Bleeding Risk Management in a Patient with Familial Macrothrombocythopenia associated with Receptor GpIIb/IIIa Deficit

Tavares F¹; Pereira C¹; Marinheiro L¹; Domingues S¹

¹Centro Hospitalar Tâmega e Sousa, EPE
Portugal

Introduction

Inherited platelet disorders are rare and their perioperative management may be challenging. We report a case of a patient with a syndrome recently described - Familial Macrothrombocythopenia associated with Receptor GpIIb/IIIa Deficit.

Clinical Case

- 22-year-old female
- Diagnosis of Familial Macrothrombocythopenia associated with Receptor GpIIb/IIIa Deficit
- Past medical history: frequent episodes of abundant epistaxis, menorrhagia, bleeding time prolonged with direct trauma and easy bruising
- Preoperative studies: isolated thrombocytopenia (92,000 platelets/microL) and a mean platelet volume of 12.3 fL
- Preoperative clotting screen (PT, APTT, INR): within the normal range.

Discussion

Familial Macrothrombocythopenia associated with Receptor GpIIb/IIIa Deficit is a recently described syndrome. Beyond quantitative platelet disorder it implies some degree of platelet impaired function. Bleeding risk is not yet quantified and may be highly variable during surgery. To the extent of our knowledge no cases have been reported of patients with this syndrome submitted to surgery. Therefore the anaesthetic management of the potential bleeding risk of this syndrome is not well known.

Learning Points

Anti-fibrinolytics and desmopressin may be useful in the perioperative management of patients with Familial Macrothrombocythopenia with Receptor GpIIb/IIIa Deficit submitted to surgery.

References

- 1 - Balduini CL, Savoia A. Genetics of familial forms of thrombocytopenia. Hum Genet (2012) 131:1821–1832.
- 2 - Nurden AT, Nurden P. Inherited disorders of platelet function: selected updates. J Thromb Haemost 2015; 13 (Suppl. 1): S2–S9.