Familial orthostatic tremor and essential tremor in two young brothers: A rare entity

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Abstract

Orthostatic tremor (OT), is usually a disease of old age and is characterized by quivering movements of the legs during quiet standing or in the state of isometric contraction in the lower limbs. This is relieved on walking or on lying down. It is diagnosed by surface electromyography, particularly over the quadriceps femoris muscles which shows a distinctive frequency of 13 to 18 Hz on standing. Some investigators consider it as a variant of essential tremor (ET) and the two conditions often co-exist. The disease is usually non-familial. Two brothers presented with tremor in the lower limbs on standing and on the outstretched hands without any family history. Subsequently, they were proved to be suffering from OT and ET by clinical examination and surface EMG. Simultaneous occurrence of OT and ET in two young brothers without any family history in the previous generation has not been described before and they also appeared at a much earlier age than what is described in the literature.

Key Words

Essential tremor, familial, orthostatic tremor, young age

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Introduction

Orthostatic tremor (OT) is a rare condition that was first reported in 1970 by Pazzaglia *et al*.,[1] though Heilman described it in three subjects as a distinctive clinical entity in 1984 and he coined the term.[2] It is usually a disease of advanced age and it consists in quivering movement, particularly of the legs and the trunk during standing or in the state of isometric contraction in the lower limbs in the supine posture and is accompanied by a curious sensation of unsteadiness that is relieved almost immediately on walking or on lying down. The condition is characterized by a distinctive 13-18 Hz bursts in electromyography (EMG) in the lower extremities on standing and sometimes in the upper extremities as well. It disappears while the patient is supine or ambulant. The condition is often considered a disease of station rather than gait and is now thought to be exaggerated physiological response to instability.[3]

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Since contraction of muscles in the limbs is central to the genesis of OT, some authors believe that this condition is a variant of essential tremor (ET). This view is further supported by the observation that positive family history of ET, head tremor, or arm tremor often co-exist with OT and the burst of tremor has a wide range of frequency (6-20 Hz).[4] However, alcohol and beta-blockers do not abolish OT and therefore, this observation does not support this hypothesis. Furthermore, the family history is usually negative and the degree of synchrony between the different groups of contracting muscles in OT is too high to support the view that one is the variant of the other. On the other hand, it has been shown in a number of studies that there is abnormal bilateral cerebellar hyperactivity in OT, as evident by positron emission tomography (PET) studies and therefore, they support the view that the two conditions are closely related, since such abnormalities are also seen in ET and primary writing tremor.[5,6] Furthermore, it is generally considered that OT is mediated by a central oscillator[7] and in this respect it resembles ET.

We report two brothers aged 12 and 7 years, respectively, who clinically and electrophysiologically satisfied the criteria for OT and both of them also displayed postural and action tremor in the upper limbs, suggestive of ET. Electrophysiological studies also confirmed the nature of this tremor in the outstretched hands.

Case Reports

Case 1

A young boy aged 12 years attended the outpatient department of tertiary care medical college hospital with the complaint of tremulousness in the legs on standing but the problem resolved on walking or on lying down. History of similar illness was seen in his younger brother but not in his parents. He further complained that his hands shake while lifting a glass of water for drinking and the tremor increased as the hand was taken closer to the lips.

On examination, we observed that the child had been quivering in the lower limbs while standing and it was also present in the upper limb to some extent. Fine ripples of movements were observed in the thigh and the calf muscles. The movements disappeared when he was asked to walk, sit, or lie down, but reappeared when asked to put the lower limbs in the state of isometric contraction while lying down. The outstretched hands showed fine tremor which increased during finger – nose test. The boy suffered from acute anterior poliomyelitis in childhood with wasting of the muscles of the right lower limb and there was scoliosis to the right as a compensatory mechanism.

Case 2

The younger brother aged 7 years also attended the clinic and complained of similar problems, though to a lesser degree. On examination, there was evidence of fine tremor in the quadriceps femoris muscles on standing which was relieved on lying down and he also displayed tremor in the outstretched hands.

Video recording was done for documenting the problem in the two brothers.

Investigations

Hemogram, chest X-ray, renal and liver function tests, thyroid function tests, electroencephalogram, and contrast-enhanced CT scan were non-contributory and copper profile was negative. Surface EMG of the quadriceps muscles showed discharges of muscle activity at the frequency of 15-16 Hz in the elder brother and 14-15 Hz in the younger brother in the standing position [Figures 1 and 2]. Surface EMG in the outstretched hands in the flexors of the fingers showed discharges of muscle activity at the rate of 6-7 Hz in both the brothers [Figure 3].

Management

Clonazepam in incremental dose from 0.5 to 4 mg a day was administered to both of them without much improvement. Gabapentin was added up to the dose of 300 mg four times a day which also turned futile. However, long-acting propranolol, 40 mg a day, considerably reduced the tremor in the hands. Soon thereafter, we lost them in the follow-up.

Discussion

OT is a rare disorder and generally occurs in the elderly subjects and is usually non-familial. The largest series of 41 patients have been published by Gerschlager *et al.*,[8] in 2004 and Piboolnurak *et al.*, have also described 26 cases later.[9] Deuschel *et al.*, recommended the clinical and electrophysiological criteria for diagnosing OT,[10] while Gerschlager *et al.*,[8] brought to attention two broad groups of the condition – those with

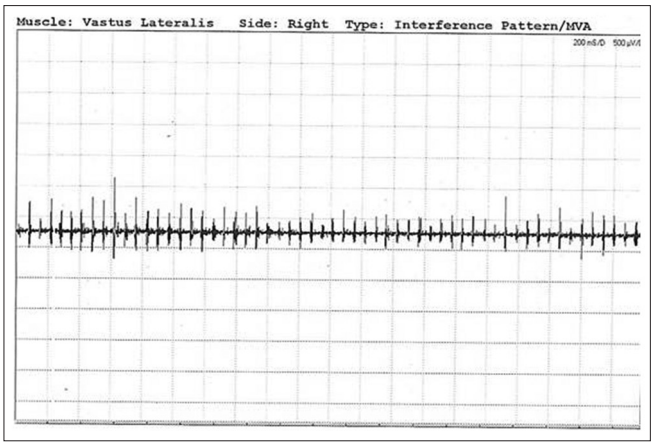

Figure 1: Surface EMG of the elder brother from right vastus lateralis showing discharge at the frequency of 15 to 16 Hz

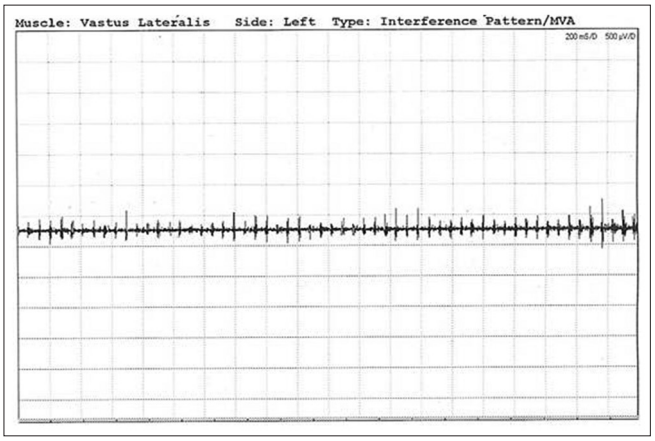

Figure 2: Surface EMG of the younger brother from left vastus Lateralis showing discharge at the frequency of 14 to 15 Hz

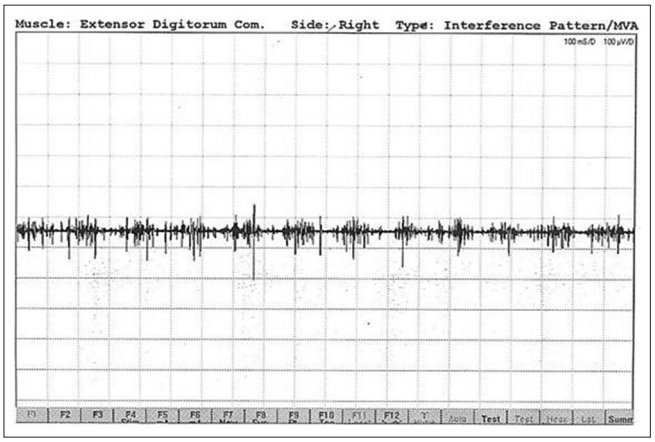

Figure 3: Surface EMG of the elder brother from right extensor digitorum communis, showing discharge at the frequency of 6 to 7 Hz

"primary OT" with or without postural arm tremor and those with "OT plus," where there are associated other movement disorders. Patients in the primary OT group were further subdivided into "OT and ET," where there was evidence of postural arm tremor and "pure OT," where such tremor in the outstretched hands was absent.

Though OT is mostly an idiopathic condition and is generally non-familial, Wee *et al.*,[4] however, hinted at the familial nature of the disease and Gerschlager *et al.*,[8] touched upon the same in their work. The latter group observed that none in the pure OT group reported any movement disorder in the relatives. However, three out of 21 patients with OT and ET reported leg tremor in two sisters, though none was personally examined by the authors. Three patients with OT and ET had first-degree relatives with arm tremor. They also reported that 24 out of the 31 cases of primary OT showed evidence of ET. Interestingly, eight patients of OT along with ET improved with alcohol and five patients had positive family history of other movement disorders in the family. On the other hand, none of the patients with pure OT had positive family history and none responded to alcohol. Since ET characteristically improves with alcohol, these findings tend to rule out the hypothesis that OT and ET are the variants of the same disease entity.

The incidence of OT and ET occurring in a family is rare. The tremor in the legs was relieved with clonazepam but not with beta-blockers, thus indicating clearly that OT is not a variant of ET. Recently, OT has been described in monozygotic twins and in two brothers as well.[11,12] However, in our study, the two siblings presented with both OT and ET. Furthermore, the reported age of onset in patients with OT and ET in the study of Gerschlager *et al.*,[8] was 50.4 ± 15.1 years, whereas in our patients, it was observed when they were aged 13 years and 7 years, respectively. The importance of the problem in our experience is that OT with ET in two siblings occurring at such an early age has not been reported in the past.

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