Tourette's Syndrome, OCD and ADHD as a Triad in Neurofibromatosis Type 1: A Case Report

To the editor,

Neurofibromatosis type 1 (NF1) is a group of distinct genetic disorders caused by a mutation of the neurofibromin (NF) gene on chromosome 17 [1] and is commonly associated with the appearance of nerve tumors (neurofibromas). NF1 has co-occurring deficits in cognitive domains such as attentional, executive, language, and visuospatial in approximately 80% of patients,[2] ultimately leading to academic difficulties. Other psychiatric comorbidities are dysthymia (21%), depressive disorder (7%), anxiety disorders (1%–6%), and personality disorders (3%).[3] Early detection of coexisting psychiatric disorders in NF1 patients opens the possibility of early treatment and better quality of life. Presented below (after taking informed consent for publication) is one such case of a patient with NF1, ADHD, obsessive-compulsive disorder (OCD), Tourette's syndrome, and depressive disorder, unlike any reported in the literature previously, for which a multidisciplinary treatment approach was utilized.

Case Presentation

A 22-year-old unmarried male, known case of NF1, with no family history of medical or psychiatric illness, with delayed developmental milestones in speech and language domain and academic difficulty, presented with an illness of 16 years with symptoms of hyperactivity, impulsivity, impaired attention and concentration, and complex motor (tapping and touching self and objects in the vicinity, shaking furniture) and simple vocal tics since six years of age, obsessive thoughts of unlucky numbers and compulsive acts (touching objects, producing sounds from mouth in response to a thought of unlucky numbers, which resembled the sound of the vocal tic- saying "aaa") and self-mutilating behavior in the form of repetitive biting of fingers as a part of tic behavior since 12 years of age, and low mood, anhedonia, ideas of helplessness and worthlessness, and decreased appetite, leading to socio academic dysfunction, for the last four months. No improvement was seen with adequate trial of sertraline (200 mg × 8 months) and risperidone (up to 4 mg × 9 years) in the past.

On physical examination, multiple café au lait spots and axillary freckling were present (2/7 clinical criteria for neurofibromatosis in liaison with a neurologist). MRI showed hyperintense lesions in the anteromedial temporal lobe, often a site of involvement in NF1, but they did not justify any of the above symptoms. Yale–Brown Obsessive Compulsive Scale (YBOCS), Yale Global Tic Severity Scale (YGTSS), and Hamilton Depression Rating Scale (HDRS) scores at the time of admission were 27, 81, and 14, respectively. Psychiatric diagnoses of Hyperkinetic Disorder—Disturbance of activity and attention, OCD, de la Tourette's syndrome, and Moderate depressive episode without somatic syndrome were made according to ICD-10.

The patient was given tab fluvoxamine (up to 200 mg) and tab haloperidol 10 mg, followed by gradually building tab clomipramine (up to 300 mg), but minimal improvement was seen after two months. Subsequently, this was augmented with tab sodium valproate (up to 1000 mg) and Habit Reversal Therapy (HRT), after which the scores were YBOCS-11, YGTSS-41, HDRS-2 by the end of three months. A few compulsive acts overlapped with tics (like saying "aaa"), and the patient was not always able to give a clear cognition behind the act, which made it difficult to categorize such acts as compulsions or tics each time they occurred. Engaging the patient in HRT also posed a challenge due to easy distractibility and hence it was discontinued after six weekly sessions. Intelligence quotient (IQ) assessment had revealed borderline IQ, which further complicated the therapy and management.

Discussion

NF1 is a neuroectodermal autosomal dominant condition known to have psychiatric comorbidities such as dysthymia, cognitive and learning deficits, and depressive and anxiety disorders in as many as 30% of patients.[3] ADHD is known to occur in approximately 30% of cases of NF1.[4] ADHD, Tourette's, and OCD as a triad is a well-known occurrence.[5] but there are no reported cases of the entire gamut of ADHD, OCD, Tourette's syndrome, and depressive disorder occurring in a patient of NF1, like this one. While symptoms of hyperactivity, inattention, and borderline IQ complicated the psychotherapy, the overlapping symptomatology between tics and compulsions, with poor reporting of cognition behind the acts by the patient, made the case even more challenging. There is mention of such cases in the literature in which a clear distinction between compulsions and tics is not possible and the patient compulsively performs tics. Hence, the acts have been labeled as "compulsive tics" or "compultics," for which optimal treatment involves anti-obsessional and tic management regimes, as was followed for our patient.[6]

Conclusion

This case had a unique presentation because of the co-occurrence of ADHD, OCD, Tourette's syndrome, depressive disorder, and borderline IQ with NF1, which has not been mentioned in the literature previously as per our knowledge, and the use of a multimodal regime for case management.

Declaration of Conflicting Interests

The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding

The authors received no financial support for the research, authorship, and/or publication of this article.

Statement of Informed Consent and Ethical Approval

Necessary ethical clearances and informed consent was received and obtained respectively before initiating the study from all participants.

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Submitted: 04 Oct. 2021
Accepted: 20 Jan. 2022
Published Online: 09 May. 2022

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HOW TO CITE THIS ARTICLE: Kaur A, Sen MS, Ganesh R, Patra BN and Sagar R. Tourette's Syndrome, Ocd With Adhd As A Triad In Neurofibromatosis Type 1: A Case Report. *Indian J Psychol Med*. 2022;44(3):317–318.

ACCESS THIS ARTICLE ONLINE
Website: journals.sagepub.com/home/szj
DOI: 10.1177/02537176221082894

Successful Combination of Interventions for Psychogenic Blepharospasm—A Case Report

To the editor,

Conversion disorder (CD) is the presentation of abnormal motor or sensory symptoms sans organic causes.[1] Psychogenic blepharospasm is a rare type of CD characterized by unintended twitching or contraction of the eyelids without structural origins.[2] The prevalence is higher in developing countries like India (up to 31% in hospital settings).[3] Limited literature exists on interventions for psychogenic blepharospasm. This letter reports an amalgam of interventions used (Transcutaneous Electrical Nerve Stimulation [TENS, retraining movement], verbal suggestions, and aversion technique [pain/discomfort]) to cause quick resolution of symptoms.

Case Description

A 12-year-old girl, studying in the sixth grade, hailing from Mauritius, came for psychiatric consultation for acute-onset bilateral blepharospasm. She claimed that upon waking one day, she was unable to open her left eye. Three months after this incident, her right eye also followed suit. The condition was continuous, lasting for eight months, accompanied by a mild headache. She used her fingers to keep her eyelids open for performing everyday tasks. No significant medical history or family history was elicited. She displayed la belle indifference. No emotional or behavioral concerns were brought forth. She went on to do her academic work as best as she could. She was examined by a neurologist, psychiatrist, and ophthalmologist in a hospital in India. Due to the presence of transient stiffness, transient tingling over the right-side extremities, and transient tremulousness in the right hand, differential diagnoses myasthenia gravis and autoimmune condition were suspected. She was put on a trial of intravenous immunoglobulin and started on pyridostigmine as empirical therapy. Because the condition began at the start of the pandemic, neurological manifestation of COVID was also suspected. Electromyography, Nerve Conduction Study, Repetitive Nerve Stimulation, HRCT-chest, MRI brain and orbit, and EEG ruled out various differential diagnoses. With this, pyridostigmine was discontinued. Psychological evaluation (Sentence Completion Test, Thematic Apperception Test, Rorschach Ink Blot Test, Millon Clinical Multiaxial Inventory) highlighted the intense need for affection and support from the immediate environment leading to underlying anxiety. A provisional diagnosis of CD (F44.4) was made. Ptosis crutches were prescribed and family counseling was suggested. She was brought to us as an outpatient by her mother through a referral from a friend. She was re-examined, and psychogenic origins were confirmed.

The treatment plan consisted of retraining the movement of eyelids (TENS) while diverting attention (trail-making tests) and providing suggestions. Previous literature claimed that TENS was effective in treating psychogenic movement disorders as it has neuro-modulatory effects.[4] On a predetermined date, she came to the lab for an initial meeting with the team of practitioners. She was first primed to believe that she had a genuine problem that could be undone using physiotherapy and psychotherapy. During her first (and only) session, she had to do a series of trail-making tests while her frontalis, corrugator supercilia, and orbicularis oculi muscles were electrically stimulated with minimal palpable muscle contraction. For each muscle, 30 contractions were given. During the stimulation and immediately after, her left eye had partially opened. The patient was informed that her eyes might open any time before the next session.