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Case Report

Brain Abscess Secondary to Visceral AVMS in Hereditary Haemorrhagic Telangiectasia

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ABSTRACT

Hereditary Hemorrhagic Telangiectasia (HHT) is an autosomal dominant disorder associated with various neurological complications arising from vascular malformations.

We present a case of a 49-year-old female who presented with focal seizures. Imaging revealed a brain abscess. Further investigation, prompted by a history of recurrent epistaxis, identified multiple pulmonary and liver arteriovenous malformations (AVMs). The brain lesion was confirmed to be an abscess and the patient was subsequently diagnosed with HHT based on clinical criteria and family history.

This case highlights the importance for a high index of suspicion for HHT in patients presenting with recurrent epistaxis and evidence of systemic AVMs. Prompt diagnosis and screening for visceral AVMs, particularly PAVMs, is essential for preventing serious neurological complications like cerebral abscesses thus significantly improving patient outcomes in this multisystemic disorder.

Keywords: Hereditary hemorrhagic telangiectasia; Vascular malformations; Arteriovenous malformations

Introduction

Hereditary haemorrhagic telangiectasia (HHT) is an autosomal dominant condition that is linked to a myriad of neurologic complications arising from vascular malformations of the brain, spinal cord and lungs. We present a 49-year-old female who came to our hospital outpatient department with

history of seizures and was found to have a brain abscess stemming from a pulmonary arteriovenous malformation (PAVM). PAVMs are associated with intracranial abscesses due to shunting and loss of the normal filtering effects of the lung capillary bed, predisposing patients with PAVMs to cerebral abscess. It is because of this association that it is imperative to treat the AVMs in patients found to have brain abscesses. In our case, embolization of pulmonary AVMs was done to minimize the risk of reinfection. PAVMs are often associated with HHT.

Case Presentation

A 49-year old female presented with history of 2 episodes of left sided focal seizures with secondary generalization that occurred 2 days ago. On examination we found right hemiparesis with predominant lower limb weakness, truncal ataxia and subtle right upper limb pronator drift which remained for a few hours. No facial or speech involvement was seen. She also gave history of repeated episodes of spontaneous intermittent, painless epistaxis since many years but denied history of haemoptysis.

On physical examination, she was hemodynamically stable. Initial investigations were unremarkable except for a white cell count of 15,000/µL. Renal and liver function tests were within normal limits. Blood culture was sent, which did not show any growth till 3rd day of incubation. A Magnetic Resonance Imaging (MRI) of the brain with contrast was performed in view of seizures. This showed a ring enhancing lesion in the left paramedian post central gyrus with surrounding perilesional edema **(Figure 1a-d)**. The central necrotic part showed restricted diffusion without any enhancement. Possibilities of Tuberculoma and cerebral abscess were suggested. She was asked to take antiepileptic drugs– Levetiracetam 500 mg twice a day and was started on intravenous ceftriaxone 2 gm twice a day.

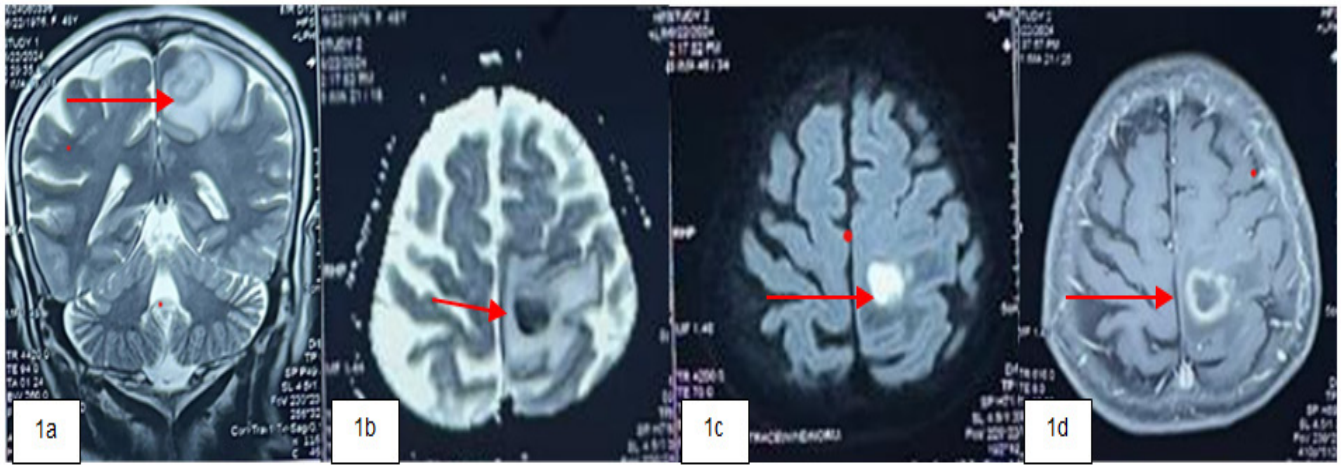

Figure 1: (A) - T2W coronal image showing a hyperintense lesion in the left paramedian post central gyrus with surrounding perilesional edema.
(B) & (C) - ADC image and its trace map showing restricted diffusion in the involved area.
(D) - Post contrast Axial image showing peripherally enhancing lesion.

A previous chest radiograph done a week back showed large subpleural oval shaped opacity in the right lower lobe. She underwent Positron Emission Tomography (PET – CT) scan in view of the suspicion of this being a neoplastic lesion.

On PET CT, the right lung showed weakly metabolic enhancing soft tissue lesion in the lower lobe measuring 33 x 25 mm and 23 x 18 mm & an enhancing pleural based lesion measuring 36 x 29 mm in the right middle lobe. The brain lesion was ametabolic, confirming it to be an abscess **(Figure 2)**.

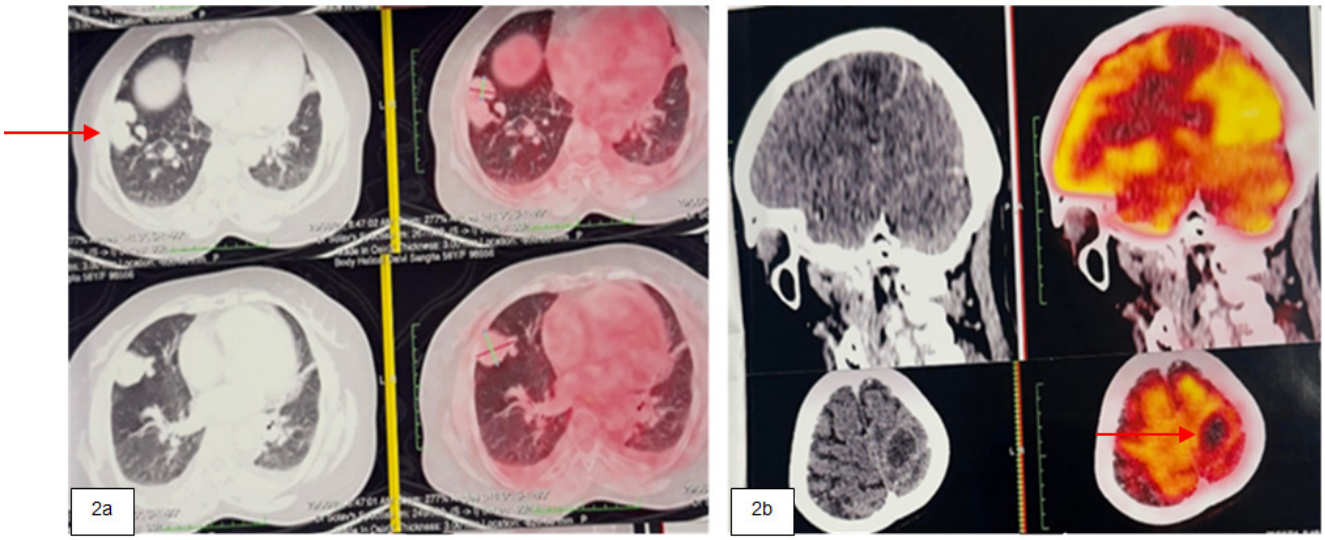

Figure 2: (A)- Axial PET CT image shows weakly metabolic enhancing soft tissue lesion in right lung lower lobe.
(B) - Brain lesion appears ametabolic, confirming it to be an abscess

A Computed Tomography (CT) Pulmonary angiogram **(Figure 3)** was suggested which confirmed the presence of 4 well defined pulmonary arteriovenous malformations, three in the right lung, of which 2 were in the lower lobe largest measuring 5.6 x 2.2 cm and one in the middle lobe measuring 2.3 x 2.5 cm and one in the left lung. All AVMs were having single arterial feeders (fistulous point). Two liver AVMs were also identified one each in the right and left lobes.

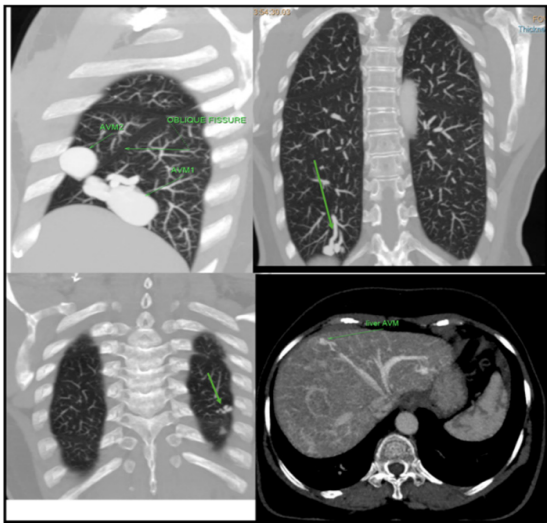

Figure 3: (A):- Reconstructed CT Sagittal image shows two pulmonary AVMs in the right lung; one in lower lobe and other in middle lobe.
(B):- Reconstructed CT coronal image shows third pulmonary AVM in the lower lobe of right lung.
(C):- Reconstructed CT coronal image shows fourth pulmonary AVM in the lower lobe of left lung.
(D):- Reconstructed CT axial image shows a hepatic AVM in the right lobe of the liver.

Conservative management of the brain abscess was planned with a 6-week course of ceftriaxone. The liver AVM was left untreated as the patient had no abdominal symptoms.

Endovascular embolization of the pulmonary AVMs was performed under general anaesthesia via a transfemoral approach.

Follow up MRI brain after a month showed reduction in size of the abscess.

In view of recurrent epistaxis, multiple pulmonary AVMs, liver AVMs, she was diagnosed as a case of HHT as her sister and another relative also had recurrent painless spontaneous epistaxis.

Discussion

This case report highlights the importance of recognizing Hereditary Haemorrhagic Telangiectasia (HHT) in patients presenting with seemingly isolated symptoms, even in resource-limited settings where comprehensive genetic testing may not be immediately feasible.

Our patient's history of spontaneous and recurrent epistaxis since childhood, also seen in her sibling, strongly suggested a syndromic diagnosis. The subsequent identification of arteriovenous malformations (AVMs) in both the liver and lungs further suggested a diagnosis of HHT.

A good clinical and family history, coupled with targeted imaging, can be pivotal in diagnosing HHT and guiding management.

HHT, also known as Osler-Weber-Rendu disease, is an autosomal dominant multisystem vascular disease with a prevalence of 1:5,000 to 1:10,000, which is associated with a myriad of primary and secondary neurologic complications[1,2]. The most common clinical manifestation is spontaneous and recurrent epistaxis beginning on an average at age 12 years of age[3], however the true burden of HHT lies in its potential for severe, often life-threatening, visceral complications. A diagnosis of HHT can be made by application of the Curaçao criteria[2] **(Table1)**.

Table 1: Summary of Highlighted Diagnostic and Surveillance Recommendations in Hereditary Hemorrhagic Telengiectasia[2].

Consideration	Recommendation
Diagnostic	• An underlying diagnosis of HHT should be considered in all persons with a PAVM. • An underlying diagnosis of HHT should be considered in persons meeting 2 or more Curaçao criteria' (1) Spontaneous and recurrent epistaxis; (2) Multiple mucocutaneous telangiectasias of the lips oral cavity, fingers or nose; (3) Visceral lesions (gastrointestinal telangiectasias or pulmonary/hepatic/cerebral/spinal AVMs); (4) A first-degree relative with definite HHT.
Targeted screening	• PAVM screening should be performed at the time of HTT diagnosis (adult/pediatric patients) and repeated at 5-y intervals (pediatric patients only)." • All adults with possible or definite HHT should receive MRI screening for brain vascular malformations at the time of diagnosis. • Spinal arteriovenous malformations are not routinely screened.

Abbreviations: HHT = hereditary hemorrhagic telangiectasia; PAVM = pulmonary arteriovenous malformation.

HHT is considered possible or suspected with 2 criteria presents, definite with 3 or more positive criteria1.

Primary neurological complications in HHT are predominantly secondary to cerebral AVMs (CAVMs), which affect nearly a quarter of individuals with HHT[4]. Given their prevalence and potential for catastrophic outcomes such as hemorrhage or seizures, expert guidelines unequivocally recommend routine brain MRI screening for all individuals with suspected or definite HHT. In contrast, spinal AVMs are considerably rarer and thus not routinely looked for[5].

PAVMs found in approximately 50% of HHT patients, represent direct shunts between pulmonary arteries and veins, bypassing the crucial capillary bed filtration and gas exchange6. This right-to-left shunt mechanism directly facilitates the paradoxical embolism of septic micro-emboli, bypassing the lungs' natural filtering capacity and allowing them direct access to the cerebral circulation.

Consequently, the risk of developing a cerebral abscess is significantly elevated in HHT patients with PAVMs, with an incidence as high as 6%[6].

While the causative organism in our patient's cerebral abscess could not be definitively identified, the strong association between PAVMs and cerebral abscesses in HHT entailed prompt intervention.

The management strategy in such cases is therefore two-fold: addressing the acute neurological insult and mitigating the future risk of recurrence. Embolization of the identified pulmonary AVMs in our patient was performed to minimize the risk of subsequent septic emboli and reinfection.

This case further reinforces a crucial diagnostic paradigm: the identification of PAVMs, even in isolation, should prompt a comprehensive evaluation for HHT. Given that nearly all PAVMs are attributable to HHT, screening for HHT is warranted in any patient found to have PAVMs, irrespective of other overt clinical manifestations[7]. The recommended first-line screening for PAVMs in patients with possible or confirmed HHT is transthoracic contrast echocardiography, with pediatric patients requiring repeat screening every five years[2].

Conclusion

This case highlights the need for a high index of suspicion for HHT in patients presenting with recurrent epistaxis, particularly when combined with evidence of systemic AVMs. Early diagnosis and screening for visceral AVMs, especially PAVMs, is important to prevent debilitating neurological complications like cerebral abscesses. Even in resource-constrained environments, a thorough clinical assessment can guide appropriate investigations and interventions, significantly improve patient outcomes and prevent life-threatening events associated with this multisystemic disorder.

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