Gilles de la Tourette Syndrome - A Severe Case in an 8 Year-Old Nigerian Male

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Abstract

Living with severe Gilles de la Tourette syndrome is physically and psychologically distressing for both the child and family and can impair quality of life and school attendance. Tourette syndrome is a chronic neurodevelopmental motor disorder manifesting with multiple motor and vocal tics. Tic disorders are common in school-aged children but there are few reports in Nigerian children and there is a dearth of physicians experienced in treating Tourette syndrome. This case report highlights the diagnosis and frustrations of management of severe Tourette syndrome in an 8 year-old male. More awareness of this neurodevelopmental disorder is needed in our locality.

Keywords: Tourette Syndrome; Tic Disorder; Management; Antipsychotics; Pimozide

Introduction

Gilles de la Tourette syndrome (GTS) is a tic disorder with onset in childhood characterized by occurrence of multiple motor and vocal tics that wax and wane in frequency, lasting more than one year and not due to a medication/substance or another medical condition [1,2]. It remains the most complex and the most severe of the three tic disorders since it presents with more frequent, more complex, more severe motor and vocal tics and a greater number and severity of co-occurring neuropsychiatric conditions [1,3]. It is common among school-aged children, commoner in boys and occurs worldwide with a prevalence of 0.6 - 0.9% [4,5].

Tics are sudden, rapid, recurrent, stereotyped, non-rhythmic motor movements or vocalizations [2]. There are three tic disorders (provisional tic disorder, persistent/chronic motor/vocal tic disorder and Tourette disorder/syndrome) which are distinguished by type(s) of tics present and the duration of symptoms [1-3]. The exact cause of GTS and other tic disorders is unknown

but like other neurodevelopmental disorders, multiple factors including genetic and environmental factors have been implicated [2,6]. Tourette disorder occurs more frequently among first-degree relatives which suggests a genetic component and the interplay between acute or chronic infections and other environmental factors in causing dopaminergic, serotonergic and glutamatergic neurotransmitter dysfunction and immunologic dysregulation has been reported by an earlier study [6]. Infections commonly associated with symptoms of tics are those caused by group A beta hemolytic *Streptococcus* (GAS), *Mycoplasma pneumoniae* and *Borrelia burgdorferi* [6]. Following streptococcal infections such as tonsillopharyngitis, the occurrence of tics, obsessive-compulsive disorder and increased motor hyperactivity or reduced fine motor coordination in a pre-pubertal child is referred to as Paediatric Autoimmune Neuropsychiatric Disorders Associated with Streptococcal infections (PANDAS) [2,6]. Thus, PANDAS is a differential diagnosis of post streptococcal tics or acute exacerbation of tics following streptococcal pharyngitis or pyoderma.

The diagnosis of GTS is clinical and based on the occurrence of both motor and vocal tics and fulfilling the criteria stipulated in the DSM-5 as already stated above [1]. Laboratory tests and neuroimaging are therefore not needed to confirm classical presentations but may sometimes be employed to exclude secondary causes of tics such as post viral encephalitis, Huntington's disease, stroke and insult to basal ganglia following head trauma [2,8]. Comprehensive evaluation of GTS involves an assessment of severity and evaluation for co-morbid disorders [2,9]. Useful symptom-rating scales include: Parent Tic Questionnaire, Tourettes's Disorder Scale, Tourette syndrome severity scale (TSSS) and Yale Global Tic Severity Scale (YGTSS) with the latter being the most commonly used instrument for clinician-rated tic severity [2]. Co-occurring psychiatric disorders include: Attention Deficit Hyperactivity Disorder (ADHD), Obsessive Compulsive disorder (OCD), mood disorders, Autism spectrum disorders (ASD), oppositional defiant disorder (ODD) and conduct disorders [2,9]. Management of GTS of mild severity causing minimal, mild or no impairment or distress involves only psychoeducation of patient, family members and teachers [2]. Severe GTS causing moderate, severe or marked functional impairment and distress would require additional measures such as class room accommodation or formal individualized education plan (IEP), behavioural treatment and pharmacotherapy [2]. The natural history of tic disorders is that of typical onset at 4 - 6 years of age, peak severity between ages 10 and 12 years and marked reduction of tics in two-thirds of individuals at age 18 - 20 years [2].

Case Presentation

Patient C.U, an 8 year-old male who was referred to the Child Neurology Clinic of Mother Healthcare Hospital and Diagnostics, Owerri due to complaints of involuntary body movements x 7 months and involuntary vocalizations x 2 months - all prior to presentation 3 years 5 months ago. Involuntary movements were recurrent, sudden, rapid, purposeless jerks of parts of the body. The abnormal movements initially involved the face (facial twitching) then neck turning and shoulder shrugging and later progressed to a sequential jerking of the upper limbs. Other times, he frequently had eye blinking, eye darting, sudden backward jerking of his head, sudden arm flexion and extension and sudden forceful leg kicking.

These abnormal movements were exacerbated by stress, fatigue or tiredness or by being idle or unengaged. They were severe and disrupted his school activities. He tore his books while writing and distracted other pupils who frequently laughed at him and made him feel very uncomfortable. On some occasions, he had sustained bruises by hitting his hand/elbow on the wall or his mouth. Due to these, he was no longer willing to go to school. However, these movements usually abated during sleep.

About 5 months into the illness, he started making involuntary vocalizations that sounded like animal noises. He would have bouts of unnecessary coughing, throat clearing, sniffing and utterance of barking noises. Frequently, he repeated what other people said, copied other people's gestures and had sudden changes in volume or pitch of speech. Later he started uttering obscene words and made complex vocal combinations.

There were no symptoms of inattention in class or hyperactivity/impulsivity and no history of obsessions or compulsions. There was no history of ingestion of substances such as cocaine and he was not on any medications prior to onset of above symptoms. Pregnancy, birth and early post natal history was unremarkable. There was no family history of tics, epilepsy or psychiatric disorders. His school performance was good. For the above complaints, he was seen by a medical officer who requested for brain MRI and planned to refer him to a psychiatrist. However, mother was verbally referred by a resident Paediatrician to our Child Neurology clinic.

The findings on physical examination at presentation were a fully conscious and alert boy with frequent involuntary, rapid, arrhythmic jerks of the limbs and vocalizations of animal-like noises. Initial diagnosis was provisional tic disorder since the duration of symptoms was less than one year. Initial management was non-pharmacologic as they were giving information about the diagnosis, its waxing and waning course, that it was not a psychotic illness or "madness", that it can resolve with age without medications and also the likelihood of the multiple motor and phonic tics persisting beyond a year which will lead to a formal diagnosis of Tourette syndrome. The parents were reassured and counseled to largely ignore the tics and increase engagement in exercises. The class

teacher was also to be educated on the child’s diagnosis so that the child will be better accommodated in class with less stigmatization.

Two weeks later, the child presented again to the clinic with complaints of increasing disruptive movements and severe tics that were disrupting both school and social activities. He was frequenting tearing his books in class while copying notes. He was further assessed and a formal assessment of tic severity was done using Yale Global Tic Severity Scale (YGTSS) from the Yale Child Study Center (October 1992 version) and the Parent Tic Questionnaire (PTQ). Screening for co-morbid conditions was done using the Vanderbilt ADHD Diagnostic Parent and Teacher rating scales. The following investigations were requested for: Full blood count (FBC), Liver Function test (LFT), serum electrolytes, urea and creatinine (SEUC), urinalysis, Thyroid function tests (T₃, T₄, TSH), throat swab for m/c/s, ASO titre and serum ferritin.

Assessment with YGTSS showed motor tic severity of 16/25, vocal tic severity of 16/25 and impairment of 30/50 (moderate) giving a total YGTSS of 62/100 (Figure 1). The Parent Tic Questionnaire (PTQ) indicated that he had 9 out of the 14 listed motor tics with many of them occurring hourly or daily with an intensity of 4 - 6/8 (obvious to others {≥ 4}, very noticeable or painful {≥ 6}) (Figure 2). Using the Vanderbilt ADHD Diagnostic Parent and Teacher rating scales, he did not fulfill the criteria for the diagnosis of ADHD and screened negative for Anxiety or depressive symptoms. However, by the Vanderbilt ADHD parent rating scale (but not the Teacher’s), he fulfilled the criteria for Oppositional defiant and conduct disorders while the performance section indicated no impairment (scored average or above average on all items).

Owing to the severity of his tics and unavailability of personnel trained in Comprehensive Behavioural Intervention for Tics (C-BIT), he was commenced on tabs pimozide 1 mg daily after a prior ECG was reported by the Cardiologist as normal. Other requested investigations were not done due to financial constraints. Psycho-education was further provided and the child taught how to build a competing response to tics when a premonitory urge is felt. On follow-up two weeks later, everybody was happy that the tics significantly reduced and he was attending school without tearing his books. About 6 weeks after commencement of tabs Pimozide, he

NAME: Christian Ogoke TODAY'S DATE: 20/06/18

RATER: Dr Christian Ogoke

MOTOR TIC SYMPTOM CHECKLIST

Description of Motor Tic Symptoms. Motor tics usually begin in childhood and are characterized by sudden jerks or movements, such as forceful eye blinking or a rapid head jerk to one side or the other. The same tics seem to recur in bouts during the day and are worse during periods of fatigue and/or stress. Many tics occur without warning and may not even be noticed by the person doing them. Others are preceded by a subtle urge that is difficult to describe (some liken it to the urge to scratch an itch). In many cases it is possible to voluntarily hold back the tics for brief periods of time. Although any part of the body may be affected, the face, head, neck, and shoulders are the most common areas involved. Over periods of weeks to months, motor tics wax and wane and old tics may be replaced by totally new ones.

Simple motor tics can be described as a sudden, brief, "meaningless" movement that recurs in bouts (such as excessive eye blinking or squinting). Complex motor tics are sudden, stereotyped (i.e., always done in the same manner) semi-purposeful (i.e., the movement may resemble a meaningful act, but is usually involuntary and not related to what is occurring at the time) movements that involve more than one muscle group. There may often be a constellation of movements such as facial grimacing together with body movements. Some complex tics may be misunderstood by other people (i.e., as if you were struggling to say "I don't know"). Complex tics can be difficult to distinguish from compulsions; however, it is unusual to see complex tics in the absence of simple ones. Often there is a tendency to explain away the tics with elaborate explanations (e.g., "I have hay fever that has persisted" even though it is not the right time of year). Tics are usually at their worst in childhood and may virtually disappear by early adulthood, so if you are completing this form for yourself, it may be helpful to talk to your parents, an older sibling, or a relative, as you answer the following questions.

• Age of first motor tics? 7 years old

• Describe first motor tic: sudden forceful jerk/kick of the leg.

• Was tic onset sudden or gradual? gradual

• Age of worst motor tics? 8 yrs years old

Motor Tic Symptom Checklist

In the boxes on the left below, please check with a mark (x) the tics the patient

1) has **EVER** experienced

2) is **CURRENTLY** experiencing (during the past week)

State **AGE OF ONSET** (in years) if patient has had that behavior.

Also, in the tic descriptions below, please **circle or underline** the specific tics that the patient has experienced (circle or underline the words that apply).

2

Figure 1

required up to 2 mg daily to achieve good control especially during his examinations.

Patient defaulted with follow-ups but presented to the clinic after 6 months with complaints of exacerbation of both motor and vocal tics and painful swallowing. Patient had run out of Pimozide and the drug was no longer available in the whole country. Examination revealed significant discrete, multiple bilateral nontender cervical lymphadenopathy, erythematous pharynx and mildly enlarged tonsils bilaterally. Diagnoses of acute tonsillopharyngitis and tic exacerbation (flare-up) were made with PANDAS as differential diagnosis. Requested investigations were throat swab m/c/s and Full blood count. The following prescription was made: Tab Augmentin 457 mg BD x 10/7; to start tab risperidone 0.125 mg

Figure 2

BD and increase to 0.25 mg BD. Throat swab m/c/s isolated *Candida albicans* - scanty growth.

He was seen a month later with complaints of increasing severity and frequency of both motor and phonic tics despite increasing tab risperidone to 1.5 mg BD. Writing exams was severely impaired and he was noticed to have occasional involuntary smile which was a new tic. After a month of poor response to tab risperidone, medication was changed to tabs haloperidol 1.25 mg BD. With poor response, the dose of haloperidol was subsequently increased to 2.5 mg BD. Despite this multiple complex motor and vocal tics persisted and made it difficult for the child to go out the house. After some months, out of frustration, mother stopped all drugs and resorted to “fasting and prayers” and attending church deliverance services. Patient defaulted and was seen again a year later during a tic “off period”. He was said to have had a 2-month period of exacerbation which was the longest since onset of illness. Exacerbations were

also noted to be more during the rainy season. On examination, he was neurologically normal with no noticeable motor or vocal tics. Diagnosis then was Tourette syndrome - off period. Patient was to try tab Aripiprazole 1.25 mg BD (to increase to 2.5 mg if necessary) if symptoms resurfaced. After this, he was lost to follow-up for two years. During this period, he consulted several other physicians in different parts of Nigeria due to persisting symptoms and poor response to available drugs. However, after a long search, he was able to procure tabs pimozide again. This was again efficacious and he continued it until he had oculogyric crisis and chest pain necessitating discontinuation of the drug. Presently, at 10 years of age,

Figure 3

FREQUENCY: Constant, almost all the time during the day
Hourly, at least once per hour
Daily, at least several times per day
Weekly, just a few times or less

INTENSITY: Mild: 1-2; Obvious to others: 4 or higher; Very noticeable or painful: 6 or higher.

VOCAL TICS	PRESENT		FREQUENCY C H D W 4 3 2 1	INTENSITY 0-8
	YES	NO		
Grunting	YES		2	4
Sniffing	YES		1	4
Snorting	YES		1	4
Coughing	YES		2	6
Animal noises	YES		3	6
Syllables				
Words				
Phrases				
Echolalia (repeating vocalizations of others)	YES		1	4
Coprolalia (obscene words)	YES		2	4
Blocking/stuttering				
Other				
Other vocal tics				
Complex vocal combinations (multiple tics at once)	YES		2	5

Figure 4

55
Vanderbilt ADHD Diagnostic Parent Rating Scale
Page 2 of 3

25. Is angry or resentful 0 1 2 3
26. Is spiteful and vindictive 0 1 2 3
27. Bullies, threatens, or intimidates others 0 1 2 3
28. Initiates physical fights 0 1 2 3
29. Lies to obtain goods for favors or to avoid obligations (i.e., "cons" others) 0 1 2 3
30. Is truant from school (skips school) without permission 0 1 2 3
31. Is physically cruel to people 0 1 2 3
32. Has stolen items of nontrivial value 0 1 2 3
33. Deliberately destroys others' property 0 1 2 3
34. Has used a weapon that can cause serious harm (bat, knife, brick, gun) 0 1 2 3
35. Is physically cruel to animals 0 1 2 3
36. Has deliberately set fires to cause damage 0 1 2 3
37. Has broken into someone else's home, business, or car 0 1 2 3
38. Has stayed out at night without permission 0 1 2 3
39. Has run away from home overnight 0 1 2 3
40. Has forced someone into sexual activity 0 1 2 3
41. Is fearful, anxious, or worried 0 1 2 3
42. Is afraid to try new things for fear of making mistakes 0 1 2 3
43. Feels worthless or inferior 0 1 2 3
44. Blames self for problems, feels guilty 0 1 2 3
45. Feels lonely, unwanted, or unloved; complains that "no one loves him/her" 0 1 2 3
46. Is sad, unhappy, or depressed 0 1 2 3
47. Is self-conscious or easily embarrassed 0 1 2 3

Figure 6

Vanderbilt ADHD Diagnostic Parent Rating Scale
Page 1 of 3

VANDERBILT ADHD DIAGNOSTIC PARENT RATING SCALE

Child's Name: [Redacted] Today's Date: 23rd - 07 - 2018
Date of Birth: NOV 17th 2009 Age: 8 years
Grade: [Redacted]

Each rating should be considered in the context of what is appropriate for the age of your child.
Frequency Code: 0 = Never 1 = Occasionally 2 = Often 3 = Very Often

1. Does not pay attention to details or makes careless mistakes, for example homework 0 1 2 3
2. Has difficulty sustaining attention to tasks or activities 0 1 2 3
3. Does not seem to listen when spoken to directly 0 1 2 3
4. Does not follow through on instructions and fails to finish schoolwork (not due to oppositional behavior or failure to understand) 0 1 2 3
5. Has difficulty organizing tasks and activities 0 1 2 3
6. Avoids, dislikes, or is reluctant to engage in tasks that require sustained mental effort 0 1 2 3
7. Loses things necessary for tasks or activities (school assignments, pencils or books) 0 1 2 3
8. Is easily distracted by extraneous stimuli 0 1 2 3
9. Is forgetful in daily activities 0 1 2 3
10. Fidgets with hands or feet or squirms in seat 0 1 2 3
11. Leaves seat when remaining seated is expected 0 1 2 3
12. Runs about or climbs excessively in situations when remaining seated is expected 0 1 2 3
13. Has difficulty playing or engaging in leisure/play activities quietly 0 1 2 3
14. Is "on the go" or often acts as if "driven by a motor" 0 1 2 3
15. Talks too much 0 1 2 3
16. Blurts out answers before questions have been completed 0 1 2 3
17. Has difficulty waiting his/her turn 0 1 2 3
18. Interrupts or intrudes on others (e.g., butts into conversations or games) 0 1 2 3
19. Argues with adults 0 1 2 3
20. Loses temper 0 1 2 3
21. Actively defies or refuses to comply with adults' requests or rules 0 1 2 3
22. Deliberately annoys people 0 1 2 3
23. Blames others for his or her mistakes or misbehaviors 0 1 2 3
24. Is touchy or easily annoyed by others 0 1 2 3

Figure 5

BRIGHT FUTURES TOOL FOR PROFESSIONALS

Vanderbilt ADHD Diagnostic Teacher Rating Scale

Name: [Redacted] Grade: Primary 6
Date of Birth: 17/11/2009 Teacher: [Redacted] School: Albright New Pri School Amakoma

Each rating should be considered in the context of what is appropriate for the age of the children you are rating.
Frequency Code: 0 = Never; 1 = Occasionally; 2 = Often; 3 = Very Often

1. Fails to give attention to details or makes careless mistakes in schoolwork 0 1 2 3
2. Has difficulty sustaining attention to tasks or activities 0 1 2 3
3. Does not seem to listen when spoken to directly 0 1 2 3
4. Does not follow through on instruction and fails to finish schoolwork (not due to oppositional behavior or failure to understand) 0 1 2 3
5. Has difficulty organizing tasks and activities 0 1 2 3
6. Avoids, dislikes, or is reluctant to engage in tasks that require sustained mental effort 0 1 2 3
7. Loses things necessary for tasks or activities (school assignments, pencils, or books) 0 1 2 3
8. Is easily distracted by extraneous stimuli 0 1 2 3
9. Is forgetful in daily activities 0 1 2 3
10. Fidgets with hands or feet or squirms in seat 0 1 2 3
11. Leaves seat in classroom or in other situations in which remaining seated is expected 0 1 2 3
12. Runs about or climbs excessively in situations in which remaining seated is expected 0 1 2 3
13. Has difficulty playing or engaging in leisure activities quietly 0 1 2 3
14. Is "on the go" or often acts as if "driven by a motor" 0 1 2 3
15. Talks excessively 0 1 2 3
16. Blurts out answers before questions have been completed 0 1 2 3
17. Has difficulty waiting in line 0 1 2 3
18. Interrupts or intrudes on others (e.g., butts into conversations or games) 0 1 2 3
19. Loses temper 0 1 2 3

(continued on next page)

Figure 7

Figure 8

he is no longer on any drugs. Vocal tics have completely stopped while motor tics are still occurring but milder with much longer off-periods of up to 6 months.

Discussion

The case presented fulfilled the criteria stipulated by DSM-5 for diagnosis of Tourette syndrome [1]. Objective assessment of severity using two severity assessment scales for tics (one parent-rated scale - PTQ and one clinician-rated - YGTSS) classified this case as causing moderately severe functional impairment. The YGTSS is the most commonly used Tourette syndrome severity assessment scale and its reliability and validity have been reported by Murphy, *et al.* [10]. Evaluation for co-occurring psychiatric disorders is critical in patients with Tourette syndrome since these disorders may cause

Figure 9

more functional impairment than the tics [2,9]. For co-morbidity assessment, the Vanderbilt ADHD Diagnostic Rating scale was used and this child did not have ADHD, anxiety or depression but screened positive for Oppositional Defiant and Conduct disorders. This child was not comprehensively evaluated for co-morbidities due to poor compliance with follow-up and financial constraints; the latter being the reason for failure to do most of the requested investigations. Multidisciplinary evaluation especially by a psychiatrist would have identified more co-morbidities in this child with moderate - severe Tourette syndrome.

A number of factors made the management of this child challenging: the poor health knowledge of people in our locality made psychoeducation difficult, the multiple and long defaults with follow-up clinic visits owing to a combination of factors including

out-of-pocket payment for health services and the poor response to many neuroleptics that culminated in reversion to unorthodox medicare and spiritual healing. Additionally, the family had unrealistic expectations from pharmacotherapy despite few sessions of psychoeducation at the beginning. Furthermore, the lack of social support groups for children with Tourette syndrome in Nigeria denied this family of some emotional support and a feeling of “not being alone” that would have made them more patient with the clinician. The results of these for this child and his family were stigmatization, social isolation during flare-ups, physical and emotional distress, “doctor shopping” and interference with schooling and impaired quality of life.

It is clear to the neurologist that children with tics have variable response to different drugs and the aim of pharmacotherapy is to reduce the tics to a level where they are no longer causing functional impairment and not to totally eliminate them [2]. Failure to appreciate this could have resulted in the family having unrealistic expectations. However, the plight of a family with a child with multiple complex motor tics, echopraxia, echolalia and coprolalia is understandable. More so, the unavailability of the only efficacious drug, in this case - pimozide and the oculogyric crisis that necessitated its immediate cessation added to their frustration.

The exacerbation of tics in this child occurred mostly in the rainy season and coincides with the period of increased upper respiratory tract infections such as GAS - pharyngitis which is known to trigger or exacerbate tics [6]. It is surprising how this child with clinical features of acute tonsillopharyngitis during one of the tic flare-ups had a throat swab culture that yielded scanty growth of *Candida albicans* which is not a known common cause of sore throat. This may have been a contaminant. Research may also be needed in investigating the role of *P. falciparum* malaria in exacerbating tic symptoms since in our locality there is also a surge in the incidence of malaria during the rainy season. Finally, the frequency and severity of tics in this child have dwindled three years after onset and is expected to disappear in late adolescence. This conforms to the known natural history of Tourette syndrome [2].

Conclusion

This case report creates more awareness and highlights the dis-

tress of living with GTS and challenges of management of moderate-to-severe Tourette syndrome in a resource-poor setting.

Competing Interests

The authors declare no competing interests.

Authors' Contributions

Dr Christian C. Ogoke was involved in the evaluation, diagnosis and management of this patient. He conceived, designed and drafted this report. Dr Emeka C. Nwolisa revised the article critically for important intellectual content. Both authors read and approved the final manuscript.

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