FAMILIAL PAROXYSMAL KINESIGENIC CHOREOATHETOSIS

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Abstract –A 14-year old boy was presented with a rare form of movement-induced drop attacks, which was also present in his father. This case was, therefore, labeled as familial paroxysmal kinesigenic choreoathetosis.

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Key words: movement disorders; choreoathetosis; drop attacks

INTRODUCTION

Movement disorders are among the most important neurological conditions to be differentiated from epileptic seizures (1). Often the clinical data, if properly collected, can differentiate between the drop attack of epileptic nature and movement-induced falls.

CASE REPORT

A 14-year-old boy from Abargho (Abarkouh, Fars Province) presented in early July of 1994 with the chief complaint of drop attacks precipitated by standing up or walking, since about 4 years ago. The attacks lasted from several seconds up to several minutes, occurring at the moment the patient initiated walking from a resting position, and in the severest form, causing him to fall and injure himself on the forehead and knees. No alteration of consciousness, tongue biting, bladder or bowl incontinence, or vomiting accompanied the attacks, nor did he become drowsy or sleepy post-ictally. Past medical history was unremarkable. No significant family history, except a close relative with febrile seizures, could be elucidated at the initial visit. The medical and neurological examinations were totally normal. Paraclinical work-up including EEG, head CT, fasting blood sugar, calcium, cardiovascular evaluation, (including EKG, and Echocardiography), nerve conduction velocity, electromyography (EMG), serum creatine phospho-kinase (CPK), and serum cerulopasmin level, were all normal. Clonazepam, 0.5 mg, 3 times daily, was started for him on the outpatient basis with the impression of dystonic attacks in mind. This initially decreased the attacks, but 2 days later the patient referred with more drops, in one of which he was injured on the scalp, with negative skull x-ray. Admission to the ward led to the observation of generalized spastic attacks with the above-mentioned characteristics making him unable to carry out his daily activities. No clinical indication of a seizure was noted. Later, his father, being absent till that time, attested to similar attacks in himself when he was 15-30 years old but had later remitted spontaneously. Hereby, the diagnosis of Familial Paroxysmal Kinesigenic Choreoathetosis (FPKC) was made, and the patient was put on phenytoin (dilantin), 20 mg/kg, loading dose, after which he developed severe vertigo, probably due to rapid loading, but later he tolerated it well. One day later the attacks decreased in intensity and duration, and he was discharged with 150 mg phenytoin daily. One week later he was able to stand up and walk freely. Later, slit-lamp examination of the eyes for Kaiser-Fleischer ring was negative, again ruling out Wilson's disease.

DISCUSSION

Familial paroxysmal kinesigenic choreoathetosis is a relatively rare form of movement disorders which was first known as movement-induced epilepsy (1). Later , it

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was called one form of reflex epilepsy, and then epilepsy originating from basal ganglia. As more patients were reported in the literature, especially those with affected family members, this disorder came to be known as a movement disorder rather than a seizure phenomenon. The movement-induced attacks start suddenly and can be a combination of dystonia, chorea, athetosis, and ballism (1). Tonic spasms of a few seconds up to maximum of 5 minutes begin abruptly as the patient tries to rise from a resting state, thus limiting patient's motor activities. Severe attacks may throw the patient to the ground. Immediately preceding the attacks, the patient may have muscle stiffness, numbness and paresthsia in the extremities, or feel tense. These attacks can be provoked by hyperventilation (1). Speech may be temporarily disturbed during the attack, but the level of consciousness, and post-ictal phenomena (incontinence, drowsiness, or sleep) never occur.

In the familial, idiopathic form, as in above patient, the disease is transmitted as an autosomal dominant trait, more in males than in females. The clinical manifestations begin most commonly at 6-16 years (range: 6 months to 40 years). The attack may reach up to 100 times per day in frequency, but decreases as the patient reaches adulthood.

In this disease all paraclinical evaluations (head CT, EMG, EKG, and central nervous system histology) are normal. EEG, except in one case which showed spike-wave activity, has been reported normal.

In a few autopsied cases the only finding has been a mild increase in melanin pigment content of locus coerleus of the brain (2). The disease responds to most anti-convulsant drugs, most notably phenytoin (dilantin). The secondary (symptomatic) form is seen most commonly in multiple sclerosis—sometimes as the first indicator—head injury, idiopathic hypoparathyroidism, perinatal hypoxic encephalopathy, putaminal as well as thalamic infarction (1). Differential diagnosis includes frontal lobe epilepsy, myoclonic epilepsies, dystonias secondary to neuroleptic drugs, and cerebellar ataxias, which are all differentiated on clinical-paraclinical grounds (1,2). In summary, patient's clinical picture fitted the diagnosis of FPKC, with a good reponse to its drug of choice.

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