

Clinical phenomenology and phenotype variability in Tourette syndrome

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Abstract

Tourette syndrome (TS) is a neurodevelopmental disorder with a rich phenomenology that includes simple and complex motor and vocal tics as well as multiple comorbidities. From a nosological perspective, it is evident that a continuum of tic severity exists, of which TS is the most severe and rare form, while transient tics and chronic tics represent milder forms. From a psychopathology perspective, TS is often concurrent with obsessive–compulsive disorder (OCD) and attention deficit hyperactivity disorder (ADHD); these disorders appear to define TS “types” TS only,

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TS+OCD, and TS+OCD+ADHD. Additional clinical aspects of TS include more frequent than expected occurrence of anger episodes, anxiety disorders, mood disorders, impulse control disorders, learning disorders, and pervasive developmental disorders. Data reduction techniques have been used more recently to define a “simple” and “complex” tic symptom clusters or factors. Phenomenologic approaches can be used in TS to guide future pathophysiologic research.

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Introduction

Tourette syndrome (TS) is a neuropsychiatric disorder which originates from central nervous system insults, genetic or acquired, to specific brain circuits that subsume motor and emotional phenomena. We review the main phenomenological features of TS including the principal comorbidities as well as related disorders. The complexity of TS phenomenology was noted from the first descriptions of the disorder in the late 19th century and continue to be the subject of ongoing research. This review surveys recent literature that addresses TS phenomenology in order to further characterize useful phenotypes for etiological and clinical research.

Definitions

TS is clinically typified by the concurrent presence of multiple motor tics and at least one vocal tic occurring relatively uninterrupted at least for, and usually longer than, 12 months. Motor tics are topographically cephalocaudal with origin in facial and oral musculature, but also affecting neck, shoulder, arm, abdominal, and other single muscle groups. Complex motor tics can include whole body movements, skipping, jumping, touching movements, and others that involve multiple muscle groups [1]. Vocal tics are caused by repetitive forced air passage and can include sniffing, snorting, coughing, yelping, squealing, and other sounds including syllables or obscene words (coprolalia). As opposed to other motor movement disorders (e.g., irregular movements in chorea, brief twitching, or jerks in myoclonus), tic phenomenology is characterized by the repetitive and stereotypic nature of the movements. The movement is experienced as suppressible, suggestible and associated with a premonitory urge that is possible to delay but not postpone

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indefinitely. After a forced delay, tics can occur with greater intensity and frequency, having been likened to “an itch you have to scratch” in association with cognitive urges and a “just right” feeling [2]. Tics wax and wane over time, are most intense in prepubertal boys and in a large proportion reach a quiescent stage around early adolescence. TS is notably comorbid with multiple conditions. Most common in TS are obsessive–compulsive disorder (OCD), attention deficit hyperactivity disorder (ADHD), anxiety disorders, and major depression. Less common are impulse control disorders including skin picking and trichotillomania. In some instances, externalizing disorders such as oppositional behaviors, impulse control problems and bipolar spectrum disorders are notable. With lower specificity, learning disorders and some forms of speech delay can also be associated with TS.

The *DSM-IV TR* lists three primary tic disorders: Tourette’s Disorder (TD) herein referred to as TS, Chronic Motor or Vocal Tic Disorder (CMVT), and Transient Tic Disorder (TTD), as well as a category for nonspecific tics that cannot otherwise be classified, called “Tic Disorder Not Otherwise Specified” [3]. TS is the most clearly defined and perhaps the best studied of all the tic disorders. TS is defined as the presence of multiple motor and one or more vocal tics (not necessarily concurrent) with onset before age 18 years, occurring nearly every day consistently or intermittently for more than a year without more than a three-month period free of tics. CMVT is defined as chronic single or multiple motor or vocal tics, but not both, and has the same frequency, duration, and age of onset specifiers as TS. TTD is defined as the presence of single or multiple motor and/or vocal tics that last longer than 4 weeks but less than a year. TTD can occur as an isolated episode or in recurring bouts. The age of onset specifier is the same as for TS and CMVT, onset less than 18 years of age, although the severity and impairment is usually substantially less. From a clinical perspective, TS can consist primarily of motor and vocal tics exclusively (pure TS) or include complex phenomena such as coprolalia, pallilalia, echolalia, echopraxia (full-blown TS) or add comorbidities, most commonly OCD and ADHD (TS-plus) [4].

Among individual tic symptoms it is noteworthy to mention that eye blinking is a common and early motor tic in many patients, while throat clearing is a common vocal tic. As such, these movements or vocalizations are not too dissimilar from normal phenomena, with intensity, frequency, and the associated sensory-cognitive urge signaling differences from normative experiences. In this vein, the neural basis of tic movements has been conceptualized as the same as those that underlie the formation of habits or “coordinated ensembles of thought and action” [5]. Among the more complex tics, coprolalia was early noted to be characteristic, although not exclusive, of TS; it appears in only a minority of cases and is dependent on overall syndromic severity [6]. The overlap of TS and OCD symptoms through ritualistic and cognitively-laden motor phenomena is exemplified by touching behaviors, “evening

up” rituals and ordering needs. When these behaviors are cognitively laden with meaning, fears, and superstitions, they are considered in the province of OCD, when they are mostly purely motor they can be considered complex tics. Counting and repeating rituals often follow these motor phenomena, constituting a sensory-motor complex which TS and OCD often share.

Phenomenology of tic disorders: is there a spectrum?

In the early phenomenological study of TS, the notion that a wide range of disorders including alcohol abuse, substance abuse, impulse control disorders, ADHD, bipolar disorder, and others were etiologically related to TS was proposed [7]. Over time, the research data has supported the view that TS is a discrete disorder with a range of increasingly complex possible combinations of symptoms and comorbidities. In specialty clinics, up to 90% of TS patients will exhibit comorbidities [8,9]. Overall, the clinical phenomenology of TS points to a high level of arousal, which can take various clinic forms. Physiologically the TS-associated hyperarousal state is captured by sensorimotor gating deficits [10] and deficient cortical inhibition [11] and is possibly subsumed by cortical thinning in motor brain areas [12]. Clinically, in addition to tics this hyperarousal state can be variously expressed as anxiety [13], hyperactivity [14], mild-moderate self-injurious behaviors [15], and other clinical internalizing and externalizing behaviors. The relationship of TS with ADHD, in particular, has been the subject of multiple research studies [16,17] since it appears in over half of TS subjects and is associated with anger episodes and increased disability [18,19]. The relationship of TS with OCD is historically better delineated. Gilles de la Tourette [20] had described compulsive traits in the original report of patients with TS, mostly taking the form of motor and sensory-based ritualistic behaviors. While the etiological relationship between TS, OCD and ADHD appears to be complex, an approach that is gaining greater heuristic valence is of considering TS to be a complex disorder with different ‘classes’ or types. The class of TS only, without major comorbidities, is generally considered to be spared of major disability in relation to motor control and executive function [21,22], while the presence of ADHD concurrent with TS consistently confers greater cognitive and behavioral disability [23], but often, these studies do not consider OCD. When OCD is studied concurrently with TS and ADHD, three classes of TS appear with greater clarity: TS only, TS+OCD, and TS+OCD+ADHD [24]. The last comorbid class, TS+OCD+ADHD, appears to be more heritable and is associated with an earlier onset of tics [24]. Whether the three disorders share a common etiological (genetic or environmental) agent is yet to be determined; however, from a phenotypic perspective, it will be important to consider the range of TS phenotypes in future clinical, treatment, and physiological studies.

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