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Case report

Natural evolution from idiopathic photosensitive occipital lobe epilepsy to idiopathic generalized epilepsy in an untreated young patient

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Abstract

Idiopathic photosensitive occipital lobe epilepsy (IPOE) is an idiopathic localization-related epilepsy characterized by age-related onset, specific mode of precipitation, occipital photic-induced seizures – frequently consisting of visual symptoms – and good prognosis. This uncommon epilepsy, which usually starts in childhood or adolescence, has rarely been observed in families in which idiopathic generalized epilepsy also affects other members. We describe a nuclear family in which the proband showed electro-clinical features of idiopathic photosensitive occipital lobe epilepsy in childhood, which subsequently evolved into absences and a single generalized tonico-clonic seizure in early adolescence. His mother had features suggestive of juvenile myoclonic epilepsy. This case illustrates a continuum between focal and generalized entities in the spectrum of the so-called idiopathic (genetically determined) epileptic syndromes.

Keywords: Photosensitivity; Idiopathic photosensitive occipital lobe epilepsy; Juvenile myoclonic epilepsy; Electroencephalography

1. Introduction

Idiopathic photosensitive occipital lobe epilepsy (IPOE) is an uncommon idiopathic localization-related epilepsy with age-related onset, specific mode of precipitation and good prognosis [1]. Occipital seizures, always photic-induced, consist of elementary visual symptoms. Interictal electroencephalography (EEG) abnormalities can be focal (occipital) and/or generalized, and intermittent photic stimulation (IPS) induces a generalized and/or an occipital photoparoxysmal response (PPR). PPR is an abnormal brain reaction to flickering lights, exhibiting a genetic component whose manifestations range from an EEG trait to clinical seizures in susceptible individuals [2].

Juvenile myoclonic epilepsy (JME) is a common syndrome of idiopathic generalized epilepsy (IGE) and is characterized by the clinical triad of myoclonic seizures, generalized tonic–clonic seizures, and typical absence seizures (one third of cases). Interictal EEG shows generalized 3–6 Hz SW/PSW and a PPR in one third to 75% of patients [3].

IPOE and JME are two idiopathic syndromes manifesting with different epileptic seizures (focal and gener-

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alized, respectively) and sharing common characteristics: polygenic aetiology, onset in childhood or adolescence, generalized EEG abnormalities and photosensitivity. Here, we describe a nuclear family in which the proband shows a phenotypic progression from partial photic-induced to generalized seizures, and the mother has a JME.

2. Case report

The proband, a 15 year-old boy with normal development, began to experience episodes characterized by a sudden vision of "colored spots...red, green, pink...moving in front of me and on the right...like a flash" at the age of six. These episodes were all triggered by photic stimuli, such as natural flickering lights, TV or computer. EEG studies demonstrated rare interictal spikes or polyspikes with posterior topography, and a generalized PPR with 3 Hz SW and PSW discharges (Fig. 1a). IPS induced focal seizures which originated from left temporo-occipital regions and were characterized by visual hallucinations followed by mental slowing (Fig. 1b and c). Electro-clinical findings suggested an IPOE and he was followed up without antiepileptic drugs (AEDs).

Subsequent video-EEG recordings showed the evolution of electro-clinical patterns over time. From 9 to 11 years of age, EEG activity was similar to that observed at onset. More recent EEG recordings (age 11–15) showed progressive change in electro-clinical features: generalized 3.5 Hz SW/PSW interictal discharges with progressively more regular and typical morphology replaced the initial focal abnormalities (Fig. 2a). An ambulatory 24-h EEG recording even showed activation of these typical generalized 3–3.5 SW/PSW on awakening (Fig. 2b). Furthermore, the patient presented a generalized tonic–clonic seizure without any evident preceding focal sign and also began to experience typical absence seizures. These absence seizures, recorded during video-EEG monitoring after IPS, consisted of a brief loss of contact, with staring and perioral myoclonic jerks, and were related to 3.5 Hz generalized PSW discharge (Fig. 2c). He still does not take AEDs and avoids precipitant factors.

The 49 year old proband's mother had a single tonic–clonic seizure at age of sixteen and experienced upper limb myoclonic jerks on awakening. Interictal EEG was characterized by generalized 3–4 Hz SW/PSW discharges and generalized PPR, thus confirming the clinical diagnosis of JME. She currently takes valproate.

3. Discussion

We report a nuclear family with two different idiopathic syndromes: the patient with IPOE evolving into generalized epilepsy, and his mother with JME. This case is unique as it demonstrates the natural evolution from exclusively photic-induced occipital seizures to the progressive emergence of a generalized electro-clinical pattern in an untreated patient.

Although the patient has a tendency towards IGE (supported by the family history) and that a coincidental association between IPOE and JME cannot be ruled out, the previous reports of IPOE seen in families with IGE suggest that these two entities share a common background and support the hypothesis of their syndromic association.

In fact, the coexistence of IPOE and JME has been previously described in 4 families [4], as well as in non-related patients with JME who exhibit a variable combination of visual symptoms and myoclonic seizures [4]. This continuum has also been suggested by recent observation of children with idiopathic focal or generalized epilepsies accompanied by additional occipital seizures, and of IPOE patients with generalized seizures, either photic-induced or spontaneous [5]. Although the same author emphasizes how rarely the association between idiopathic focal epilepsy and IGE occurs, these observations indicate that, in these idiopathic conditions, the border between focal and generalized seizures is not so clear and the similarities cannot be confined to photosensitivity.

From a pathophysiological standpoint, the thalamocortical system seems to be involved in the generation of both focal and generalized seizures. In photosensitive patients the discharge arises from visual cortex, where magnocellular projections synchronize first cortical and then subcortical reticular activity [2]. This could explain different semiologies observed in photosensitive patients. Predominant cortical involvement is reflected by early visual symptoms, promptly followed by phenomena due to discharge spread, especially to motor areas. Similarly, early (but likely secondary) involvement of subcortical structures may be responsible for generalized manifestations such as absences, according to Gloor's concept of generalized cortico-centrencephalic epilepsies [6]. In patients with syndromic IPOE/IGE overlap, visual stimuli can trigger abnormal activation of this cortical–subcortical network, whose variable involvement can generate different (focal and/or generalized) electro-clinical features in the same family or in the same patient. This can be interpreted under the light of the more recent "cortical focus theory", showing that the cortex plays a leading role in the generation of generalized spike–wave discharges [7].

In the present report the natural progression (which to our knowledge has never been reported) of the proband's untreated epilepsy from exclusively reflex occipital to generalized seizures, seems to reinforce the modern concept of the continuum in the spectrum of some idiopathic conditions. Such co-existence is debated and the overlap is considered limited by some authors