Perioperative Management of Coagulopathy in Lowe Syndrome

A 33 year old male with Lowe Syndrome, end stage renal disease receiving hemodialysis twice per week suffered a left hip femoral neck fracture. Repaired with a left hemiarthroplasty and complicated by immediate post operative hematoma. No history of bleeding.

3 weeks post op he had persistent hematoma formation requiring surgical evacuation, no source of bleeding was found intraoperatively. Branches of the superior gluteal artery and were embolized although active bleeding was not visualized on angiography.

5 weeks from initial surgery, he had recurrent hematoma with another surgical evacuation. Hematology was consulted for suspected bleeding diathesis.

Final hematoma evacuation managed with periop antifibrinolytics, cryoprecipitate, desmopressin

Post op initiation of oral tranexamic acid 1300mg BID with no recurrence of hematoma at 10 weeks from initial surgical intervention.

• Surgeons should be aware of the increased bleeding risk in LS patients and consider early consultation with hematology and use of antifibrinolytics, cryoprecipitate and desmopressin in the perioperative period to reduce the risk of hemorrhage.

• Delayed hemorrhage more consistent with disorder of primary hemostasis

• Initial work up of coagulopathy: assess for uremia, platelet levels, PT, PTT testing, mixing study to assess factor levels or presence of factor inhibitor.

• To assess primary hemostasis assess platelet function with PFA-100 and fibrinogen levels, assess function of fibrinogen by thrombin clotting time and reptilasetime in setting of heparin use.

• Optimize platelet function with hemodialysis

• Lowe Syndrome may cause disorders in primary hemostasis due to abnormal platelet function and even dysfibrinogenemia, as is the case in our patient, leading to delayed perioperative hemorrhage and hematoma. Mechanism is unknown.

INTRODUCTION

Lowe syndrome (LS) is an exceedingly rare X linked disorder characterized by congenital bilateral cataracts, renal Fanconi syndrome and motor and intellectual delays that result from a mutation in the oculocerebrorenal gene (OCRL).

OCRL encodes a phosphatidylinositol 5-phosphatase which includes a Rho GTPase activating protein that is central to vesicular trafficking and cytoskeleton development on the cellular level all vital to platelet function.

Procedures that pose risk of hemorrhage such as cataract surgery, creation of arteriovenous fistula and orthopedic surgeries are common and have led to the discovery of a primary hemostasis disorder in these patients.

CASE PRESENTATION

LEARNING OBJECTIVES

• Recognize the clinical features and pathogenesis of Lowe Syndrome

• Evaluate for and recognize clinical features of primary & secondary hemostasis disorders in perioperative period

• Understand management of post operative hemorrhage in primary hemostasis disorders

REFERENCES