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CASE REPORT

OSLER – WEBER – RENDU SYNDROME: A RARE CASE OF UPPER GASTROINTESTINAL BLEEDING

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ABSTRACT

BACKGROUND: Osler-Weber-Rendu syndrome (Hereditary hemorrhagic telengiectasia) cases present with recurrent epistaxis, gastrointestinal bleeding (hematemesis, melena), and arteriovenous malformations involving almost all organs of body. Internal gastrointestinal bleeding can occur from arteriovenous (telengiectasia) from stomach or small bowel.

CASE PRESENTATION: We present a case of Osler-Weber-Rendu syndrome who presented with recurrent epistaxis, melena and gross anemia (hemoglobin 3.5 gm/dL). He was admitted and transfused red cell concentrates along with intravenous fluids. His investigations including oesophagogastroscopy, colonoscopy were normal. His Computed tomography mesenteric angiography revealed 02 sites of telangiectasias. The telangiectasias were embolized and coiled resulting in successful control of melena and symptomatic improvement.

CONCLUSION: Osler-Weber-Rendu syndrome can present with gross anemia in emergency. Their management can be worrisome and challenging for both clinicians and cases of Osler-Weber-Rendu syndrome.

KEYWORDS: Telangiectasia, Hereditary Hemorrhagic (MeSH); Gastrointestinal (Non-MeSH); Melena (MeSH); Telangiectasias (MeSH)

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INTRODUCTION

Osler-Weber-Rendu syndrome also known as Hereditary hemorrhagic telengiectasia (HHT) is a rare autosomal familial disease which presents with recurrent epistaxis, gastrointestinal bleeding and abnormal structure of blood vessels.[1] It was first reported in 19th century.[2] Osler, Hanes and Weber brought this rare but important syndrome to light; naming it as Osler-Weber-Rendu syndrome.[3] The typical combination of recurrent epistaxis, gastrointestinal bleeds and characteristic abnormal/malformed blood vessels (called telangiectasias) on lips, nasal mucosa, oral cavity mucosa and multiple organs with iron deficiency anemia are its diagnostic features.[4] This syndrome is diagnosed with help of Curacao criteria. The diagnosis is "definite" when 3 or more of the following criteria is fulfilled and when 2 of the criteria are present, the diagnosis is "Probable"; epistaxis, telangiectasias, visceral vascular malformations and presence of first degree relative with Hereditary hemorrhagic telangiectasia.[4] This disease has autosomal dominant inheritance. Around 20% cases have no family history.[5] The incidence of HHT is reported 1-2/100,000.[6] Treatment is mainly supportive. Most common presentation is recurrent epistaxis, followed by gastrointestinal bleeding which occurs in 13-30% of HHT cases.[4]

We are presenting this case report of a man who presented with iron deficiency anemia and later reported in emergency department with upper gastrointestinal bleeding, to better understand the rare disease presentation and its management.

CASE DESCRIPTION

A 49 years man reported to outpatient department with recurrent epistaxis and melena for many years off and on. He had been visiting various physicians and hospitals for these complaints. He had been treated for recurrent epistaxis with chemical and electrical cauterization multiple times but the problem persisted. Similarly, melena was also treated by various physicians with proton pump inhibitors and symptomatic therapy on multiple occasions. On examination he was pale and he had telangiectasias on nasal mucosa and tip of fingers. His elder sister had similar complaints who died at age of 64. His blood pressure (BP) was 90/60 mmHg, his pulse was 131/minute, he was afebrile and respiratory rate was 25/minute. Rest of examination was unremarkable. He was admitted and all base line investigations were advised urgent. His hemoglobin level was 3.5 mg/dL. He was shifted to intensive Care unit (ICU) and managed with immediate blood transfusion (02 RCCs), intravenous fluids, proton pump inhibitors (intravenous omeprazole) and hemodynamic care under strict monitoring of vital signs. Rest of investigations including liver function test (LFTs), renal function test and prothrombin time (PT) and partial thromboplastin time with kaolin (PTTK) were normal as shown in Table 1. Considering his disease and episodes of melena; diagnosis of bleeding from gastrointestinal telangiectasias was made. Next morning, his oesophagogastroscopy was carried out which showed a small hiatus hernia with no evidence of source of bleeding. Subsequently, his colonoscopy, CT mesenteric angiography and RBC labeled scan were done which were all normal. Patient's hemoglobin was built and he was discharged after the complaints settled. He reported again after 2 weeks with similar episodes of melena. He was readmitted and he underwent same cascade of investigations as before. His oesophagogastroscopy and colonoscopy came out to be normal again. However, this time CT

mesenteric angiography (CTA) revealed a site of telangiectasias (arteriovenous malformation) as shown in Fig 1. He was diagnosed as a case of HHT upon history, examination, investigation findings and family history of a sibling with similar complaints. Keeping in view the findings of CT mesenteric angiography, embolization of involved telangiectasias and coiling was planned and carried out successfully. After the procedure, he was observed for 02 days in hospital. He symptomatically improved and was discharged with weekly followup in outpatient department. He was advised to avoid nose picking and to be cautious about any kind of trauma to his lesions on body parts as they could cause excessive bleeding. He was referred to ENT department for recurrent epistaxis where he underwent nasal surgery (cauterization and septoplasty). Till date, he is alright and had no episode of melena or profuse nasal bleed and he is maintaining his Hb.

TABLE 1:LABORATORY INVESTIGATIONS OF STUDY SUBJECT

Laboratory Test	Normal Range	Patient Result
Hemoglobin (mg/dL)	12 – 18	3.5
Total Leucocyte Count (/mm^3)	4 – 11 × 10^3	11.1 × 10^3
Platelets Count (/mm^3)	150 - 400 × 10^3	410 × 10^3
Serum Bilirubin (umol/L)	0 - 17	39
Serum Alanine Transaminase (IU/L)	< 42	21
Serum Alkaline Phosphatase (U/L)	< 279	88
Serum Urea (mmol/L)	1.7 – 8.3	3.3
Serum Creatinine (umol/L)	53 - 105	75
Serum Sodium (mmol/L)	135 - 145	141
Serum Potassium (mmol/L)	3.5 - 5.1	4.6
Blood Sugar Random (mg/dL)	< 140	116
D-dimers (ng/ml)	0 - 500	406
LDH – Lactate dehydrogenase (U/L)	0 - 480	212
ESR (mm at end of 01 hr)	0 - 15	20
Prothrombin Time (seconds)	11 - 13	12
Partial Thromboplastin Time with Kaolin (PTTK) (seconds)	30 - 40	34

DISCUSSION

HHT also called Osler-Weber-Rendu syndrome occurs commonly due to three genetic mutations; ENG (HHT1), ACVRL (HHT2) and SMAD4.[7] Gastrointestinal bleeding is the second commonest clinical presentation in HHT cases after recurrent epistaxis. Gastrointestinal bleeding starts usually after 50 years of age in HHT patients.[4] Canzonieri et al has described 22 cases of HHT, who had undergone gastroduodenoscopy and colonoscopy. They showed that duodenum was most common site of involvement.[7] Proctor et al performed endoscopy in 27 HHT cases and showed presence of gastric, deodenal and jejunal lesions in these cases.[8]

HHT cases are anemic because of recurrent bleeding from nose and gastrointestinal bleeds which lead to iron deficiency anemia. Adequate management includes treating anemia, replenishing iron depletion either orally or intravenously and blood transfusion in excessive bleeds.[9] Oral tranexamic acid also decreases need for transfusion and gastrointestinal bleeding episodes. Important task of a gastroenterologist is to identify the site of bleeding and control it effectively. Argon plasma laser is very a very effective latest modality for this purpose.[10] Long acting somatostatin analogues have also shown to reduce gastrointestinal bleeding episodes and requirement of transfusions.[11]

As far as nasal bleeding is concerned, in majority of cases these are occasional and few have significant major episodes.[1] So, majority of patients with hereditary hemorrhagic telangiectasias require no treatment except oral iron therapy and reassurance. In severe cases, many need nasal packing or surgical procedures including cauterization, septodermoplasty etc.[1]

CONCLUSION

Osler-Weber-Rendu syndrome can present with gross anemia in emergency. Anemia can result from recurrent nasal bleeding and gastrointestinal bleeding in form of hematemesis and melena. Their management can be worrisome and

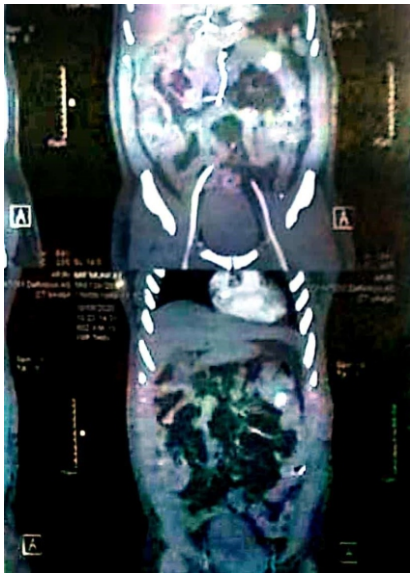

Figure 1: Contrast Enhanced Computed Tomogram (CECT) & Computed T omograghy Angiography (CTA) of abdomen showing mesenteric arteriovenous malformation

challenging for both clinicians and cases of Osler-Weber-Rendu syndrome.

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AUTHORS' CONTRIBUTION

Following authors have made substantial contributions to the manuscript as under:

FS: Identification, diagnosis and management of case, drafting the manuscript, approval of the final version to be published

MAK & SA: Identification of case, drafting the manuscript, critical review, approval of the final version to be published

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

CONFLICT OF INTEREST

Authors declared no conflict of interest

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DATA SHARING STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request