Tourette's Syndrome
An Often Misunderstood Disorder

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What is Tourette's Syndrome?

While it is tempting to objectify and intellectualize any neuropsychiatric disorder, Tourette's Syndrome (TS) often defies anything beyond a descriptive, verbal-picture, painted with an experienced brush and an empathetic mind. The incredibly broad spectrum of symptoms associated with TS is, alone, a testament to the unique and fascinating aspects of what could be characterized as a model neuropsychiatric disorder. This article is not intended as a comprehensive review of TS literature or as a definitive introduction to TS. This is meant as a brief overview of TS for those who might be somewhat unfamiliar with the disorder. The author's own experiences with TS, as a patient and a researcher, provide for what is hoped to be a refreshing insight into this idiosyncratic disorder.

Named after Georges Gilles de la Tourette, a French physician and pupil of Charcot, TS was also seen and described by other physicians. Sigmund Freud is thought to have seen the disorder in his practice but is said to have not really taken any notice of it. In his defense, even Charcot and Gilles de la Tourette conceded that tics and coprolalia could be symptoms of hysteria—Freud often diagnosed hysteria (Kushner, 1998). The disorder, as described by Gilles de la Tourette, is a pathological milieu in the sense that it creates a setting, a characterized mood, around itself. The disorder is, for all intents and purposes, one with the patient. Intertwined so that the person, if he/she were to be cured of TS, would cease to be themselves—almost like a personality disorder. But, relatively few diagnosed cases of TS actually fit this original description. Most people with TS have mild cases where a few facials tics and a couple of phonic tics might be their worse-case scenario. Echolalia, coprolalia, multiple motor and phonic tics, and motor incoordination are the key symptoms of Gilles de la Tourette's syndrome (Robertson, 2003). Coprolalia, though, is not common in most cases of TS and most TS cases involve just motor and phonic tics.

The current diagnostic criteria for TS are somewhat disputed. Two governing bodies for sets of criteria are often cited: the World Health Organization (WHO) and the American Psychological Association (APA). The Diagnostic and Statistical Manual of Mental Disorders (DSM) of the APA includes a maximum age of onset—18 years old in DSM-IV-TR. This version of the DSM, however, is controversial because, in addition to the presence of both motor and phonic tics, this edition of the DSM also stipulates that the patient must be experiencing impairment and distress in order for a diagnosis of TS. To some physicians, this seems as absurd as stating that a patient who deals well with their illness does not have the illness. For this reason, it is the DSM III-R criteria, which fail to make any mention of distress or impairment, which are most-often used for TS.

With an age of onset of about 7 years (Wand et al., 1992) and a male:female prevalence ratio of about 4:1 (Freeman et at., 2000), TS is most often seen in adolescent or preadolescent males. While TS can be very disturbing to children afflicted with the disorder, it is comforting to know that symptoms tend to disappear by adulthood in at least half of all cases (Robertson, 2003).

It is easy to get wrapped-up in an academic discussion of TS and much harder to have an intellectual appreciation for the complexities, idiosyncrasies, and spectrum of TS. For this reason, it is important that a clinician or researcher understands TS, as best she/he can, from the patient's perspective.

While most individuals with TS tend to have mild symptoms that do not usually interfere with daily activity, both tics and behavioral problems, such as aggression, can cause social problems in individuals with TS. Tics such as head jerking, unusual vocal noises, and bouts of coprolalia can be deterrents to normal socialization in a child with TS. In adulthood, these, and similar symptoms, can make it hard for an individual with TS to keep a job, make friends, and have normal relationships.

The tics, from head jerking to seemingly deliberate profanities are, to the patient, much like an itch that needs to be scratched. When a person with TS jerks their head or blinks their eyes it is not a movement which they have no control over (as in Parkinson's

Disease or some other tic disorders) rather it is a response to an intense urge, a need that must be satisfied. The person has to jerk their head or blink their eyes – to not do so feels incredibly wrong. Likewise, the profanity is also the result of responding to a need, an intense urge. The person may not mean to call you a bad name, she or he simply has to.

I have heard parents of children with TS remark that their child is "doing things on purpose." In one case I saw, a 9 year old girl would often call her mother a "f*cking-b*tch." Her mother was adamant that her daughter was doing this on purpose, had full control over herself, and that this offensive profanity had nothing to do with the girl having TS. In fact, neither the mother nor the father was convinced that their daughter actually had TS. They believed that she was simply a "naughty-girl" and so they would punish her, or rather try to punish her, by sending her to her room or refusing her requests for toys. Of course, the punishments did not work because the girl was not doing these things on purpose. She did not want to call her mother such horrible names, she had no choice, she had to, it was a tic. Imagine the torment that this little-girl must be feeling. This would be difficult enough for an adult but even more so for a child. Confused and bewildered by her own behavior, she needs help, not punishment.

Unfortunately, the girl's story is all too common. TS was once thought to be an extremely rare disorder and it is only in the past decade that it has gained any notoriety outside of the medical community. Indeed, even within the medical community knowledge of TS is growing but scarce.

Another reason why TS is often hard for the clinician to recognize is that comorbid disorders such as Attention Deficit Hyperactivity Disorder (ADHD) and Obsessive Compulsive Disorder (OCD) can make it difficult to see where the TS begins and where it ends. Indeed, one TS researcher asserts that TS begins where ADHD, OCD, tics, and behavioral problems meet (Jankovic, 2001). This brought much uproar in the form of letters to the editor as TS has always been identified as a tic disorder with none of the aforementioned disorders or problems required for a TS diagnosis. However, it is entirely possible that they have a common etiology.

Pharmacology and Current Research Foci

Most studies suggest that abnormalities in dopamine and serotonin metabolism are found in TS patients. However, just what those disturbances are is not known

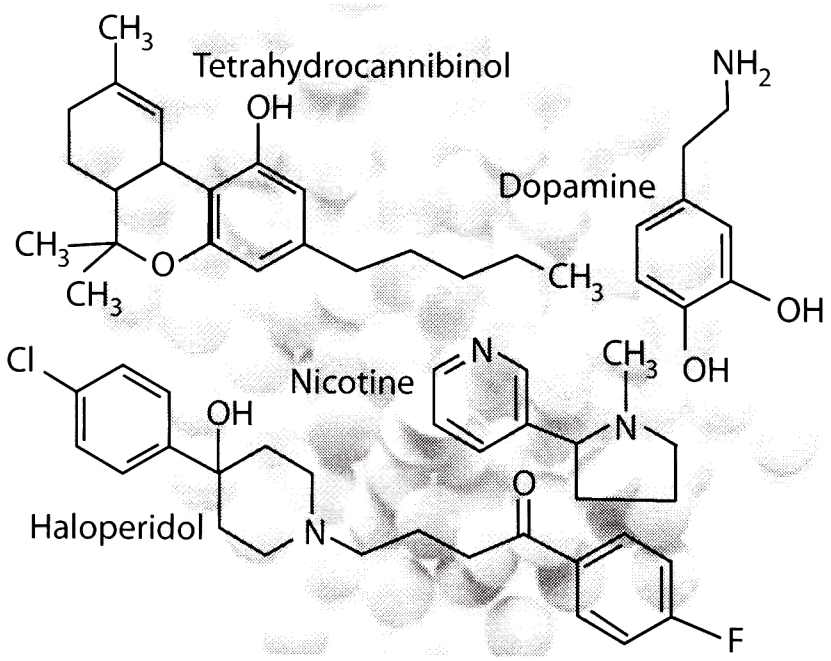

Figure 1
Haloperidol, dopamine, tetrahydrocannibol and nicotine have all been used with variable success to treat Tourette's Syndrome.

and the strongest evidence towards any answers in this direction comes from the use of haloperidol in the treatment of TS.

Pharmacological treatment for TS started in the early 1960s with the use of haloperidol, a dopamine receptor antagonist (Caprini, 1961 & Seignot, 1961). Relatively small amounts of haloperidol are needed to achieve saturation of the D2 receptors and most TS patients do respond to it (Fitzgerald et al., 2000). Clinical practise, though, has moved towards using atypical dopamine antagonists such as risperidone and olanzapine (Sandor, 2003). In general, dopamine receptor antagonists have been the most successful class of pharmacological agent used for the treatment of TS as they are able to reduce tic frequency and severity in about 70% of cases (Shapiro and Shapiro, 1998).

Other treatments, including nicotine and tetrahydrocannabinol (marijuana) have been tried with variable success. Early nicotine studies were inefficient and the most promising study suggests that nicotine is only helpful when combined with other treatments, such as haloperidol (Silver et al., 2001).

In addition to the many clinical trials of dopamine receptor antagonists and other pharmacological interventions, there are 2 main foci in current TS research: genetics studies and brain imaging studies.

TS has, for a long time, been seen as a hereditary disorder, running in families and passed-on from parent to child. Thus, there are many international efforts to locate the gene(s) responsible (or at least partly responsible) for TS. Since 1990, 6 family studies have been completed, all of which show TS is familial and genetic linkage studies have provided enough data to suggest possible candidate genes for TS. Unfortunately, most of the studies examining candidate loci have had negative results. The common thread among potential candidate genes which have been excluded has been their relation to dopamine metabolism with almost all of them regulating the release of dopamine or serotonin in one way or another (Pauls, 2003). Despite the negative results, segregation analysis studies have reported data sufficient to suggest that TS is due to an autosomal dominant gene. In line with most of the TS genetic work however, is that this gene has remained elusive and, if it does exist, the penetrance and variability will be an extremely interesting matter for study.

Conclusion

Gilles de la Tourette's syndrome has gone from being a supposedly rare 19th century tic disorder to being a more common neuropsychiatric spectrum disorder linked to OCD and ADHD. The psychopharmacological revolution offered the first ever effective treatment for TS, as it did for a myriad of other neuropsychiatric disorders, and the ensuing genetics revolution allowed the first-ever window into the root cause of TS, or at least we think. Future genetics studies will likely yield promising candidate genes and, as we work out the genetic and biochemical pathways of the human organism, these studies will probably lead to a different class of pharmacological treatment for TS – drugs that can directly alter the chemical imbalance of TS.

As for the patients, those who have to suffer because of their TS, those who fear going to a movie theatre so as not to disturb the peace, those who cannot work a job at the grocery store because they constantly tell Mrs. Smith and her daughter to "f*ck-off and have a nice day," it is for these people that we must continue to work, learn as much as we can, and attempt to ask questions that nobody knows the answer to. M