

**lek. Justyna Kaczyńska**

## **Streszczenie w języku angielskim**

### **Title: Atypical tics in Gilles de la Tourette syndrome**

Gilles de la Tourette syndrome (GTS) is a neurodevelopmental disorder that begins in childhood and predominantly presents with tics. Tics are defined as sudden, rapid, recurrent, nonrhythmic motor movements (motor tics) or vocalizations (vocal tics). Classic clonic tics fulfil the criteria of the above definition, however, the clinical picture of the disease also includes atypical types of tics. These include cognitive tics, dystonic tics, as well as tonic tics (TTs) and blocking tics (BTs) which are the subject of this dissertation. TTs are sustained (> 500ms) isometric muscle contractions that produce little or no noticeable movement. BTs are defined as sudden and transient cessations of motor activity with maintained consciousness. Despite the fact that TTs and BTs are known elements of the clinical picture of GTS, there is little information about them in medical literature. The publications that are the subject of this dissertation are the first to analyze the clinical characteristics of TTs and BTs and investigate the correlations between these atypical tics and other clinical factors.

Tics have certain characteristics that allow them to be differentiated from other movement disorders. Most patients experience a non-specific sensory phenomenon called premonitory urges that occur immediately before the tic. The performance of the tic causes an immediate, short-term dissolution of unpleasant warning signals, producing a feeling of relief. Other characteristic features of tics include the short-term possibility of refraining from performing them and the variability of tics over time (waxing and waning course). More than 85% of patients with GTS have a coexistence of psychiatric disorders, the most common of which are obsessive-compulsive disorder (OCD) and attention deficit hyperactivity disorder (ADHD).

Publication 1 focused on TTs. The study included 241 patients (153 children and 88 adults; mean age  $16.3 \pm SD 9.2$  years; 188 males - 78.0%). In publication 2, which pertained to BTs, the number of patients enrolled was 195 (140 children and 55 adults; mean age  $15.0 \pm SD 9.2$  years, 148 males - 75.9%). Both publications are original articles whose common aims were to determine the incidence, onset, and clinical characteristics of TTs and BTs, to investigate

their correlation with other factors, and to determine whether they belong to the tics spectrum or other disorders in GTS.

All patients were systematically interviewed with the aid of a semi-structured interview comprised of demographic and clinical data and a physical examination was performed. The collected data was statistically analyzed.

The obtained results confirmed the initial assumption that TTs and BTs have many common features. Both types of tics appear in patients with GTS who are on average a little over 10 years old, which is the period of the greatest severity of tics in the natural course of the disease. The statistical analysis showed a significant correlation of TTs and BTs with a greater number and severity of tics, i.e. with a more severe course of GTS. It has also been shown that TTs and BTs have similar features to classic tics: variable occurrence (possibility of resolution) during the course of the disease, and in the case of TTs the presence of premonitory urges and the feeling of relief after the performance of the tic. Both TTs and BTs may appear singly, but it is also possible to have several types of these tics in one patient.

The presence of at least one TT at some point in the disease was documented in 72% of patients (85.2% of adults and 63.9% of children), and at least one BT occurred in 37.4% of patients (52.7% of adults and 31.4% of children). BTs appear to be more involuntary because, unlike TTs, they are not associated with the presence of premonitory urges.

Publication 2 emphasizes the division of BTs into primary and secondary. Primary BTs are defined as a sudden cessation of movement that is not caused by other motor tics or vocalizations, while secondary BTs are the result of other tics, usually prolonged TTs or dystonic tics. The study also emphasized the need to differentiate between vocal BTs and stuttering. Publication 1 concluded that there are numerous similarities between TTs and dystonic tics in terms of phenomenology and clinical correlations, and there is often a coexistence between these two types of tics.

The most common types of TTs were tensing of the abdomen (58.7%), slightly less common was tensing of the neck muscles (52.7%) and tensing of the upper limbs (50.3%), and least frequent were contractions of the muscles of the lower limbs, tensing of the muscles of the whole body, or buttocks tensing. Cessation of walking was the most commonly found BT (80.8%), while cessation of speech and cessation of running or writing were diagnosed less frequently.

Although the analysis of selected therapeutic procedures and the effectiveness of the treatment of atypical types of tics was not the subject of this doctoral dissertation, the confirmed affiliation of TTs and BTs to the spectrum of tics suggests that the treatment of these tics should be the same as for classic tics.

In conclusion, the presented publications characterized TTs and BTs, and the obtained results indicate that these two types of tics have many common features. Both TTs and BTs have been shown to be early and frequent symptoms of GTS, and TTs are twice as common as BTs. The frequency of both types of tics increases throughout the duration of the disease. Unlike TTs, which are the result of involuntary muscle contractions, BTs have been shown to disrupt various motor activities. An important conclusion from both publications is the occurrence of the analyzed tics in the period of the highest intensity of the disease and their relationship with a more severe course of GTS. The studies have confirmed that TTs and BTs belong to the spectrum of tics and comorbid psychiatric disorders in patients do not significantly affect their occurrence.

In patients with GTS, it is extremely important to be aware of the presence of all types of symptoms and actively search for them as this allows for a proper assessment of the severity of the disease and, thus, to make the right therapeutic decisions.