Introduction:
Malignant infantile osteopetrosis (MIOP) or “marble bone disease” is a rare autosomal recessive disorder of increased bone density due to defects in osteoclasts ability to reabsorb and remodel bone. See Table 1 for MIOP characteristics (1). An infant with MIOP presented for emergent craniosynostosis surgery, which resulted in a sudden massive blood loss, hemodynamic instability, hypocalcemia and QT prolongation.

Case Report:
A 9 month-old, 5.6 kg infant was admitted with FTT & hypotonia. A diagnosis of MIOP with severe multiple suture craniosynostosis (causing increased ICP & papilledema) was made. He was urgently scheduled for cranial expansion surgery. Preoperatively, he was optimized in the ICU by the hematology, metabolic, and pulmonary services. Preoperative anemia (Hct 20) and thrombocytopenia (Plt 80) were treated; Hct 31.4 & plt count 190. Coagulation profile was WNL. The patient was consented & enrolled in a randomized, double-blind, placebo-controlled clinical trial of the antifibrinolytic, Tranexamic Acid (TXA) (2). He was a difficult intubation due to limited neck extension & mandibular hypoplasia. After cranial dissection, the intraoperative course was complicated by sudden excessive bleeding from the abnormal, spongy, vascular bone. One blood volume was lost in 30 minutes requiring immediate volume resuscitation with PRBC’s & vasopressors. There were several brief episodes of hypotension, hypocarbia and desaturation. The study was immediately unblinded & upon discovery of placebo administration, TXA was started at bolus of 50mg/kg/15 min followed by an infusion at 5mg/kg/h as per protocol at the time.

Table 1: Characteristics of MIOP:
- Craniosynostosis
- Difficult intubation (mandibular hypoplasia & bony overgrowth)
- Reduced bone marrow volume
- Restrictive lung disease
- Pathological fractures
- Hypocalcemia
- Hematological abnormalities (anemia, thrombocytopenia, leukocyte & macrophage dysfunction)

Total products administered were PRBC 182 mL/kg, platelets 32 mL/kg, FFP 40 mL/kg, 2U cryoprecipitate, and 5% albumin 90 mL/kg. Postoperative course was complicated by prolonged intubation, respiratory compromise & enterobacter bacteremia. After several months in the hospital, the parents decided to place him in palliative care. Unfortunately, he later died at home from respiratory complications.

Discussion:
Anesthesia management of hemostasis in MIOP patients undergoing craniosynostosis repair should focus on a multimodal patient blood management strategy which includes maintaining hemodynamic stability, resuscitation with blood & specific blood products & control of intraoperative bleeding. The administration of Tranexamic acid, in pediatric craniosynostosis surgery can significantly reduce surgical blood loss & transfusion requirements (2). Point of care tests such as TEG may aid in the diagnosis & treatment of coagulopathy. Despite careful management, perioperative mortality & morbidity remain high.