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# Delayed Diagnosis of Hereditary Hemorrhagic Telangiectasia

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**Summary:**

***Hereditary Hemorrhagic Telangiectasia (HHT), also known as Osler-Weber-Rendu Syndrome, is an autosomal dominant disorder. The typical findings of the disease are telangiectasias in skin and mucous membranes and arteriovenous malformations and aneurysms presenting in the organs like brain, lung, intestine and liver. The most common symptoms are recurrent epistaxis and gastrointestinal bleeding.***

***We report here a case of 38-year-old male who presented with typical history of recurrent epistaxis, hematemesis and melaena. Although the manifestations occurred since childhood , it took years to reach the diagnosis. Because most of clinician thought that upper gastrointestinal bleeding are usually due to peptic ulcer disease and chronic liver disease. The aim of this case report is not only to remind of this rare chronic disease but also to to increase the clinical suspicion for early diagnosis of HHT.***

***Key words: Epistaxis, Hematemesis, Melaena, Telangiectasia, Hereditary Haemorrhagic Telangiectasia.***

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## Introduction:

The Osler-Weber-Rendu syndrome or Hereditary Haemorrhagic Telangiectasia (HHT) is a rare systemic fibrovascular dysplasia which bears an alteration in the elastic and muscle layers of vessel walls, making them more liable to spontaneous ruptures and injuries[1,2] . 95% of affected individuals experience recurrent epistaxis, with a mean age of approximately 12 years and a mean frequency of 18 episodes per month.[3] The prevalence of intestinal telangiectasia varies from 10% to 33% [4]. They occur anywhere in the gastrointestinal tract, most commonly in the stomach and upper duodenum. Common causes of recurrent upper gastrointestinal bleeding are peptic ulcer disease and oesophageal varices. The aim of this case report is increase the suspicion of the clinician toearly diagnosis of HHT which is a rare cause of upper gastrointestinal bleeding.

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## Case Presentation:

A 38-year- old male patient was admitted into Dhaka Medical College hospital with recurrent epistaxsis for 33 years, haematemesis and melaena 1year. With these complaint, he went to different ENT specialists and underwent cauterization twice. He was admitted into several hospitals during each episode of melaena and received total 9 units of blood transfusions during last 4 months in addition to oral iron therapy on discharge each time. His father had similar history of epistaxis during his childhood and his 9-year old daughter has been suffering from epistaxis for about 5 years. On examination, patient was anaemic, with telengiectasic spots at oral mucosa of cheek and lower lip (figure1, 2).

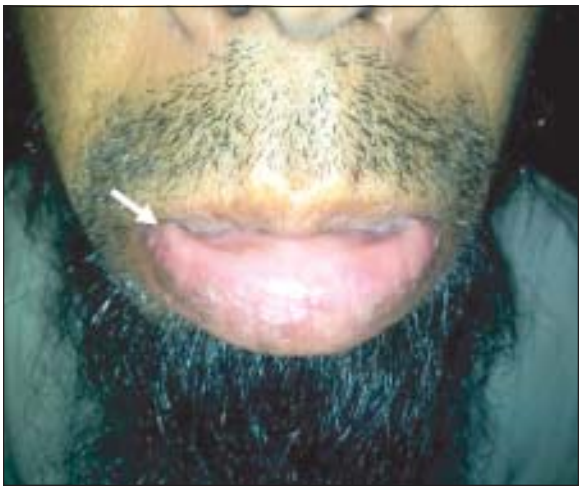

**Fig.-1:** *Telengiectasic spot over lower lip (white arrow)*

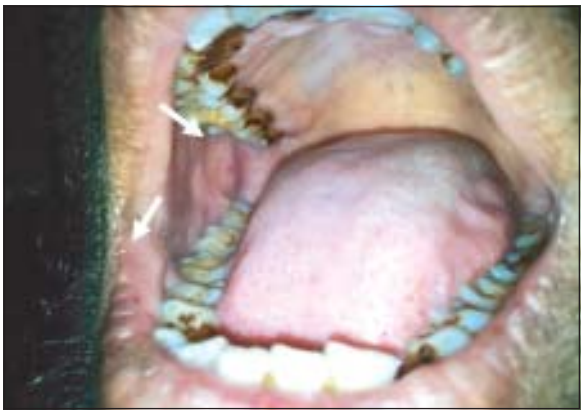

**Fig.-2:** *Telengiectasic spot over lower lip and oral mucosa (white arrows)*

No purpura, petechiae or ecchymosis were found. Laboratory investigation showed microcytic hypochromic anaemia with haemoglobin 8.8 gm/dl and Platelet count 4,13,000/cubic mm, and coagulation profiles were PT 18.5 seconds , APTT 33.5 seconds with a control of 31 seconds. Other blood biochemistry including liver function tests were within normal limit. Ultrasonogram of whole abdomen shows hepatomegaly with multiple ill-defined space occupying lesion (SOL) of variable sizes suggestive of vascular alterations .These structures correspond to dilated arterial branches and passive venous congestion in the liver. Upper GI Endoscopy showed a telagiectatic spot at pyloric antrum of the stomach and colonoscopy showed a telagiectatic spot in caecum (figure 3). We diagnosed the case as Hereditary Haemorrhagic Telangiectasia.

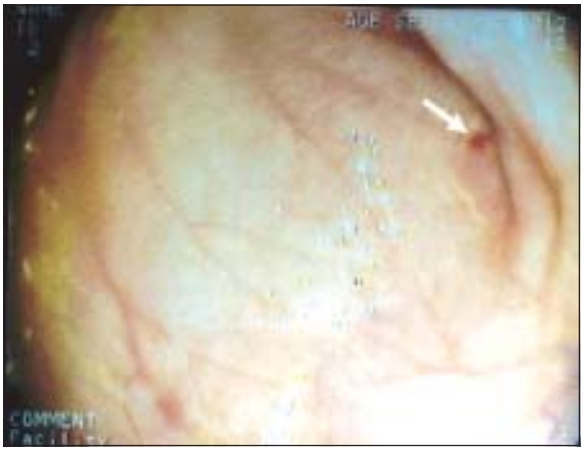

**Fig.-3:** *Telengiectasic spot in the caecum(white arrow)*

**Discussion:**

Diagnosis of Hereditary Hemorrhagic Telangiectasia is based on four main clinical features called Curacao criteria which were established in 1999 to improve and facilitate admission of individuals with Hereditary Hemorrhagic Telangiectasia . These features are as follows: spontaneous recurrent epistaxis (nosebleeds), mucocutaneous telangiectases (abnormal capillary connections that mostly appear on the skin and mucosa), visceral arteriovenous malformations (inadequate connection between arteries and veins in the liver, lungs, digestive system and brain) and an affected first degree relative as indication of autosomal dominant inheritance. Definite diagnosis of HHT is made if three of the four mentioned criteria are present. Hereditary Hemorrhagic Telangiectasia can be suspected if there are two positive criteria and if only one criteria is present, Hereditary Hemorrhagic Telangiectasia is considered to be unlikely [5] .

Almost all the criteria of HHT was present in our patient. But his patient was lately diagnose because of lack of suspicion by the physicians as well as rarity of this disease in our community. Epistaxis is the commonest presentation but commonly overlooked because of so many other common conditions causing epistaxis. Our patient underwent endoscopic examination previously without a definitive diagnosis as endoscopists probably failed to detect telangiectatic spot . It collapsed usually during active gastro intestinal bleeding. After correcting anaemia, telangiectatic spot becomes prominent or overt, so easy to detect during follow up endoscopy, that's why this case was missed before. Once detected it is better to cauterize in accessible areas.

Catastrophic haemorrhage occurs in the Hereditary Haemorrhagic Telangiectasia population due to pulmonary and cerebral AVMs. Asymptomatic screening for cerebral AVMs remains the subject of debate because of the risks of diagnostic and treatment modalities, and unclear natural history. The prevalence of liver involvement in hereditary hemorrhagic telangiectasia ranges from 8–30%, with more than half the patients being asymptomatic[6]. Doppler color flow study and CT scan of the abdomen are the confirmatory to see the hepatic involvement[7]. Though in our case hepatic involvement is present, we cannot confirm it due to financial support.

Regarding management, treatment of Hereditary Haemorrhagic Telangiectasia is symptomatic (it deals with the symptoms rather than the disease itself), as there is no therapy that stops the development of telangiectasias and AVMs directly. Episodes of severe bleeding are treated with endoscopic argon plasma coagulation (APC) or laser treatment of any lesions identified; this may reduce the need for supportive treatment[8]. During active severe bleeding mainstay of treatment is blood transfusion as happened in our case. After blood transfusion because of severe gastro intestinal bleeding, re-endoscopy was done which detected multiple telangiectatic spots in our patient. He is in need of cauterization of telangiectatic spots .

**Conclusion:**

Always upper gastrointestinal bleeding is not due to peptic ulcer disease and chronic liver disease .Though HHT is a rare disease, it may be considered if bleeding is recurrent and from different sites. So, any unexplained and recurrent bleeding from a particular site must be taken seriously. Screening is required for the family members in case of HHT. Proper education of patients, family members and more importantly medical practitioners is required in order to diagnose and proper management of the patients with HHT.

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