Hereditary Hemorrhagic Telangiectasia in a Sudanese Patient, A case Report

Fadi Toum Ahmed[1], Moh. Eljack[2], Mohammed Abdulkarim [3], and 4. Hiba Faroug Alamin Mohammed[1]

[1]Sinnar University Faculty of Medicine and Health Sciences
[2]Wad Medani center For heart disease and Surgery
[3]Alzaiem Alazhari University Faculty of Medicine

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Abstract

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Fadi M. Toum Ahmed11 Supervisor and first author Internal Medicine Specialist, Sinnar University fadi.mtoum2016@gmail.com , Moh.Mah.Fadelallah.Eljack22 Co-author MBBS, University of Bakht Alruda m.mahmmoud96@gmail.com, Mohamed Abdulkarim 33 Co-author and corresponding author Alzaiem Alazhari University, Khartoum-Sudan mohd.karim0095@gmail.com , Hiba Faroug Alamin Mohammed 44 Co-author Registrar of internal medicine Methag70@hotmail.com

Authors Affiliations and positions:

Fadi M. Toum Ahmed Supervisor and first author Internal Medicine Specialist, Sinnar University fadi.mtoum2016@gmail.com

Moh.Mah.Fadelallah.Eljack

Co-author and corresponding Author MBBS, University of Bakht Alruda Medical Doctor at Medani Heart Centre m.mahmmoud96@gmail.com

Mohamed Abdulkarim

Co-author Alzaiem Alazhari University, Khartoum-Sudan mohd.karim0095@gmail.com

Hiba Faroug Alamin Mohammed

Co-author Registrar of internal medicine Methag70@hotmail.com

Abstract

Hereditary hemorrhagic telangiectasia (HHT) is a rare disorder affecting skin and body's internal organs with tendency for bleeding. We reports a case of Sudanese 42-year-old with family history of HHT presented with recurrent epistaxis and telangiectasias.

Key Clinical Message:

Patients whom presents frequently with epistaxis or any other abnormal bleeding should be evaluated probably with digging deeply on their own and their family history .

Introduction

Hereditary hemorrhagic telangiectasia (HHT) (also known as Osler Weber Rendu syndrome (OWRS)) is a rare dominant autosomal disorder with a frequency of 6.1 to 12.1 per 100,000 (1). There are no differences between genders. It is clinically characterized by telangiectasia, recurrent epistaxis, visceral vascular lesions (arteriovenous malformations - AVMs). Usually a person with HHT has a family history of the disorder. This article reports a case that is clinically compatible with this rare entity.

Patient information

Our patient is a 42- year-old male who presented with a lifelong bleeding problem. On admission, he was concerned about an extensive bleeding through his nose (right nostril). His condition was diagnosed previously as idiopathic thrombocytopenic purpura (ITP) for which he is on steroids, osteoprotection, tranexamic acid with no improvement. He also takes PPI. He has 2 family members with the same bleeding problem including a brother and a sister. They were already diagnosed with HHT and passed away due to refractory HF, DCM, AF, and massive blood loss. He is not known to be have any allergies and has never received any drug that might interfere with blood homeostasis.

Clinical findings:

History:

This is not the first time that the patient has presented with such presentation with many episodes reported since childhood. He has initially been diagnosed with idiopathic thrombocytopenic purpura since 2007 based on presentation and investigations including complete blood count and bone marrow biopsy which showed thrombocytopenia at that time for which he was started on corticosteroids and osteoprotection with no improvement of his condition. He also takes proton pump inhibitors and tranexamic acid to help reduce the bleeding. He is not known to have any allergies, and has never received any drug that might be interfering with blood homeostasis.

Examination:

Upon examining his skin and mucus membrane multiple purpuric, punctuate, tiny macules in palms and fingertips of both hands were noted (figure 1). These macules also appeared on the oral and nasal mucosae as well as on the tongue (figure 2 and 3). His cardiac examinations revealed normal first and second heart sounds with pansystolic functional murmur. When examining his abdomen, his liver and spleen were palpable (10 and 6 cm below the costal margin, respectively) with normal other measures.

Timeline:

His condition has started since his childhood when he presented with same symptoms for which his labs showed thrombocytopenia (15 thousands/cmm) that the bone marrow sample has confirmed. Therefore, he was diagnosed as idiopathic thrombocytopenic purpura (ITP) for which steroids have been started with no improvement and he continued to have many bleeding episodes. He was admitted last time few months ago with large amounts of bleeding from his right nostril. A more detailed history and examination was intriguing for hereditary hemorrhagic telangiectasia. Based on the presence of epistaxis, telangiectasias and a first-degree relative with HHT confirmed using the Curacoa criteria.

Diagnostic assessment

Diagnostic work-up that has been done includes blood tests, Imaging and GI endoscopy. His blood tests showed anemia (Hb =6g/dl) along with normal WBCs and platelets. Upper GI endoscopy showed gastric and duodenal ulcers and his coloscopy showed hyperemic mucosa with severe inflammation and colonic polyps. He was diagnosed with ulcerative colitis and put on mesalazine and steroids for three months. However, no

telangiectasias or visible AVMs were found. Additionally, we found him to be HCV positive, and we owe that to the frequent blood transfusions which he received.

Treatment and follow-up

With regard to the treatment this patient received, it was mainly supportive in nature since no cure has been found till now. This patient was treated through nasal packing for his epistaxis along with tranexamic acid and blood transfusions. While we were not able to do contrast-enhanced pulmonary CT scan looking for pulmonary AVMs, it is usually managed with embolization. The patient was followed-up in the referred clinic for a period of 3 months with no significant complications apart from his epistaxis.

Discussion:

Hereditary hemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu disease is a rare autosomal dominant (AD) vascular disorder affecting the skin and body's internal organs in which there is an increased tendency for bleeding.

A consensus upon diagnostic testing is applied as Curacoa criteria which includes 2 criteria out of 4 for possible diagnosis and 3 out of 4 criteria for confirming the diagnosis. These criteria include recurrent episodes of epistaxis, telangiectasias in the skin and mucosal surfaces, arteriovenous malformations (AVMs) in visceral organs (lung, liver, brain and spine) as well as having a first-degree relative with confirmed disease using the same criteria (2).

There are not many cases reported from the African continent with a case of a 60-year-old African woman from Nairobi, Kenya has been described (3). We think that the reason might be that diagnosing such a condition can be challenging in low-resource countries such as Sudan in this case. Availability of complex investigation modalities away from big cities can be a big hinderance in terms of diagnostic approach to patients with suspected HHT.

In this article, we report a case of a 42-year-old Sudanese man who presented with a lifelong bleeding problem with recurrent epistaxis and known first-degree relative (a brother) with HHT diagnosed using the same Curacoa criteria who has died. He has multiple telangiectasias in his palm skin. His colonoscopy revealed no telangiectasia, though. Due to financial difficulties and availability investigations modalities, the patient was not able to do vascular imaging studies to look for AVMs in lungs, liver or brain. He was found to be positive for hepatitis C virus, which we suggest that it could be related to recurrent transfusions he received due to his anemia from chronic blood loss. While hepatomegaly has been reported in the case of the Kenyan woman, this patient has enlargement of not only his liver but also his spleen (10 and 6 cm below the costal margin, respectively) along with visible purpuric rash. We aim in this article to expand the literature about this disease in Africa.

Recurrent epistaxis is the single most common manifestation of HHT that is affecting up to nearly 90% of patients (4). It can be massive and progress to an emergency affecting the hemodynamic stability of patients. It is usually managed and controlled by nasal packing and tranexamic acid. Recurrent bleeding results in anemia that can be managed using blood transfusion and iron replacement therapies. While this patient has the predominant mucocutaneous manifestations, visceral organs involvement can lead to some serious complications including hypoxemia from secondary shunting of the blood due to pulmonary AVMs and possible paradoxical pulmonary embolism for the mechanism which was reported in the literature (4). Strokes, both ischemic and hemorrhagic have also been reported.

Ethical approval:

Ethical approval was obtained from Ethical committee at University of Sinnar and informed consent was taken from the patient for purposes of publications.

Consent:

Written informed consent was obtained from the patient parents for publication of this case report and accompanying images.

Conflict of interest:

The Authors Reports no conflict of interest

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