ADHD, TOURETTE SYNDROME AND KABUKI SYNDROME
– CASE PRESENTATION –

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ABSTRACT
This paper presents a case of an inpatient in Child and Adolescent Psychiatry Clinic, “Prof. Dr. Al. Obregia” Psychiatry Hospital, Bucharest, admitted for hyperactivity, impairments in school functioning, multiple motor and vocal tics and distinctive facial features. The data obtained from his personal history, clinical examination, genetic lab results and psychological evaluation were significant for DSM IV-TR and ICD 10 diagnostic criteria for ADHD, Tourette Syndrome and Kabuki Syndrome.

Keywords: impairments in school functioning, tics, distinctive facial features

INTRODUCTION
Tourette syndrome (TS) is a childhood neuropsychiatric disorder characterized by motor and phonic (vocal) tics. It is often associated with behavior disorders, particularly obsessive - compulsive disorder (OCD) and attention deficit hyperactivity disorder (ADHD). These behavior disorders often accompany the tics and may dominate the clinical picture in some patients. TS is a genetic condition that runs in the family. However, the precise genetic abnormality responsible for the phenotype has not yet been elucidated. Tourette syndrome (TS) is clinically significant because it is much more common than previously thought, is easily misdiagnosed, and is often associated with significant comorbidity [1].

CASE PRESENTATION
Patient L.C, aged 11, known with ADHD and mild mental retardation, from 3 years, referred to our clinic for multiples motor tics (involuntary movements of the eyes, facial grimaces, shoulder movements, later - involuntary movements of the abdomen) symptoms that persisted for over 1 year.

Her history reveals: delayed psychomotor development (she held her head up at 6 months, she sat at 1 year, she walked at 1 year and 9 months, she said her first words after 2 years) and a brother with history of psychoactive substances use and mental retardation. The patient lives with her parents and brother in an urban area, near our clinic and she is in IVth grade, having satisfactory academic result.

Clinical somatic, ocular and neurological examinations were normal, with few exceptions regarding distinctive facial features including: arched eyebrows, long eyelashes, long openings of eyelids (long palpebral fissures) with the lower lid turned out (everted) at the outside edges, a flat, broadened tip of the nose and large protruding earlobes and skeletal abnormalities. This facial pattern is commonly described in Kabuki syndrome and therefore genetic tests aiming to confirm the diagnosis are in progress.

Laboratory tests (CBC, blood glucose, serum calcium, transaminases) were normal. EEG line performed while the patient was awake – without pathological graphic elements.

Psychological examination performed in the current presentation, highlights: Hyperkinetic behavior, easily distracted, verbosity, undiscriminating attachment, spontaneity, cognitive capacity (IQ = 67).

The Mental State Examination: The patient (a girl) is cooperative, self-conscious, oriented, visual and psychic contact easily obtained, abnormal involuntary movements (involuntary eyes movements, facial grimaces, shoulder movements, abdomen movements) and vocal tics (voice readjust). The patient doesn’t have quali-

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tative perception disorders at the present examination, difficulty focusing, distractibility, no altered thought content, with a wide range of emotional expression during the assessment, normally spontaneous speech, hyperactive behavior, without appetite alteration and no sleep disorders.

Tourette syndrome is diagnosed in the presence of multiple motor tics and one or more vocal tics that appear before 18 years of age. Therefore, no specific laboratory or genetic tests is needed in order to establish this diagnosis. The keys to diagnosis are recognition and an index of suspicion.

The practitioner could potentially diagnose tics as chorea, myoclonus or stereotype movements and initiate a work up to evaluate these conditions. In the present case our assessment excluded these alternative conditions [2].

Phonic tics, such as throat clearing, sniffing or cough, may be mistaken for allergies or asthma which were all eliminated by clinical investigations.

TS should be differentiated from transient tic disorder of childhood, which is similar to TS but lasts for less than one year.

The psycho-pharmacological treatment that was established in the first days of hospitalization was meant to diminish involuntary movements. Therefore, small doses of an atypical antipsychotic were used together with ADHD specific medication. In addition, patient education was performed including measures that were taken to nurture self-esteem and self-correction: individual counseling, cognitive and behavioral therapies, and group therapy were considered as well.

At discharge, recommendations also were made in favor of supportive therapy sessions in order to help the patient alleviate the discomfort due to tics, therefore improving social functioning.

**Evolution and prognosis**

Initially, involuntary movements had single localization (facial grimaces), but under family pressure (especially her mother who thinks that these involuntary movements are intentional made by the patient in need of attention) they continued and new additional tics developed.

Having ADHD as a comorbid condition, a more severe outcome is expected. The associated behavioral disorders or ADHD often cause more morbidity than the tics themselves. In children, behavioral complications frequently lead to poor academic performance, social isolation, and emotional problems. Moreover, the associated tics of TS can be somewhat volitionally suppressed, and the mental and emotional effort used to suppress tics may also interfere with attention and concentration in school and work.

**CONCLUSIONS**

The management of Tourette syndrome is multifaceted. Treating patients with TS should be a collaborative effort. The neurologist, psychiatrist, psychologist, family members, and school professionals all have important roles. The approach is primarily aimed at medically managing frequent or disabling tics, treating coexisting behavioral symptoms, and patient and family education. Ideally, patients with mild tics who have succeeded in adapting to their conditions can avoid the use of medication. Individual, group, or family counseling may help in facilitating a healthy adaptation to the illness.

Educating patients, family members, peers, and school personnel regarding the nature of TS; restructuring the school environment and providing supportive counseling are measures that may be sufficient to avoid pharmacotherapy. Pharmacologic therapy for tics is considered when tics interfere with social interaction, school performance, or daily activities. The goal of such therapy is not completely eliminating the tic, but rather controlling tics in order to alleviate the social embarrassment or discomfort caused by them, therefore improving social functioning.

Areas of strength should be emphasized, such as talents and skills, interests, any family or peer support, and psychological resilience. Social skills training can help develop and reinforce more effective methods used to raise confidence and improve children communication. Parents or other guardians may benefit from parent behavior management and discipline training, recognizing that the underlying purpose is to instill a sense of self-control and responsibility for one’s behavior.

Allowances must be made for the child’s uncontrollable behaviors that result from the disorder, but some behaviors, such as spitting at others or
obscene gestures have negative social connotations and require special guidance. Methods to help the child manage these behaviors include nonjudgmental acceptance of the child regardless of the nature of the behaviors and working with the child to adapt or find alternative, more appropriate behaviors that satisfy premonitory urges, such as spitting into a handkerchief instead of spitting openly.

Parenting skills books, workshops, and trained specialists are widely available and emphasize on practical methods in positive reinforcement of desirable behaviors through giving praise or rewards, modeling appropriate behaviors, and administering “time-outs” from rewards or attention for inappropriate or uncontrolled behaviors. Parents may also benefit from group support and education or other topical groups and from individual supportive counseling to cope with accompanying stress. Several relaxation or stress management approaches reportedly improve tics in TS. For example, tics are known to worsen from stress and to improve during periods of relaxation. Whether such therapies have a direct effect on the tics or exert an indirect influence by allowing patients to deal more productively with life stresses is unclear [1].

REFERENCES