Case Report

Thrombasthenia — A Rare Cause of Upper Gastrointestinal Haemorrhage

H J DE SILVA[1], U DE SILVA[2], A MANAWASINGHE*[3]

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Summary

We report a woman with thrombasthenia, first presenting at the age of 60 years. This case also highlights the importance of perforrming haemostatic screening tests for patients with upper gastrointestinal haemorrhage in whom upper gastrointestinal endoscopy is negative.

Introduction

Bleeding disorders with qualitative platelet defects comprise a complex group of abnormalities. Thrombasthenia, though rare, is one of the most clearly defined of this group. The accepeted diagnostic criteria are: excessive haemorrhage with a prolonged bleeding time but normal coagulation time, a normal platelet count, failure of platelet aggregation by adenosine diphosphate (ADP), and absent or deficient clot retraction[1]. There is also failure of platelet aggregation by adrenalin[2]. Bleeding is commonly cutaneous or mucous, visceral haemorrhage being rare[2,3]. The haemorrhagic manifestations usually start in early childhood[2], and the only effective treatment consists of administration of fresh whole blood, platelet rich plasma or platelet concentrates[2,3]. The disease is inherited as an autosomal recessive trait[2,3,4].

1 *Senior Lecturer*
2 *Registrar*
3 *Registrar*
*Department of Medicine and *Pathology, Faculty of Medicine, University of Peradeniya.*

Case Report

The patient, a 60 year old woman presented to our unit with haematemesis and melaena of 2 days duration. This was the fifth episode of upper gastrointestinal (GI) bleeding in the last 3 months. Prior to that, she had not had any bleeding from the upper GI or from elsewhere. The four previous episodes had been investigated at another hospital with a barium meal and three upper GI endoscopies, all of which had been negative. A diagnosis of 'possible gastric erosions' had been made, and she was treated with cimetidine and antacids. The patient had no epigastric pain, anorexia, nausea, symptoms suggestive of gastro-oesophageal reflux or weight loss. There was no past history of a bleeding tendency or excessive blood loss. She denied use of steroids, aspirin, non steroidal anti inflammatory drugs or alcohol intake. There was no past history of hepatitis, and despite careful questioning there was no family history of a bleeding diathesis.

On examination she was very pale, had a regular, small volume pulse of 110 per minute, and the BP was 90/70mmHg. The rest of the clinical examination was normal; especially, she was not icteric, had no lymphadenopathy, did not have any evidence of bleeding into the skin, and her liver and spleen were not palpable.

The haemoglobin was 4 g/dl and haematocrit 28%. After resuscitation, an upper GI endoscopy was perfomed about 36 hours after admission to hospital. Apart from pallor of the upper GI mucosa, the endoscopy was entirely normal. The following investigations

were perfomed; Bleeding time 16 minutes (normal 2-5), whole blood clotting time 8 minutes (normal 5-11), prothrombin time 12 seconds (control 12 seconds), Partial thromboplastin time in Kaolin 83 seconds (normal < 100 seconds), white cell count 8.2 x 10^9/1 with a normal differential count, platelet count 310 x 10^9/1, ESR 18mm in the first hour, blood film showed no abnormal white cells, bone marrow aspiration showed an active marrow with reduced iron stores and normal megakaryocytes with active platelet budding, blood urea level, serum proteins (including electrophoresis), and liver function tests including an ultrasound scan of the liver were normal. Because the bleeding time was prolonged, tests of platelet aggregaton were performed. Platelets from the patient and one normal control were obtained in the following manner[5]; first platelet rich plasma was obtained by centrifuging 2ml of venous blood in EDTA at 1500 r.p.m. for 15 minutes. The platelet rich plasma was then separated and centrifuged again at 3000 r.p.m. for 15 minutes. Most of the supernatant was discarded and the button of platelets resuspended. To study platelet aggregation[6], either 0.1 ml of ADP (10μ g/ml) or 0.1ml of 1 in 10,000 adernalin was added to 0.2ml of the platelet suspensions in test tubes placed in a water-bath at 37°C, and mixed well. After 2 minutes a drop of each sample was transferred onto a glass slide and examined microscopically for the presence of platelet aggregation. The patients' patelets did not aggegate in the presence of either ADP or adernalin, while the control platelets aggregated in the presence of both substances. Finally, to assess clot retraction, 5ml of venous blood was obtained from the patient and the control into calibrated plain glass tubes. In the sample obtained from the patient, clot retraction was absent at one hour. On these findings, a diagnosis of thrombasthenia was made.

Since then, the patient has been readmitted to hospital more than 20 times with upper GI haemorrhage, once with spontaneous bruising in addition to the GI bleeding. Each of the episodes were treated with fresh blood and platelet rich plasma transfusions.

Discussion

Our patient fulfilled the criteria which are necessary to diagnose thrombasthenia[1]. This is the second report of this condition in the Sri Lankan medical literature. The first was that of an 11 year old boy and his 5 year old sister presenting with bleeding gums, purpura and epistaxis[5]. Our patient denied a family history of a bleeding disorder, which is sometimes the case diseases inherited by an autosomal recessive trait. Futhermore, acquired causes of defective platelet function such as liver disease, uraemia and dysproteinaemia[7] have been excluded in our patient with reasonble confidence. What is unusual about our case is the fact that the patient first manifested the haemorrhagic tendency as late as 60 years of age. We could not find any reports in the literature of such a delay in presentation of the disease. Thrombasthenia usually manifests itself in early childhood[2]. It may be possible that the patient did have minor bleeding previously, which had gone unnoticed.

The other reason for reporting this patient is to emphasize the importance of perfoming haemostatic screening tests for patients with upper gastrointestinal haemorrhage in whom upper gastrointestinal endoscopy is negative. Had this been done, several unnecessary investigations and inappropriate drug treatment could have been avoided in our patient.

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