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Papillon Lefevre Syndrome with Abdominal Epilepsy

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Abstract

Observation: Papillon-Lefevre syndrome is a rare autosomal recessive disorder characterized by diffuse palmaplantar hyperkeratosis combined with early loss of primary and permanent teeth. Abdominal epilepsy is a rare condition most frequently found in children, consisting of gastrointestinal disturbances caused by epileptiform seizure activity. Extremely limited number of palmoplantar keratoderma cases associated with epilepsy was reported in the literature. Here we are presenting a case of PLS with abdominal epilepsy that to our knowledge has never been previously reported.

Introduction

Papillon-Lefevre syndrome (PLS) is a very rare syndrome of autosomal recessive inheritance characterized by periodontitis and palmoplantar keratoderma. It has prevalence of 1-4 cases per million persons [1]. Cranial calcification or symptoms due to brain abscess associated with this syndrome were rarely reported [1, 2]. Here we are presenting a case of PLS with abdominal epilepsy that to our knowledge has never been previously reported.

Case Report

A-20-year-old man with a previous diagnosis of PLS visited our outpatient clinic with chief complaint of paroxysmal nausea and epigastric pain which happens almost monthly and lasting approximately 30 minutes during the last three years. His history revealed that only four of his deciduous teeth had erupted normally after birth then lost all of them gradually. He also gave a history of development of fissures and thickening in his palmaplantar skin since age of 1-2 years but no improvement with the given treatments was seen.

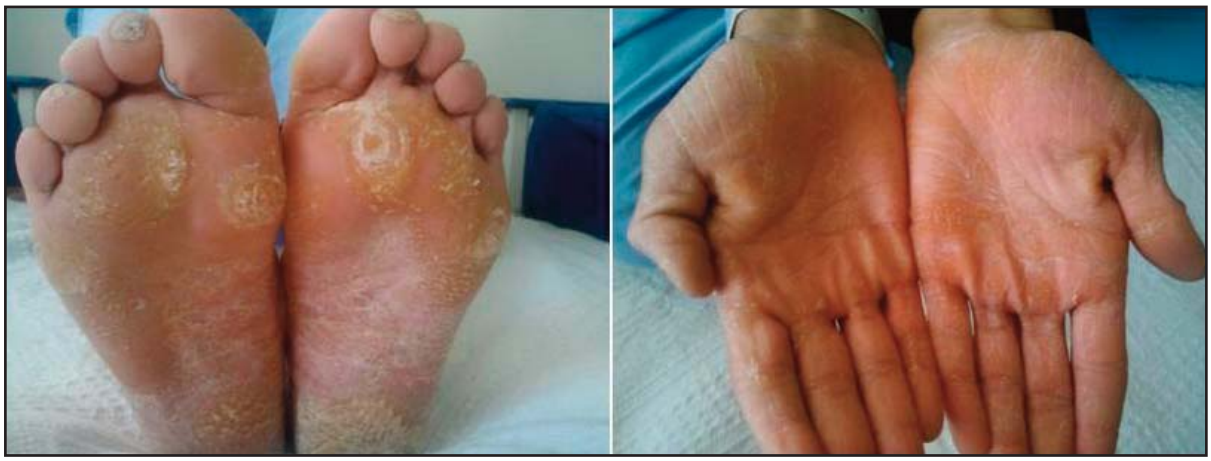

Figure 1. Keratotic plaques on skin of both palms and soles

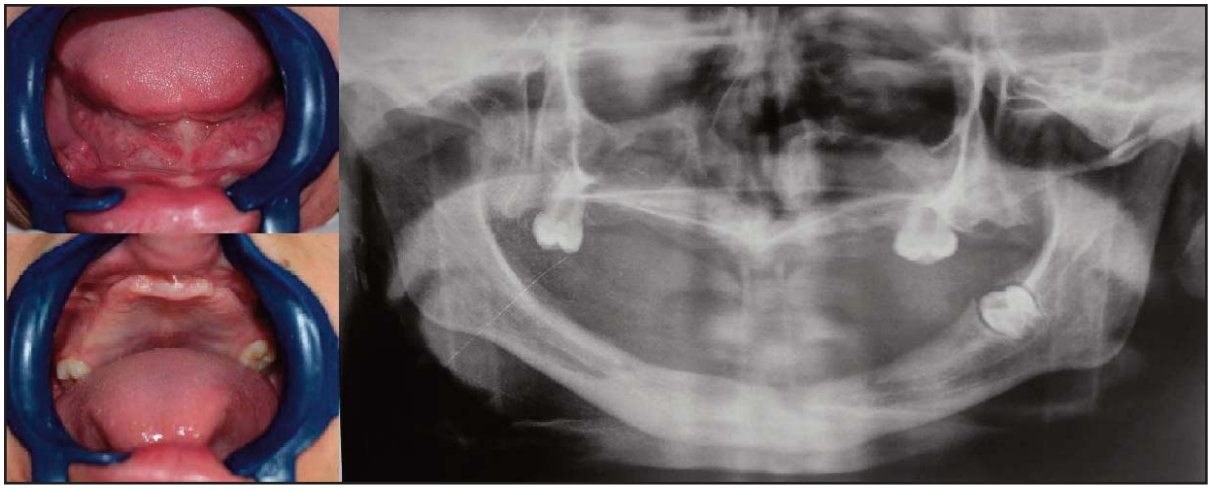

Figure 2. Only two molar teeth can be seen at intra-oral examination (Left). Radiographically, there was another molar on the left mandibula (Right)

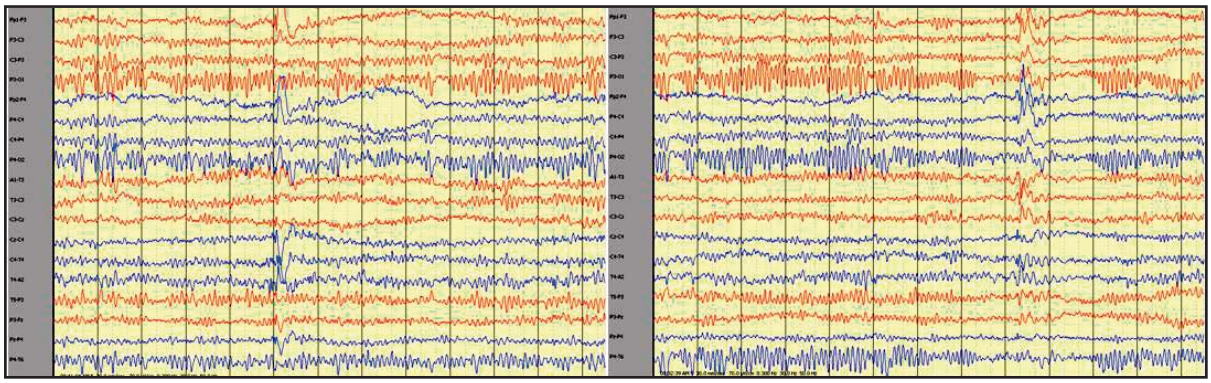

Figure 3. EEG findings, generalized sharp waves

After complaining of periodic nausea and epigastric pain he went to a gastroenterologist, however examination and all the laboratory results were in normal ranges. His neurological examination revealed nothing abnormal. Skin of both palms and soles was peeling off suggestive of keratoderma (**Figure 1**). On the intraoral examination, it was observed that the patient was edentulous in both the upper and lower jaws except two maxillary molar teeth resembling third molars. A dental panoramic radiograph showed the presence of an impacted lower third molar and severe alveolar bone loss (**Figure 2**). Although electroencephalogram (EEG) showing bitemporal sharp and slow waves and rare generalized spike and waves (**Figure 3**) discharges the cranial magnetic resonance imaging (MRI) scan was normal. After these results, he was considered as abdominal epilepsy and started carbamazepine, then gradually increased up to 600 mg/daily. The patient responded to this treatment very well and attacks of abdominal pain with nausea completely ended since last 6 mounts.

Discussion

Abdominal epilepsy is a rare condition most frequently found in children, consisting of gastrointestinal disturbances caused by epileptiform seizure activity. A criterion for diagnosis of abdominal epilepsy includes frequent periodic abdominal symptoms, an abnormal EEG and significant improvement of gastrointestinal symptoms after taking antiseizure medication [**3**]. To our knowledge this is the first report of PLS with epilepsy.

Extremely limited number of palmoplantar keratoderma cases associated with epilepsy was reported in the literature. In a case of inherited palmoplantar hyperkeratosis with facial dysmorphism, clinodactyly, deafness and hypodonti the concurrence with epilepsy was reported [**4**]. Serrano Castro et al. reported the coexistence of focal epilepsy with Vohwinkel syndrome which is characterized by diffuse palmoplantar keratoderma, sensorineural de-

afness and skeletal abnormalities [**5**]. They asserted that missense mutation in a connexin gene (connexin 26) can include epileptic manifestations. Connexins are transmembrane proteins rolled in intercellular communication processes. They also have been experimentally demonstrated to participate in epidermal differentiation [**6**].

Although, it is not possible to reach a definitive conviction, we find it interesting to observe a coexistence of these two rare diseases. For confirming whether an association between these two conditions controlled clinical and genetically trials are needed.

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