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CASE REPORT

A case of factor XI deficiency in a patient undergoing an oesophagogastrectomy

Natasha Parrott[1]

Leigh Kelliher[1]

Author details can be found at the end of this article

Correspondence to:
Natasha Parrott;
tashy13@hotmail.co.uk

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Introduction

Factor XI deficiency (variously known as haemophilia C or Rosenthal syndrome) is a rare disorder of the clotting cascade which increases the risk of bleeding, particularly after surgical procedures or trauma. Therefore, it is important that clinicians involved in the treatment of these patients are aware of the potential risks and take measures to mitigate them, especially in the context of major surgery. Factor XI deficiency can occur in patients of all races, but is more common among Ashkenazi Jews (Bolton-Maggs, 2000). This case report discusses a patient with factor XI deficiency who underwent an oesophagogastrectomy for oesophageal carcinoma. He required treatment with factor XI concentrate before induction, as well as intraoperative antifibrinolytic therapy, to reduce his risk of perioperative bleeding.

Discussion

Factor XI deficiency, also known as haemophilia C or Rosenthal syndrome, may be a congenital (autosomal) or, more rarely, acquired condition. It is the most common of the

Case report

A 75-year-old man with a recent diagnosis of factor XI deficiency required an oesophagogastrectomy for oesophageal adenocarcinoma. During the course of investigation for his cancer, he was found to have a slightly prolonged activated partial thromboplastin time ratio of 1.2, with an otherwise normal coagulation profile. Further tests revealed a low factor XI level of 35 iu/dl (normal range 69–129 iu/dl). He had no personal or family history of abnormal bleeding, no Jewish heritage, and had previously undergone dental surgery without incident. During the work-up for his oesophagogastrectomy he had also undergone an uncomplicated staging laparoscopy. Given this history, his age of presentation and the new discovery of cancer, the working diagnosis was an acquired factor XI deficiency. There was no other significant past medical history, and preoperative cardiopulmonary exercise testing showed good baseline function with an anaerobic threshold of 16.1.

He was referred to a tertiary haematology centre and a plan was made for the perioperative management of his condition during his oesophagogastrectomy. As he was about to undergo major surgery, with the potential for significant blood loss, it was recommended that he receive factor XI concentrate plus antifibrinolytic cover. His preoperative haemoglobin level was 135 g/litre, so no intravenous iron or blood transfusions were required for optimisation. Platelet count was within the normal range. Factor XI concentrate is not routinely available at all sites and arrangements were made to have this supplied from a tertiary centre.

On the morning of surgery 1000 units of factor XI concentrate were given as an intravenous infusion. Anaesthesia was then induced with remifentanil and propofol target-controlled infusions plus rocuronium. He was intubated with a left-sided double lumen endotracheal tube. A thoracic epidural was inserted, arterial and central venous access gained and a urinary catheter placed. Tranexamic acid 1 g was given on induction and a subsequent two doses given at 8 hours. Blood pressure was maintained using goal-directed fluid therapy and a phenylephrine infusion that continued into the postoperative period. The estimated blood loss was 420 ml. His haemoglobin level at the end of surgery was 132 g/litre. He remained metabolically stable throughout.

He was extubated and transferred to the intensive care unit for ongoing vasopressor support and monitoring. Tranexamic acid was continued 8-hourly for 4 days postoperatively. At no stage did he develop bleeding complications or require transfusion of blood products. He was treated as per the local enhanced recovery pathway and was discharged home after a total of 14 days.

Learning points

- When preparing a patient for major surgery, abnormalities of coagulation should be thoroughly investigated. Factor XI deficiency, while rare, may be an acquired condition and its diagnosis is not precluded by a history of no bleeding problems.
- In cases of unexplained or unexpected haemorrhage following major cancer surgery, factor XI deficiency should be on the list of differential diagnoses.
- This is the first case report of a patient with factor XI deficiency undergoing an oesophagogastrectomy and demonstrates how, with careful planning and multidisciplinary teamworking, it is possible to safely perform such a high-risk procedure in this context.

rare bleeding disorders, estimated to affect 1 in 1 000 000 people in the general population (Batty et al, 2015). Although factor XI is required for normal coagulation pathways, the association with factor XI levels, indicated by the activated partial thromboplastin time and bleeding tendency, is weak. Generally the bleeding tendency is variable and often does not correlate with the factor XI levels (Wheeler and Gailani, 2016). It is not purely recessive as heterozygous carriers may present with severe bleeding (Bolton-Maggs, 2000).

As mentioned, factor XI deficiency is usually an inherited condition caused by a variant in the F11 gene. Acquired factor XI deficiency is rare and reportedly caused by the development of autoantibodies in patients with autoimmune disease such as systemic lupus erythematosus or cancer. While it may be picked up during preoperative screening or during investigation of unexplained bleeding, these cases more commonly present with abnormal bleeding after trauma or surgery.

Potential therapies include fresh frozen plasma, factor XI concentrate, fibrin glue, desmopressin and antifibrinolytics such as tranexamic acid (Batty et al, 2015).

Oesophagogastrectomy necessitates surgical resection around major blood vessels – including the aorta and pulmonary vessels – and the heart and as such has the potential for significant bleeding. Major haemorrhage during this procedure is associated with increased morbidity and mortality. It is therefore important to optimise patients' coagulation perioperatively and take all measures to mitigate this risk. This case highlights the importance of thorough preoperative assessment and screening in high-risk surgery. The only abnormality identified was a mildly prolonged activated partial thromboplastin time ratio, something that could easily have been overlooked with catastrophic consequences. However, through close multidisciplinary working and effective perioperative planning it was possible to perform this procedure safely.

Author details

[1]Department of Anaesthetics, Royal Surrey County Hospital, Guildford, UK

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