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Case Report

Hereditary Hemorrhagic Telangiectasia presenting as migraine: A case report

Carlotta Facini [a,*], Elena Pavlidis [a], Emanuela Claudia Turco [b], Francesco Pisani [a]

[a] *Child Neuropsychiatry Unit, Neuroscience Department, University of Parma, Parma, Italy*
[b] *Child Neuropsychiatry Unit, Mother and Child Department, University-Hospital of Parma, Parma, Italy*

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Abstract

Background: Hemorrhagic Hereditary Telangiectasia (HHT) is an autosomal dominant disease characterized by the presence of multiple arteriovenous malformations (AVMs). Migraine is described in association with HHT in adulthood, while only few paediatric cases are reported in the literature.

Aim: In this paper, we describe an atypical case of HHT in a 7-year-old boy, who presented severe and recurrent episodes of migraine-like headache as the first symptom of the disease.

Methods: The patient was accurately investigated both clinically (general, neurological and dermatological examinations), instrumentally (electroencephalogram, brain magnetic resonance, transcranic Eco-color-Doppler, contrast echocardiography and enhanced chest computed tomography) and genetically.

Results: Familial history was positive for HHT in the paternal line. Both general and neurological examination were normal. Brain magnetic resonance imaging showed a minor old infarct in the right parietal and occipital lobes. Transcranic Eco-color-Doppler, contrast echocardiography and enhanced chest computed tomography revealed a pulmonary AVM. Cephalalgia resolved after transcatheter embolotherapy. A genetic test, identifying the mutation in endoglin gene both in the patient and in the father, confirmed the suspected diagnosis of HHT.

Conclusions: Although headache is rarely reported in children as the first symptom of HHT, we warn clinicians on this possible link, as a promptly diagnosis is advisable in order to prevent potential complications.

Keywords: Hemorrhagic Hereditary Telangiectasia; Migraine; Secondary cephalalgia; Arteriovenous malformations

1. Background

Hemorrhagic Hereditary Telangiectasia (HHT) is an autosomal dominant vascular dysplasia with a high penetrance but variable expression [1]. Its estimated prevalence is 1/5000–10000 [1,2]. It is caused by mutations in genes of endoglin (ENG), activin receptor-like kinase 1 (ACVRL1/ALK1) and SMAD family member 4 (SMAD4), which are involved in the transforming growth factor-β (TGF-β) signaling cascade, that plays an important rule in the regulation of angiogenesis [1]. The clinical diagnosis can be delayed and the incidence underestimated, because the typical signs, epistaxis and telangiectases, occur often after puberty [3] and arteriovenous malformations (AVMs) can remain asymptomatic for a long time [2]. Thus, life-threatening events and severe complications of AVMs,

* Corresponding author at: Child Neuropsychiatry Unit, Neuroscience Department, University of Parma, Via Gramsci 14, 43126 Parma (PR), Italy. Tel.: +39 521 702205; fax: +39 521 704708.
E-mail address: carlottafacini@gmail.com (C. Facini).

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such as intracranial hemorrhage or stroke, could be the first manifestation of HHT in children [2,4,5]. Neurological symptoms can be due to cerebral AVMs (CAVMs) or, more frequently, to pulmonary AVMs (PAVMs) causing a right-to-left shunt with paradoxical embolism [6]. The prevalence of migraine in adult HHT patients is higher than in the general population, varying between 16% and 57% [7]. The medical literature provides only few information about headache in HHT children [2–4,8].

In this paper, we report a peculiar case of a 7-year-old boy, who presented with a severe migraine-like headache as the first symptom of HHT. We suggest clinicians to consider this possible HHT presentation, in order to

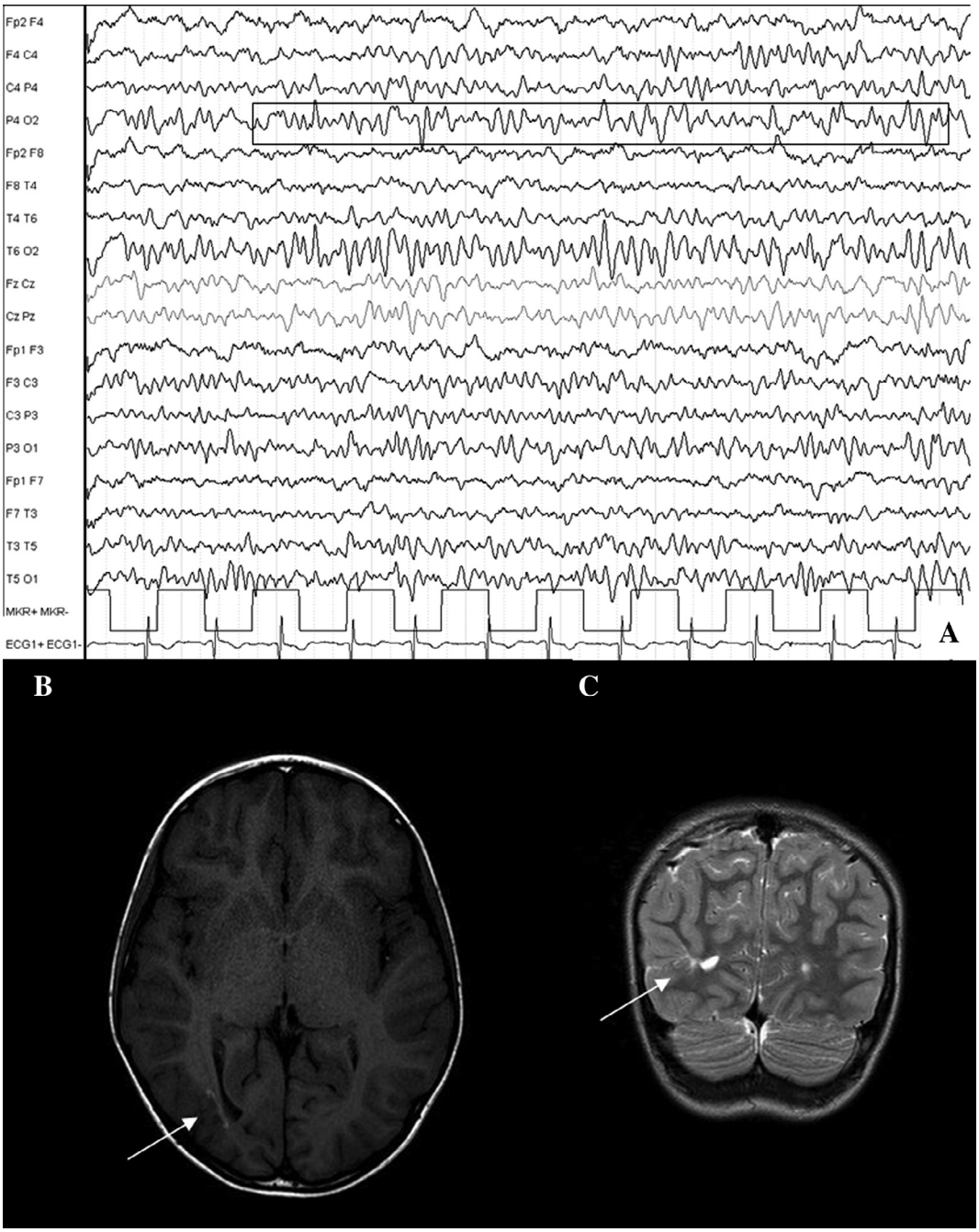

Fig. 1. (A) EEG recording during wakefulness, showing an asymmetry of the background activity with theta-delta rhythms in the right posterior areas (bandpass filter: 1, 6–70 Hz; notch 50 Hz; sens: 100 μV/cm). (B) (axial T1-weighted image) and (C) (coronal T2-weighted image): MRI images showing a small infarct in the right occipito-parietal lobe.