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Case Report

Importance of Early Diagnosis of Hereditary Hemorrhagic Telangiectasia and Detection of Visceral Involvement Before Significant Clinical Complications Occur

Jeff Redleene Samuel[1], Heber Anandan[2], V M R Vairamuthuraja[3], Renuka Devi[4], Ravi Amal Raj[4]

[1]Senior Resident, Department of General Medicine, Tirunelveli Medical College Hospital, Tirunelveli, Tamil Nadu, India, [2]Senior Clinical Scientist, Department of Clinical Research, Dr. Agarwal's Healthcare Limited, Tamil Nadu, India, [3]Professor, Department of General Medicine, Tirunelveli Medical College Hospital, Tirunelveli, Tamil Nadu, India, [4]Assistant Professor, Department of General Medicine, Tirunelveli Medical College Hospital, Tirunelveli, Tamil Nadu, India

Abstract

Hereditary hemorrhagic telangiectasia is an autosomal dominant disorder that affects blood vessels throughout the body and results in a tendency for bleeding. The prognosis varies, depending on the severity of symptoms but generally, it is good when diagnosed early as bleeding is promptly recognized and adequately controlled. We report the case of a 45-year-old woman who developed telangiectasia over labial mucosa and ventral surface of the tongue, nasal septum, and multiple cherry angiomas over trunk was diagnosed as a case of hereditary hemorrhagic telangiectasia. She was successfully treated and prevented from developing further complications and followed up. Long-term systematic follow-up is indicated because known lesions may recur or progress and new manifestations of the syndrome may develop over time. Patients should be screened for pulmonary, hepatic, and central nervous system arteriovenous malformations (AVMs) at the time of diagnosis and at the onset of any suggestive symptoms and signs. Children of those with Hereditary hemorrhagic telangiectasia have a 50% chance of harboring the same mutation. Accordingly, pulmonary AVM screening and long-term follow-up are advocated.

Key words: Hereditary hemorrhagic telangiectasia (HHT), Arteriovenous malformations (AVMs), Recurrent epistaxis

INTRODUCTION

Hereditary hemorrhagic telangiectasia (HHT) manifested by mucocutaneous telangiectases and arteriovenous malformations (AVMs), a potential source of serious morbidity and mortality.[1] Lesions can affect the nasopharynx, central nervous system (CNS), lung, liver, and spleen as well as the urinary tract, urinary tract infections, conjunctiva, trunk, arms, and fingers.[2,3] Recurrent epistaxis is the most common presentation. ~90% manifest by the age of 40 years.[4] The reported case incidence is 1-2 cases per 1,00,000 populations annually. However, the prevalence may be underestimated because many cases may be asymptomatic. HHT occurs with equal frequency and severity in males and females.[5]

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CASE REPORT

A 45-year-old female patient was admitted to Tirunelveli Medical College Hospital with complaints of hemoptysis 2 episodes, breathlessness for 2 months, and epistaxis on and off since 2 years. The patient had regular menstrual cycles with normal flow. The patient gave a history of multiple skin lesions present in her mother. There was no similar illness in any other family members. On general examination, telangiectasia was present over labial mucosa and ventral surface of the tongue (Figure 1) and nasal

Corresponding Author: Dr. Heber Anandan, Senior Clinical Scientist, Dr. Agarwal's Eye Hospital, No. 10, South By-Pass Road, Vannarpettai, Tirunelveli - 627 003, Tamil Nadu, India. Phone: +91-9894067910. E-mail: clinicalresearch@drgarwal.com

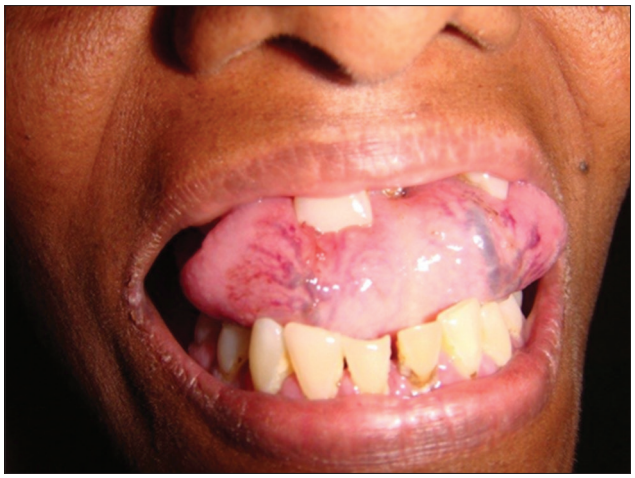

Figure 1: Telangiectasia was present over ventral surface of the tongue

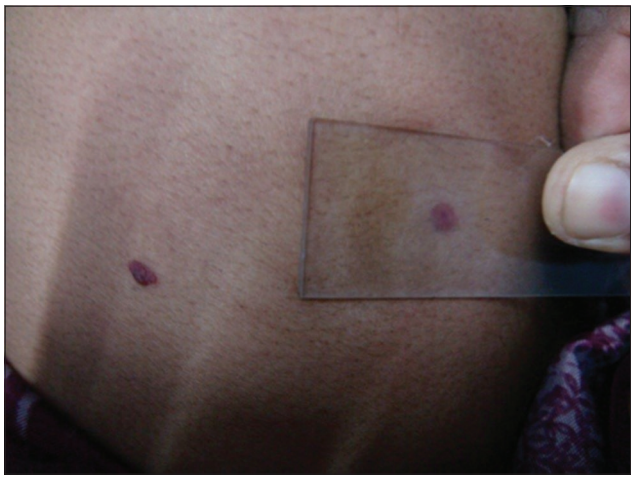

Figure 2: Cherry angiomas over trunk, which was blanching on pressure

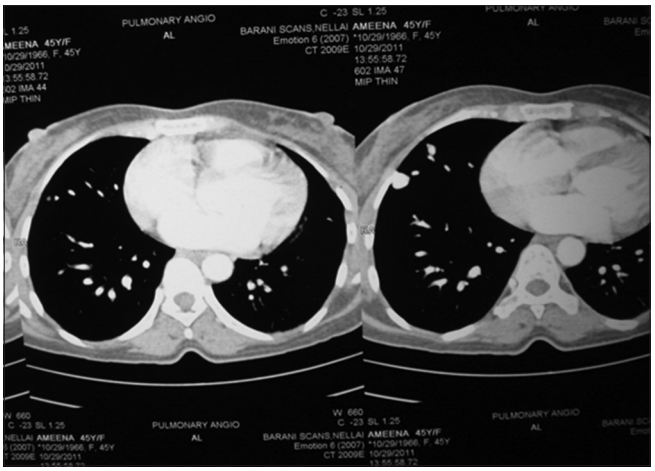

Figure 3: CT chest revealed- multiple lung nodules(AV malformation)

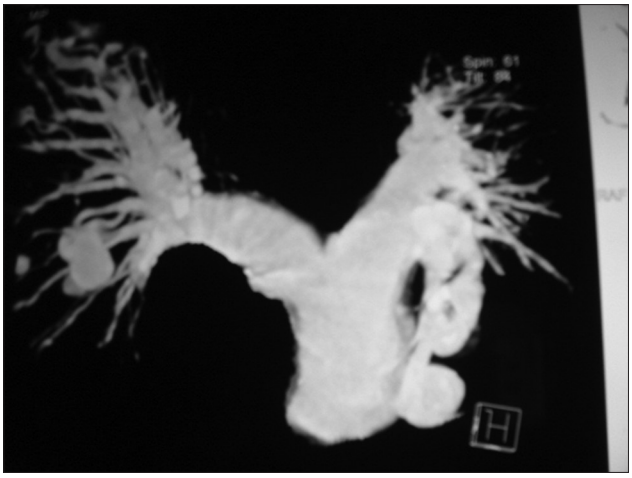

Figure 4: CT pulmonary angiography revealed multiple pulmonary AV malformations

septum. She had multiple cherry angiomas over trunk, which was blanching on pressure (Figure 2). She had pandigital clubbing. On auscultation, heart sounds were normal, and a bruit was heard over the right infrascapular area. Her vitals were stable, and other system examinations were normal. Her chest X-ray PA was normal. On further evaluating her, taking computed tomography (CT) chest revealed interesting findings (Figure 3) multiple lung nodules which were suggestive of AV malformation. So, we evaluated her by taking CT pulmonary angiography. Her CT pulmonary angiography (Figure 4) revealed multiple pulmonary AV malformations at least 12 with nidus of varying sizes in both lung fields supplied by segmental and subsegmental branches of pulmonary arteries and venous drainage through tributaries of pulmonary veins. Hence, we decided to take bubble echocardiogram which revealed small air bubbles in the left atrium.

DISCUSSION

Hereditary hemorrhagic telangiectasia is an autosomal dominant disorder manifested by mucocutaneous telangiectases and AVMs. Epistaxis is the most common manifestation. Onset of symptoms may be delayed until the fourth decade of life or later decades.[6-8] Diagnosis of HHT is made clinically by the Curacao criteria.[9]

1. Epistaxis - Spontaneous, recurrent nosebleeds
2. Telangiectases - Multiple at characteristic sites (lips, oral cavity, fingers, and nose)
3. Visceral lesions - Gastrointestinal (GI) telangiectasia,[10] pulmonary AVM,[11,12] hepatic AVM, cerebral AVM, and spinal AVM[13]
4. Family history - A first-degree relative with HHT

Definite diagnosis - 3 criteria

Possible diagnosis - 2 criteria
Unlikely - <2 criteria

Skin lesions are managed by topical agents, hypertonic saline sclerotherapy, and Laser ablation.[14] Pulmonary AVM is treated by embolization. GI bleeding is treated by aminocaproic acid[15] and endoscopic photoablation or electrocautery. central nervous system AVM is treated by Embol therapy.[16] Our patient gave the similar history in her mother, had episodes of epistaxis, had telangiectasia and her investigations revealed pulmonary AV malformations. Hence, she had all the criteria for definite diagnosis, and so our patient is a case of HHT.

CONCLUSION

This case illustrates the manifestations of HHT and its early diagnosis and management. Telangiectasia of skin and mucous membranes, epistaxis, and positive family history make up the classic triad of HHT. The onset of symptoms begins with epistaxis, continues with pulmonary AVMs, and proceeds to cutaneous and mucous telangiectases.[10] Screening with multiple imaging modalities is employed in HHT because of the prevalence of AVMs. Medical therapy and surgical treatment in patients with HHT are aimed at decreasing the amount of hemorrhage and minimizing the sequelae of AVMs and that necessitates the early diagnosis of HHT.

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