Exposure and response prevention in the treatment of tics in Tourette's syndrome

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1.1 History

In 1885, the French physician George Gilles de la Tourette reported on nine patients with chronic tic disorders characterized by motor and vocal tics. One famous case history was the Marquise de Dampierre, a French noblewoman, and also a patient of Charcot, who had persistent body tics, barking sounds and utterances of obscenities. Tourette's work was an important advance in the classification of the movement disorders. He proposed that the symptoms he observed constituted a new disease which should be separated from the chorea movement disorders. Charcot rewarded Tourette for his description of the symptoms and their developmental course by naming this disease after him. In his early writings, Tourette suggested that Tourette's syndrome (TS) was an inherited disorder, but in a later paper, he (as cited in Shapiro, Shapiro, Bruun, & Sweet, 1978) focused on the patients's mental instability and the nervous or mental disorders in the family background. Although some of the original clinical characteristics still remain valid, many diagnostic features have been redefined since these first reports. Similarly, suggestions that TS was a rare disorder and of psychogenic origin have been replaced by evidence suggesting a more common disorder of genetic origin with neurotransmitter abnormalities (Shapiro et al., 1978; Singer, 2005).

1.2 Phenomenology, definition and diagnosis

Motor and vocal tics

The main feature of TS is motor and vocal tics. Tics are characterized by sudden, rapid, recurrent, non-rhythmic, stereotyped motor movements or sounds (APA, 2000). Tics have different degrees of severity and duration and can be divided into simple and complex tics. Simple motor tics are brief, rapid movements that manifest themselves suddenly, frequently in bouts, and involve one muscle group. Common simple motor tics include eye blinking, head jerking, and shoulder shrugging. Complex motor tics are abrupt movements or a more coordinated sequence of movements that involve multiple muscle groups and may persist longer. Complex motor tics serve no purpose, even when they seem to be more goal-directed, e.g., touching, hitting, smelling, jumping, or obscene gestures. Simple vocal tics include sounds and noises such as sniffing, throat clearing, coughing, and grunting. Complex vocalisations include the repetition or mimicking of other people's words (echolalia), the repetition of one's own words (palilalia), or the uttering of obscene words (coprolalia) (Leckman, 2003; Singer, 2005).

Premonitory sensations

Prior to the tics patients often experience so-called premonitory sensations, i.e., unpleasant

sensations of a somatosensory nature (such as itching, tension, prickling, pain, heat or cold) in the skin, muscles, joints, bones or vocal cords. These sensations prompt the performance of tics followed by feelings of momentary relief (e.g., Leckman, Walker, & Cohen, 1993). Besides the local or regional premonitory sensations, there may be nonlocalizable and less specific premonitory sensations, such as urges or general tension. In order to relieve the uncomfortable urge a particular movement is repeated until 'it feels good' or 'it feels just right'. These 'just-right' perceptions frequently involve the sensory modalities of sight and touch (Leckman, Walker, Goodman, Pauls, & Cohen, 1994).

Premonitory sensations had already been observed by Patrick in 1905 (as cited in Scahill, Leckman, & Marek, 1995), who reported that sensations were the origin of tics which, in turn, were the result of yielding to those urges. However, it was not until 1980 that Bliss, who suffered from Tourette himself, drew attention to these sensations. He published a detailed report of his own experience of premonitory sensations and hence questioned the conventional view that tics are involuntary in character. For how involuntary is a movement that can be suppressed? Lang (1991), in accordance with Bliss (1980), suggested that the motor execution of the tic is 'intentional'. He found that of the patients with a movement disorder other than tics, 93% assessed their abnormal movements as completely involuntary, as compared to 32% of the patients with a tic disorder. The remaining 68% of the tic disorder patients felt that their tics were voluntary but at the same time compelling, since they were necessary in order to achieve an end. The percentage of TS patients (96%) experiencing the tic as intentionally executed was even higher (Lang, 1991). Fahn (1993) proposed that the tics could best be described by using the word 'unvoluntary'. He stated that the motor or vocal action produced in response to a local premonitory sensation is voluntary, whereas the preceding discomfort is involuntary. Currently, premonitory sensations are considered the involuntary component of the tics, i.e., the stimulus, whereas the actual movement is the volitional response to the premonitory sensation (Kwak, Vuong, & Jankovic, 2003). The term 'sensory tics', introduced by Shapiro, Shapiro, Young, & Feinberg (1988) to describe another type of tic was abandoned by most researchers, because a sensory sensation is not considered a tic behavior anymore. In line with this scientific debate, the term 'involuntary' that was included in the DSM-III-R (APA, 1987) definition of tics, was omitted in the DSM-IV edition (APA, 1994).

The literature on TS has inconsistently used the term premonitory sensations. The Tourette Syndrome Classification Study Group (1993) for example used a broad definition to describe the different sensations that precede tics. The Study Group defined 'sensory phenomena' as 'generalized or focal uncomfortable feelings or sensations preceding tics that usually are relieved by a movement' (1993, p. 1016). This description encompasses urges that are not associated with sensations as well as those that are preceded by (visual, auditory, tactile, or olfactory) sensations. Alternatively, Chee and Sachdev (1997)

excluded urges and 'just-right' perceptions in their definition of what they called 'sensory tics', since these urges usually involve cognitive experiences, or, when felt in a body part, are not localized close to the skin (Chee & Sachdev, 1997). Research has demonstrated that premonitory sensory sensations are a distinctive feature of TS and that 'just-right' perceptions occur in both TS and obsessive-compulsive disorder (OCD; Leckman et al., 1994; Miguel et al., 1995, 2000).

Table 1-1 summarizes the findings of studies that examined the occurrence of premonitory sensations in TS. The different percentages of TS patients who recognized these sensations reported in the studies may reflect differences in method of data collection, sample size, sample age, and the definition of sensations that was used. Shapiro et al. (1988) reported the lowest incidence of premonitory sensations in a large sample size (see Table 1-1). Shapiro et al. acknowledged that their figure was likely to be an underestimation because they had been 'less sensitive to the existence of these symptoms' in their retrospective data collection. To date, one study (Chee & Sachdev, 1997) assessed the occurrence of premonitory sensations in TS patients as compared to a healthy control group. The TS group (28%) experienced significantly greater life-time prevalence of 'localizable physical sensations' than the healthy control group (8%). In summary, it can be concluded that premonitory sensations are a common feature of TS (also see Table 1-1).

To have premonitory sensations does not mean that every tic is preceded by a sensation or urge. The anatomic regions that have the highest density of premonitory sensations are the head, shoulder girdle, throat, hands, midline of the stomach, and front of the thighs and feet. Simple tics involving rapid behaviors, like eye blinking, often are not preceded by premonitory sensations (Leckman et al., 1993). Lang, Consky, and Sandor (1993) suggested that the development of a new tic (especially a complex tic) consists of a voluntary, purposefully performed movement that is accompanied by a premonitory sensation. When the tic is repeated, it develops into a perpetuated automatic behavior, of which the patient is unaware. Only when the tics are pointed out, the patient becomes aware of the tic and its intentional, premonitory aspect. This theory is supported by the observation that premotor potentials are absent during automatic tic performance, whereas these potentials are present when patients are asked to mimic the tic (Obeso, Rothwell, & Marsden, 1982). However, even long-standing simple tics can continue to be associated with a consciously perceived premonitory sensation or urge (Lang, 1991).

Premonitory sensations occur on average 3 years after the onset of tics, and it is reported that children under the age of 10 years are not usually aware of these sensations (Leckman et al., 1993). Banaschewski et al. (2003) studied a relative large sample of 254 TS patients and found that rates for awareness of premonitory sensations increased from childhood to adolescence, from 34% (8 to 10 years), to 56% (11 to 14 years) to 68% (15 to 19 years). Contrary to these findings, Woods, Piacentini, Himle, and Chang (2005) reported that only one (2%) of 42 youths with a mean age of 11 years denied the presence

of any premonitory sensation. However, children younger than 10 years old were not as consistent in their reporting of these sensations as older patients (Woods et al., 2005). To date, it remains unclear whether premonitory sensations are present from the onset of the disorder, but do not enter young children's awareness, possibly due to a lack of introspective ability, or that the sensations emerge with development, after the onset of tics, as a result of disease progression or cognitive development (Leckman et al., 1993; Woods et al., 2005). Banaschewski et al. (2003) reported that tic awareness increases with age independent of tic duration and age of tic onset, which seems to indicate that development of premonitory sensations reflects cognitive development rather than disease progression.

Other associated features and classification

TS patients are not only sensitive to perceptions of internal body sensations. A wide range of auditory, tactile or visual external cues may also prompt tics (Cohen & Leckman, 1992; Eapen, Moriarty, & Robertson, 1994). The nature of these external cues is usually highly specific for individual patients. Echo phenomena such as echopraxia, echolalia and palilalia appear in response to external cues. Tics can also be triggered by other people coughing and sniffing. Sometimes, when a patient is interviewed about his repertoire of tic symptoms, tics immediately (re)appear. In addition, a number of TS patients report urges to perform acts that are senseless, dangerous, or forbidden in response to external stimuli. Examples range from the urge to count letters while watching television, to squeeze the tail of one's beloved cat, to touch a hot iron, to break a glass and subsequently cut oneself with the broken glass, to suddenly turn over the steering wheel of a car while driving down a highway, or to shout in a quiet church. Furthermore, some patients show 'site sensitization', meaning that they are unusually aware, distracted and distressed by particular sensori stimuli, such as long hair or tags in new clothing. Unless the hair is cut or the tags are removed, it is difficult to pay attention to other things than to perform tics, i.e., to shake one's head in order to move off the hair, or to pull at the clothing tags (Leckman, 2003; Verdellen & Keijsers, 1999).

Another feature of tics is their suppressibility. TS patients are able to suppress tics voluntarily for minutes up to hours (Jankovic, 1997; Kurlan, Lichter, & Hewitt, 1989). There are indications that the intentional character of tics facilitates tic suppression. Relationships have been found between the experience of premonitory sensations and the ability to suppress tics (Banaschewski et al., 2003; Kurlan et al., 1989; Leckman et al., 1993; Peterson et al., 1998). Both the ability to suppress tics and the intentional character of tics helps to differentiate TS from other hyperkinetic movement disorders, such as chorea, athetosis, myoclonus, dystonia, and paroxysmal dyskinesias (Koller & Biary, 1989; Lang, 1991). Although voluntary tic suppression is often believed to result in a rebound of tics, i.e., an intensified tic expression due to a build-up of inner tension (e.g., Fahn, 1993; Jankovic,

Table 1-1. Reports of premonitory sensations in TS

Study	Sample size	Method	Definition and symptoms included	Frequency		Age range
				n	%	
Shapiro et al., 1988	1237	Retrospective	Sensory tics: physical sensory sensations	105	8.5	4-69
Kurlan et al., 1989	34	Interview	Sensory tics: local physical sensations, generalized physical sensations and generalized mental sensations (e.g., urge, compelling thought, nervousness)	26	76.5	14-64
Lang, 1991	48	Interview/questionnaire	Tics: intentionally executed behaviors in response to an inner need or involuntary urge	45	95.8	7-57
Cohen & Leckman, 1992	28	Interview	Sensory phenomena: physical sensations and urges, impulsivity	22	79.5	9-60
Leckman et al., 1993	132	Questionnaire	Premonitory urges: local physical sensations, general tension, mental sensations and urges ('just-right' feelings)	123	93.2	8-71
Miguel et al., 1995	17	Structured interview	Sensory phenomena: local physical sensations, generalized feelings or urges, 'just-right' perceptions	12	71	22-43
Chee & Sachdev, 1997	50	Interview	Sensory tics: localizable physical sensations	14	28	10-32
Banaschewski et al., 2003	254	Questionnaire	Premonitory sensory phenomena: uncomfortable bodily and cognitive sensations, local tension or sensations, generalized inner tension, sensory urge	94	37	8-19
Kwak et al., 2003	50	Questionnaire	Premonitory sensory phenomenon: localized sensations, generalized urges, tension, 'just-right' perceptions	46	92	6-40
Woods et al., 2005	42	Self-report rating scale	Premonitory urge phenomena: 'phenomenological descriptions of premonitory urge experiences found in the literature (e.g., pressure, itch, 'just-right' perceptions)'	41	98	8-16

1997; Leckman, 2003), there is no empirical evidence to support this theory (Himle & Woods, 2005).

Characteristically, tics change in number, frequency, anatomic distribution, and complexity over time. In a typical TS course, tics first appear before the age of seven years and usually begin with transient periods of eye blinking or other simple facial tics. Vocal tics usually appear 1 to 2 years after the onset of the simple motor tics. Motor and vocal tics frequently change and evolve into more complex tics. The severity and nature of symptoms may wax and wane, with peeks of severity at 8-12 years of age in most cases. Although in most patients symptoms decrease during and after adolescence, only 8% of the patients have complete and permanent remission (Leckman, 2003; Rampello et al., 2006; Singer, 2005). Tics not only vary over time within patients, there is also a large inter-individual variation in its severity. The environmental or biological factors that cause tics to wax and wane are unknown. Stress, anxiety, physical exercise, excitement, fatigue, exposure to heat, caffeine, central nervous system stimulants, and streptococcal infection may exacerbate tics, whereas distraction, concentration, use of alcohol, nicotine or cannabinoids, relaxation and sleep may decrease tic severity (Rampello et al., 2006; Silva, Munoz, Barickman, & Friedhoff, 1995; Singer, 2005).

TS is diagnosed based on the patient's history and clinical assessment. There is no diagnostic laboratory test to establish the diagnosis. Although there is strong support that the disorder is hereditary and genetically predisposed, responsible genes remain to be identified (Pauls, 2003). According to DSM-IV-TR (APA, 2000), TS is characterized by the presence of both multiple motor tics and one or more vocal tics throughout a period of more than 12 months, during which time there was never a tic-free period of more than 3 consecutive months. DSM-IV-TR requires a TS tic onset before the age of 18.

Attention deficit hyperactivity disorder (ADHD) and OCD are the most frequently reported comorbid diagnoses in TS. Comorbidity rates for ADHD range from 50 to 75% (Comings & Comings, 1987; Spencer, Biederman, Harding, Wilens, & Faraone, 1995), and comorbid OCD is reported to occur in approximately 20 to 60% of TS patients (Grad, Pelcovitz, Olson, Matthews, & Grad, 1987; Pitman, Green, Jenike, & Mesulam, 1987). There has been debate as to the relationships of TS to ADHD and OCD. Some authors have suggested that TS and ADHD are not related, but that the high comorbidity rate is the result of ascertainment bias (Pauls, Leckman, & Cohen, 1993). It may be difficult to distinguish ADHD from the typically abrupt, often explosive movements of tic behaviors. Others have suggested that TS and ADHD are genetically related (e.g., Comings & Comings, 1987). Pauls et al. (1993) reported that there may be two types of patients with TS plus ADHD, one where ADHD is independent of TS and another where ADHD is secondary to TS. When ADHD symptoms precede the development of tics, the disorders do not appear to be related, whereas when ADHD symptoms follow the development of tics, the disorders appear to be related (Pauls et al., 1993). Moreover, the high ADHD comorbidity rates

reported in the literature may reflect TS related impaired inhibition (Walkup et al., 1999). As to the relationship of TS to OCD Shapiro et al. (1978) and Shapiro and Shapiro (1992) noted that in the research literature many TS patients were probably misdiagnosed with a comorbid diagnosis of OCD due to mistakenly viewing TS symptoms (e.g., repetitive touching, sensory sensations, symmetry behavior) as compulsions. Shapiro et al. (1978) did not find OCD symptoms more often in TS patients than in a control group of psychiatric outpatients (13% in the TS group compared to 15% in the control group). However, Shapiro et al. identified a small subgroup of TS patients with comorbid OCD. Cath et al. (2000, 2001) carried out a series of studies aimed at differentiating TS from OCD and proposed two subtypes of OCD: one with and one without tics. Whereas tics were associated with an increased frequency of repetitive behaviors that are unrelated to avoidance of harm, such as mental play, echo phenomena, touching and self-injurious behavior, the absence of tics was associated with an increased frequency of contamination and washing rituals, aggressive repetitive thoughts, and repetitive requests for reassurance. In line with Shapiro et al. (1978), Cath et al. (2000) concluded that the repetitive behaviors seen in TS patients have an impulsive character that should be considered as an integral part of TS and should be differentiated from OCD.

1.3 Pathophysiology

The pathophysiological basis of tics in TS and its relation to voluntary/involuntary aspects is not yet understood. However, several lines of biochemical, neurophysiological, neuroimaging, and genetic research indicate that basal ganglia structures and cortico-striato-thalamo-cortical (CSTC) circuits may play a crucial role in the pathophysiology of tic disorders (Hoekstra et al., 2004; Singer & Minzer, 2003). Sensorimotor, orbitofrontal, and association cortices are considered relevant, and probably includes the limbic system as well. Hypothesized abnormalities are disorders of excess excitation or diminished inhibition, disruptions in frontal cortex, striatum, and abnormalities of various synaptic neurotransmitters (Peterson et al., 1999; Singer, 2005).

There is evidence that the basal ganglia and related cortical and thalamic structures are involved not only in motor control but also in the occurrence of premonitory sensations. A recent functional MRI study (Fattapposta et al., 2005) showed that the supplementary motor area (SMA) is continuously activated in TS during the performance of voluntary movements, indicating an abnormal activation pattern of the motor cortex. In addition, electrical stimulation of the SMA evokes feelings of tension and an urge to move (Fried et al., 1991; Lim et al., 1994).

Studies into the existence of premotor potentials, to investigate the voluntary nature of tics, have contradictory outcomes. Although Obeso et al. (1982) found no evidence