinek.pdf), available without cost.

Scholastic data, including academic performance and behavior rating scales completed by teachers and parents, are vital in the initial assessment. Several of these scales are available free, such as the Vanderbilt Assessment Scales in the National Initiative for Children’s Healthcare Quality ADHD Toolkit (www.nichq.org/resources/toolkit).

Commonly, tics will not be observed during the office visit. Remember that tics are often suppressible (especially in the practitioner’s office), and they wax and wane. Some children will substitute more apparent tics with less apparent ones in the clinical setting, or will wait to release tics precisely when you are not looking at them. Others may disguise tics as seemingly purposeful movements, such as brushing hair out of the face in concert with a head jerking tic, sometimes referred to as parakinesias.

A careful physical examination may reveal telltale consequences of tics. These stigmata may include excoriated lesions on the skin from picking, ulcers on the buccal mucosa from biting, or circumoral chapping from lip-licking. The neurologic examination should be nonfocal in tic disorders, although “soft neuromaturational” signs may be present, such as ambidexterity or bedwetting beyond the age of 5 years.

Diagnostic laboratory, imaging, or electroencephalographic studies are not indicated in the evaluation for TS. On occasion, such studies may be indicated to rule out other specific diagnostic considerations when findings on the physical examination or history warrant further investigation.

**Diagnostic pitfalls to sidestep**

Although the diagnostic criteria for TS are straightforward, the diagnosis is often missed. Misconceptions about the syndrome explain much of this oversight. Most of the items listed in the differential diagnosis are uncommon; tics are not. Also, consider other tic disorders, including a chronic tic disorder (identical to TS except that the tics—past and present—are motor or phonic tics, not both) or a transient tic disorder (identical to a chronic tic disorder but the tics last less than a year [and longer than four weeks]).

In the absence of a careful history and exam, tics should never simply be explained away by telling parents that their child “will outgrow them.” When the tics persist, such ersatz reassurance may plant the mistaken belief that a nervous disposition is causing the tics, or that the child could stop the tics if only he or she really wanted to.

More often than not, presenting complaints will not be tics but rather complications of a comorbid condition. Complaints may include school failure, abdominal pain, or sleep problems suggesting depression; social skills problems; hyperactivity; or bedwetting. (Parents or patients may also misinterpret tics—complaining of “allergies” because of eye blinking or sniffing tics, for example.) In such cases, be on the alert for tics. Consider having parents provide videotaped samples of the child having tics at home, or try to discretely observe the child in the waiting area, where the child may be less likely to suppress the tics.

Some families of children with TS trek from physician to physician in search of an answer, and wait years before they are given a correct diagnosis. The understanding that the diagnosis brings to patient and family, and the chance to begin to truly manage the condition, is, for many who have or who live with someone who has TS, an incredible relief.
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