

## Tourette's Syndrome

Stephen P. Flynn, MD, Irwin Jacobs, MD, and William Guinter, PhD  
Cleveland, Ohio

**D**R. STEPHEN P. FLYNN (*Associate Director, Fairview General Hospital Family Practice Residency*): Today's Family Practice Grand Rounds involves a young boy with Tourette's syndrome. This patient raises several interesting issues for family physicians: the delayed recognition and response of the medical system, the role of the school in which the problem surfaced, and the importance of convening and working with the entire family.

The identified patient, Greg, is an 8-year-old third grader, who was initially brought to the Family Practice Center because of "fidgety" behavior and anal itching. Past medical history included recurrent ear infections and an episode of aseptic meningitis at the age of 4 years. No obvious behavioral problems were seen until the first grade. At that time the parents noted the onset of fidgeting, repetitive snorting noises, and nervousness. Because of these snorting noises and a family history of asthma, Greg was evaluated by an allergist, but with negative results. Later that year Greg began to have crying spells at night, stating that he did not want to attend school because "The kids pick on me," "I'm too short," and so on. An evaluation by the school psychologist revealed that Greg had average intelligence and grade-appropriate achievement. The school personnel felt he lacked self-confidence, recommending more positive support from both the teacher and parents.

In the second grade Greg continued his restless behavior. He also began to scratch and pull at the seat of his pants. These symptoms markedly increased in the third grade. Greg was unable to sit still during dinner and continued with his snorting and pulling at his pants. He was easily upset and agitated. He worried excessively about his homework, school performance, and his two brothers. The teacher began calling Greg's mother regularly about concerns over his behavior. After a classroom observation, the school psychologist suggested a pediatric neurology consultation to evaluate the possibility of Tourette's syn-

drome. At no time did the parents notice any facial tics or coprolalia (the involuntary utterance of obscenities) in Greg. There was no family history of tic disorders.

Greg's mother first brought him to our office for a second opinion during the spring of his second-grade year. At that time he was under the care of a pediatrician. The parents had expressed concern about Greg's behavior to his physician on various occasions. They were told that his examination was normal, and that his behavior would probably improve over time.

On his initial visit, a physical examination revealed an active child in the 25th percentile for height and weight, with otherwise normal physical findings. A pinworm preparation, done at the mother's request, was negative. Over the following 11 months Greg and his family made many visits to our office. We also received many telephone calls.

Primarily because of the teacher's concern, a more comprehensive evaluation of Greg's behavior was started. A consultation was obtained with a pediatric neurologist, Dr. Irwin Jacobs. He concurred with the diagnosis of Tourette's syndrome. Greg was started on a low dose of haloperidol. At the same time, a family assessment was felt to be indicated. The primary physician convened the entire family, including Greg, his two brothers, and parents. After this interview, the family was referred to the residency's family therapist for short-term counseling. The family had several sessions with the family therapist, who maintains regular contact with their family physician. Family therapy helped to reduce the emphasis on Greg as the identified patient, and also helped the family adjust to his medical condition. The parents reported a decrease in Greg's fidgeting and snorting.

This history is not atypical for Tourette's syndrome. Frequently, the condition goes unrecognized for years, as parents repeatedly seek medical attention. Often allergy and otolaryngic evaluations are performed because of certain vocal tics; something unusual in this case is the lack of multiple facial and other tics. Essentially, Greg had only a vocal tic (snorting) and a complex motor tic (pulling at his pants). Another important aspect was the concern of the schoolteacher, who helped precipitate a more thorough evaluation in this case. We will address the role of the school system later in this conference.

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From the Department of Family Medicine, Case Western Reserve University, and Fairview General Hospital, Cleveland, Ohio. Requests for reprints should be addressed to Dr. Stephen P. Flynn, Hassler Center for Family Medicine, 18200 Lorain Avenue, Cleveland, OH 44111.

**TABLE 1. DIAGNOSTIC CRITERIA\* FOR TOURETTE'S DISORDER**

1. Both multiple motor and one or more vocal tics 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 one year
3. The anatomic location, number, frequency, complexity, and severity of the tics change over time
4. Onset before the age of 21 years
5. Occurrence not exclusively during psychoactive substance intoxication or known central nervous system disease, such as Huntington's chorea and postviral encephalitis.

\* From the DSM-III-R<sup>1</sup>

DR. IRVIN JACOBS (*Pediatric Neurologist*): Greg is a wrestler at school. Even when wrestling, he tugged at the seat of his pants. It is unusual that he would do this when involved in something he liked doing. If this movement were just a habit, it would have probably disappeared when Greg was intensively involved in activities such as wrestling. Although Tourette's syndrome can be very severe, we are beginning to recognize milder cases more often. The statistics that applied to Tourette's syndrome ten years ago are not really applicable today.

Tourette's syndrome was first described by Gilles de la Tourette in 1885. He described nine cases. Some of these cases involved people jumping up and down and spitting. The syndrome was an oddity, and very few cases were reported. I recall hearing about my first case as a resident in neurology 15 years ago. People began to recognize the syndrome more often in the early 1960s.

With the discovery that haloperidol was an effective medication for treating this disorder, interest in Tourette's syndrome increased, and more patients came to physicians for treatment. Another reason for the increased attention was because of the efforts of the Tourette's Syndrome Association. This organization, which consists of patients with Tourette's syndrome and their families, has played a major role in educating both physicians and the public.

The DSM-III-R criteria<sup>1</sup> for Tourette's syndrome are listed in Table 1. The age of onset is usually 2 to 15 years. Greg's case is typical, in that Tourette's syndrome is much more common in the male patient, more common in whites, and probably is genetic in origin. The disorder is characterized by multiple, involuntary, spasmodic, frequently repeated, stereotyped movements. We used to believe that these movements disappeared during sleep, but now there is evidence to suggest that they may persist at night. Symptoms are clearly worse during times of stress. Patients can voluntarily suppress these movements

for a brief period of time. The motor tics often begin in or around the face (blinking eyes, facial twitching, shoulder jerking), but can present anywhere. In addition to motor tics, these patients have vocalizations, which may be a particular word, a sniff, clearing of the throat, or a whole variety of noises. Perhaps the most disturbing of these is coprolalia. In fact, not only do those afflicted say these words out loud, but also they sometimes have obsessive thoughts and keep saying these words to themselves or write them repetitively. The definition of Tourette's syndrome requires that these symptoms continue for more than one year. Over time, the particular symptoms wax and wane, which is important to recognize because this specific characteristic makes management very difficult. Drug therapy must be regulated accordingly to how the patient is doing.

Tic disorders can be divided into three different syndromes. The most common syndrome is called *transient* tic disorder of childhood. These tics can be either motor or vocal; it can be just a sniffing sound. Transient means occurring less than one year. The movements or vocalizations can occur with varying frequency. There may be eight to ten tics in the course of a minute. The tics may quiet down for several hours, or there may be very rapid movements for many hours. The tics are not only variable throughout the day, but vary over the course of weeks and months as well.

If the tic continues for more than one year and remains only motor, it is then called a *chronic* motor tic disorder. If the tic continues over one year and it consists of vocalizations and motor components, we give it the name *Tourette's syndrome*. It is not possible to predict when a patient comes in with a tic that has been present for six weeks whether that tic will stop, or whether the patient will develop Tourette's syndrome.

Findings on physical examination are usually normal except for the tics. Mild abnormalities may be seen on a computerized tomography scan of the head, or in the electroencephalogram, but nothing diagnostic of Tourette's syndrome will be revealed. The diagnosis is made on the basis of observation and history. The history is most important because the child can suppress these movements and may not feel comfortable enough to demonstrate the tics in the office until the third or fourth visit. If the child does suppress the tics, he or she begins to feel what some describe as inner tension and may be compelled to release it. Some patients say that they do not feel they are adequately controlled unless this inner tension is gone. When you ask why they do it, their response is usually, "Because I feel better when I do it."

The differential diagnosis is limited. When a patient presents with tic movements and a typical history, there is little else the diagnosis can be. You can consider focal seizures, but focal seizures would not change in character

*continued on page 30*

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The onset of chronic active hepatitis may be insidious, and patients receiving long-term therapy should be monitored periodically for changes in liver function. If hepatitis occurs, the drug should be withdrawn immediately and appropriate measures taken. Peripheral neuropathy, which may become severe or irreversible, has occurred. Fatalities have been reported. Conditions such as renal impairment (creatinine clearance under 40 ml per minute), anemia, diabetes mellitus, electrolyte imbalance, vitamin B deficiency, and debilitating disease may enhance the occurrence of peripheral neuropathy. Cases of hemolytic anemia of the primaquine sensitivity type have been induced by nitrofurantoin. Hemolysis appears to be linked to a glucose-6-phosphate dehydrogenase deficiency in the red blood cells of the affected patients. This deficiency is found in 10 percent of Negroes and a small percentage of ethnic groups of Mediterranean and Near-Eastern origin. 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Use of this drug in women of childbearing potential requires that the anticipated benefit be weighed against the possible risks. **Labor and Delivery:** In case CONTRAINDICATIONS. **Nursing Mothers:** Nitrofurantoin has been detected in breast milk, in trace amounts. Caution should be exercised when Macrochantin is administered to a nursing woman, especially if the infant is known or suspected to have a glucose-6-phosphate dehydrogenase deficiency. **Pediatric Use:** Contraindicated in infants under one month of age. (See CONTRAINDICATIONS.) **ADVERSE REACTIONS: Gastrointestinal:** Hepatitis, including chronic active hepatitis, and cholestatic jaundice occur rarely. Nausea, emesis, and anorexia occur most often. Abdominal pain and diarrhea are less common. Gastrointestinal reactions of dose-related nature can be minimized by reduction of dosage. **Respiratory:** Chronic, subacute, or acute pulmonary hypersensitivity reactions may occur. Chronic pulmonary reactions are more likely to occur in patients who have received continuous treatment for six months or longer. Malaise, dyspnea on exertion, cough, and altered pulmonary function are common manifestations which can occur insidiously. Radiologic and histologic findings of diffuse interstitial pneumonitis or fibrosis, or both, are also common manifestations of the chronic pulmonary reaction. Fever is rarely prominent. The severity of chronic pulmonary reactions and their degree of resolution appear to be related to the duration of therapy after the first clinical signs appear. Pulmonary function may be impaired permanently, even after cessation of therapy. The risk is greater when chronic pulmonary reactions are not recognized early. In subacute pulmonary reactions, fever and eosinophilia occur less often than in the acute form. Upon cessation of therapy, recovery may require several months. If the symptoms are not recognized as being drug-related and nitrofurantoin therapy is not stopped, the symptoms may become more severe. Acute pulmonary reactions are commonly manifested by fever, chills, cough, chest pain, dyspnea, pulmonary infiltration with consolidation or pleural effusion on x-ray, and eosinophilia. Acute reactions usually occur within the first week of treatment and are reversible with cessation of therapy. Resolution often is dramatic. **Neurologic:** Peripheral neuropathy, which may become severe or irreversible, has occurred. Fatalities have been reported. Conditions such as renal impairment (creatinine clearance under 40 ml per minute), anemia, diabetes mellitus, electrolyte imbalance, vitamin B deficiency, and debilitating diseases may increase the possibility of peripheral neuropathy. Less frequent reactions, of unknown causal relationship, are nystagmus, dizziness, headache, and drowsiness. **Dermatologic:** Exfoliative dermatitis and erythema multiforme (including Stevens-Johnson Syndrome) have been reported rarely. Transient alopecia also has been reported. **Allergic Reactions:** Lupus-like syndrome associated with pulmonary reaction to nitrofurantoin has been reported. Also, angioedema, maculopapular, erythematous or eczematous eruptions, urticaria, rash, and pruritus have occurred. Anaphylaxis, sialadenitis, pancreatitis, arthralgia, myalgia, drug fever, and chills or chills and fever have been reported. **Hematologic:** Agranulocytosis, leukopenia, granulocytopenia, hemolytic anemia, thrombocytopenia, glucose-6-phosphate dehydrogenase deficiency anemia, megaloblastic anemia, and eosinophilia have occurred. Cessation of therapy has returned the blood picture to normal. Aplastic anemia has been reported rarely. **Miscellaneous:** As with other antimicrobial agents, superinfections by resistant organisms, e.g., *Pseudomonas*, may occur. However, these are limited to the genitourinary tract because suppression of normal bacterial flora does not occur elsewhere in the body. **OVERDOSAGE:** Occasional incidents of acute overdosage of Macrochantin have not resulted in any specific symptoms other than vomiting. In case vomiting does not occur soon after an excessive dose, induction of emesis is recommended. 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continued from page 28

from week to week or month to month. In Huntington's or Sydenham's chorea, the movements are irregular, more constant, and more exaggerated than in tic disorders. Wilson's disease, cerebral palsy, and other movement disorders are excluded because the movements associated with these disorders are not stereotyped, as they are in Tourette's syndrome.

Children with Tourette's syndrome can also present with the symptoms of an attention-deficit disorder—impulsiveness, distractibility, and lack of concentration. Family studies have suggested that the same genetic factors that influence Tourette's syndrome can also be associated with an attention-deficit disorder. Some patients started on methylphenidate because of an attention disorder have developed Tourette's syndrome. Studies suggest that Tourette's syndrome would probably have developed anyway in these patients because of family and genetic factors, but certainly the methylphenidate can precipitate or aggravate the tic disorder. How do you treat someone who has Tourette's syndrome and an associated attention-deficit disorder? There is no contraindication to using a second medication, eg, both haloperidol and methylphenidate. Some patients with an attention-deficit disorder and Tourette's syndrome will improve on haloperidol alone.

The mainstay of medication for Tourette's syndrome is haloperidol. Haloperidol is a dopamine-blocking agent that has potential side effects. Children can become depressed, sedated, and start to eat excessively as well as develop some of the complications of all the major tranquilizers, including Parkinsonian symptoms and tardive dyskinesia. For these reasons, I often try to take patients off haloperidol for a period of time, perhaps once a year.

Another medication that can be used is pimozide, another dopamine-blocking agent that has been approved recently for use in Tourette's syndrome. Essentially it has the same effect as haloperidol but may cause less sedation. It can be used if haloperidol does not work. The long-term effects are not known, but pimozide may have some cardiotoxic effects. Electrocardiograms are required when using pimozide, so the initial drug of choice is still haloperidol. Clonidine, an antihypertensive medication, has been tried, but is generally not so effective as haloperidol. Once you have established that the medication works, you can let the parents try to regulate the medication themselves. If the child is doing well for a period of time, the dosage can be lowered. If the patient looks as though he is under a lot of stress, then the dosage can be raised, within limits. If the symptoms are not well controlled, I ask the parents to call me.

When should medication be prescribed? Only in those cases where the condition is causing a problem. If a child has a mild tic and seems to be handling it well, if his friends do not care, and if the teacher is not complaining,

I would not recommend medication. I prescribe medication only when the condition begins to affect the functioning of the child or the family. I always prescribe the smallest effective dose.

It was interesting to hear about the family counseling sessions that have been occurring with Greg. For many years psychotherapy was the only available treatment for Tourette's syndrome, and it just did not work. Family counseling is important in helping families to deal with and understand what is going on and, as such, is a valuable adjunct.

Finally, there is the problem of knowing what to do when you encounter a patient who has very early signs of tics, such as a child blinking his eyes. Should you put him on haloperidol if he's only had the tic for several weeks? Could you prevent the evolution of the disease? No study has been done to answer these questions.

Until recently we thought Tourette's syndrome was a life-long problem. This impression was based on the earlier reports of the more severe cases of Tourette's syndrome. Now it is becoming more evident that patients are coming in with much milder symptoms, and parents may say that they had similar tics in childhood that disappeared after several years. So the incidence of spontaneous resolution in Tourette's syndrome may be 10 to 20 percent.

DR. WILLIAM GUINTER (*School Psychologist*): In treating behavior disorders in children, I favor a cooperative approach with the family, the school, and the medical system. The school's initial role is to identify a behavior disorder; the best places to discover a behavior disorder are daycare centers or schools. Next, the school psychologist can observe and record the type and duration of any abnormal behavior. The school psychologist can act as the physician's eyes and ears. When physicians administer medication, it would be appropriate to ask the school psychologist to observe the student. We can provide data about how the student is progressing.

I caution you about using teachers to monitor progress. Many teachers would want children with behavior disorders medicated, and they will give you the data to support the need for prescriptions. An objective observer, whether the principal, the counselor, or the school nurse, can be an excellent advocate for kids. The school psychologist is responsible for documenting the behavior of the students in the classroom, and we will do that for physicians. Many physicians have questionnaires they want given in the school setting. Our primary concern is getting the family and physician involved to help handle a behavior disorder.

There are two aspects of behavior management in which I often become involved. The first is promoting consistency in the disciplinary environment throughout the school. In most schools, there are many teachers in one building and each may approach behavior management

in a different way. Principals also have different styles of behavior management. From a psychologist's perspective, one of the most important elements in handling behavior or attention-deficit disorders is consistency.

The second aspect of behavior management is in designing programs for specific children and their teachers in the classroom. We identify the desired behavior and what factors lead to this behavior. We try to establish actions by the teacher to reinforce the desired behavior. We also try to involve the family so that the same behavior is reinforced at home.

Behavior does not change in a brief period of time, so monitoring is another major area where the school psychologist can help. Because change often occurs in small steps, people tend to overlook the child's progress. Without proper monitoring, growth will not be recognized. It is important to say to the child, "You're improving, you're going in the right direction." What often happens, however, is that only the residual misbehavior is noticed, and this leaves the child thinking, "I'm trying like crazy and nobody sees it."

Finally, it is our responsibility to foster some kind of internal behavior control with the child, so that when medication is reduced or removed, the child does not regress.

DR. JACK MEDALIE, (*Chairman, Department of Family Medicine, Case Western Reserve University*): We often see tics in childhood. For example, a child will get a foreign body in his eye, and after it is removed, the child goes on blinking for some time. Then the mother or little brother exhibits a tic that tends to occur under stressful conditions.

There seems to be a family tendency to react to anxiety in certain ways. Behavior problems in children are often symptoms of family dysfunction, and family diagnosis is also very important. From this discussion it seems the whole focus of attention centers on the one child, and one wonders what happened to the others. Often in families where medical attention is focused on one individual, there is also a hidden patient, another family member at high risk for illness. In this case, I think the hidden patient was the whole family. I think family counseling would help to get some equilibrium back.

#### Addendum

Greg was placed on a small dose of haloperidol for four to five months and appeared to improve. After the commencement of the new school year, the medication was stopped by his parents, and Greg continued to do well. His mother reported that he performed well in fourth grade for six months without medication. Recently, with the stress of moving to a new house, Greg has exhibited some recurrence of his vocalization tics.

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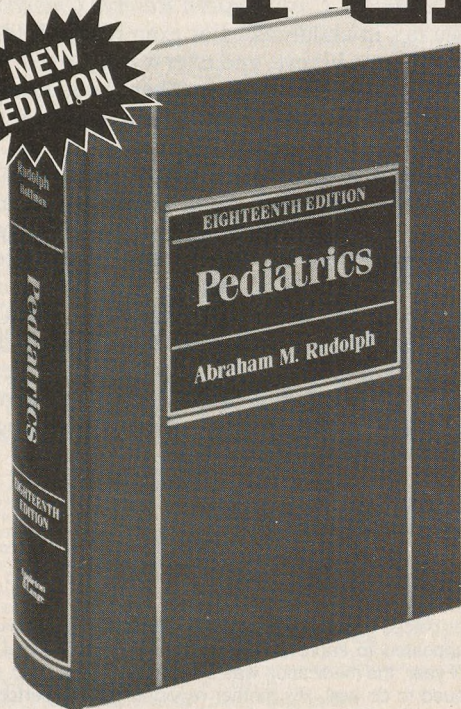
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