The colorful spectrum of Tourette syndrome and its medical, surgical and behavioral therapies

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ABSTRACT

Tourette syndrome (TS) is a common neuropsychiatric disorder, more common in males than females, with onset before age 18. TS is characterized by multiple motor tics and one or more vocal/phonic tics, persisting for more than a year. Tics are involuntary, abrupt, rapid, repetitive, but non-rhythmic movements or sounds (vocalizations). They are preceded by an inner urge. Tics can be temporarily suppressed, but this leads to a powerful re-emergence. The performance of tics results in immediate but transient relief.

Motor and vocal tics are classified as simple or complex. Tics emerge many times during the day and display spontaneous fluctuations in frequency, type, severity and complexity. Tics improve during concentration, worsen during relaxation or when under stress and occasionally are persistent in sleep. Psychiatric comorbidities such as attention deficit hyperactivity disorder (ADHD), obsessive compulsive disorder (OCD) and others frequently are present.

Patients, families and teachers benefit from receiving instruction regarding the character of TS and its specific symptoms and from receiving counseling. Pharmacological treatment is not always necessary. Atypical antipsychotics (e.g. risperidone, ziprasidone, olanzapine, aripiprazole) are often the first-line treatment; typical antipsychotics (e.g. haloperidol, pimozide, fluphenazine), benzodiazepines (clonazepam) and tetrabenazine are employed less frequently. Alpha adrenergic agonists (clonidine, guanfacine), the selective noradrenaline re-uptake inhibitor, atomoxetine, and the amphetamine-like stimulant, methylphenidate, are useful in patients with tics and ADHD; selective serotonin re-uptake inhibitors can be useful in individuals with tics and OCD. Botulinum toxin can be effective in focal tics. In severe, treatment-resistant TS, deep brain stimulation may be beneficial.

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1. Introduction

Tourette syndrome (TS) is a common neuropsychiatric disorder, more common in males than females, with onset before age 18. TS is characterized by the presence of multiple motor and phonic tics persisting for at least 1 year. Psychiatric comorbidities, including attention deficit hyperactivity disorder, obsessive compulsive disorder and others, are frequently present. The characteristic clinical course is characterized by spontaneous fluctuations. Children with TS have normal intellect. However, their school attendance and personal life may be complicated by a variety of TS symptoms with a consequent negative effect on quality of life.

1.1. History

In 1885, the French neurologist Gilles de la Tourette, working in the Salpêtrière hospital in Paris under the supervision of Prof. Jean Martin Charcot, described in the journal Archives de Neurologie a neurological condition that he described as ‘maladie des tics.’ The article was based on the observation of nine individuals with features of childhood-onset, hereditability, waxing and waning severity of symptoms and premonitory urge.

2. Epidemiology of TS

The prevalence of TS is 0.3–0.8% of the pediatric population [1,2]. TS is more common in males than females (the ratio 4.3:1) [3]. The typical age of onset is between five and seven years; during
adolescence tics tend to decline and in approximately two thirds of individuals go into remission by early adulthood [3].

Psychiatric comorbidities are frequent (approximately 50–90% of TS patients), and include obsessive-compulsive disorder (OCD), attention deficit hyperactivity disorder (ADHD), impulse control disorder (ICD), self-injurious behavior, aggression, depression, anxiety and oppositional behavior [3,4]. Psychiatric symptoms, especially ADHD and OCD, tend to persist into or even increase in severity in adulthood. In approximately 20% of patients, symptoms have a negative effect on the social, educational and professional quality of life.

The clinical course of TS is one of waxing and waning, with long-lasting spontaneous fluctuations of both motor and vocal tics; it also may be characterized by the persistence of some subtypes of tics and the appearance and disappearance of new subtypes of tics.

In adulthood tics often disappear or decrease in intensity, but some mental symptoms, particularly OCD may get worse [1].

3. Clinical symptomatology

3.1. Tics

Tics are involuntary, abrupt, rapid, repetitive, but non-rhythmic movements or sounds (vocalizations). They are preceded by an inner urge (a mental phenomenon with a “have to do it” sensation). The urge may be a general feeling of tension and increasing intensity of energy to carry out certain activities. Another form of the urge can be local unpleasant, premonitory somatosensory sensations in a specific region of the body. Execution of the tic can be temporarily suppressed, but is then followed by increased rebound severity of the tic. Thus, tics could also be understood as voluntary reactions to involuntary stimuli. The performance of tics results in immediate but transient relief.

Tics usually appear first in the face, most typically with eye blinking; they subsequently may involve the head, neck, shoulders and proximal parts of the upper extremities. They appear many times during the day and display spontaneous fluctuations in frequency, type, severity, and complexity. They improve with concentration on activities, worsen during periods of relaxation or stress, and may persist in sleep. Many patients can suppress tics during social interactions following which, when alone, a burst of dramatic motor or vocal tics may appear.

3.1.1. Motor tics

**Simple motor** tics are the most common tics in the general population [5]. They involve one muscle or one segment of muscle. The most typical presentation involves eye blinking, jerks of head, tongue protrusion, and grimacing. Less frequent are jerks of the upper extremities, shrugging of the shoulders, and hand clapping. Simple motor tics may differ in velocity and character; they can be dystonic or myoclonic.

**Complex motor** tics are sequences or patterns of movement that may imitate purposeful or well-coordinated actions such as touching objects, repetitively fiddling with clothes or eyeglasses, jumping, hopping or bizarre gesturing. Specific subtypes are echopraxia (mimicking other people’s movements), copropraxia (obscene or socially inappropriate gestures), or rarely automatism (e.g. self-hitting, self-biting, banging objects).

3.1.2. Vocal tics

**Simple vocal** (or phonic) tics are various sounds such as sniffing, coughing, throat clearing, whispering or any simple meaningless noise.

**Complex vocal** (or phonic) tics are compulsive uttering of syllables, words or even short phrases. There are specific types of vocal tics, such as echolalia (repeating words spoken by someone else), coprolalia (obscene or socially inappropriate shouts) or palilalia (repeating the last words of one’s own speech). Coprolalia, although the best-known feature of TS in the lay community, is not frequent, occurring in 10–20% of patients.

3.2. Psychiatric comorbidities

**ADHD** is a mental disorder characterized by decreased ability to maintain attention and difficulty in controlling behavior, resulting in hyperactivity.

**OCD** is a mental disorder in which the individual feels the urge to perform repeatedly some activities (rituals), or suffers from recurrent intrusive thoughts. The patient is unable (or only transiently able) to control the urges or thoughts. Trying not to execute the ritual, may result in significant anxiety.

**ICD** is a psychiatric disorder characterized by exaggerated impulsivity and lack of self-control, with inability to resist an urge or impulse.

Darrow et al. [6] identified two phenotypes of TS: symmetry (checking obsessions, ordering, arranging, counting, writing, rewriting compulsions, repetitive writing tics) and disinhibition (uttering syllables or words, echolalia, palilalia, coprolalia, copropraxia, and obsessive urges).

3.2.1. Etiology

TS is a complex disorder with both genetic and environmental pathogenic factors.

Twin and family studies have established that TS bears a strong genetic component [2]. In many cases bilineal heredity is present; first-degree relatives are at significantly high risk for TS. Many studies based on linkage analysis, single nucleotide polymorphism, copy number variation, genome-wide association research, epigenetics studies and whole exome sequencing have been completed, but no single or multiple causative genes have been identified [7,8].

There is evidence that environmental factors connected with development of the embryo, with birth and with postnatal life may play a significant role in the development of TS [9] (e.g. maternal smoking during pregnancy, low birth weight, perinatal trauma). The connection between the onset and course of TS and streptococcal infection has been pursued for many years, but convincing evidence is lacking. The clinical entity, Pediatric autoimmune neuropsychiatric disorders associated with streptococcal infections (PANDAS), resembles TS in many features [10] (see Differential Diagnosis). However, the whole PANDAS concept remains in dispute and altered immune regulations are most probably not causal factors in TS.

3.2.2. Structural and functional correlates of TS

The precise localization of brain structures involved in TS pathogenesis remains unknown. Structural and functional neuroimaging studies demonstrate brain changes connected closely with TS [11]. The most significant neuropathological abnormalities are present in the gray matter – predominantly supplementary motor cortex, striatum (smaller volumes of putamen and caudate nucleus) and thalamus (in the whole cortico-striatal-thalamo-cortical circuit) [12,13]. Some studies have reported changes in the cerebellum [14] and others have emphasized cortical thinning (predominantly in sensorimotor cortex) as a primary pathological component [15]. A few fMRI studies have demonstrated altered functional connectivity in tracts connecting the supplementary motor areas with the basal ganglia and frontal cortico-cortical circuits [16].

Changes in many neurotransmitters may play a role in the pathogenesis of TS. There is evidence that dopamine is the primary candidate involved in TS [12], but other disturbances...
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