

CM-AVM SYNDROME AND SEVERE HEMOPHILIA B IN AN INFANT: CASE REPORT AND STAGED TREATMENT

Karen Chen, MD

Background: Hemophilia B is an X-linked deficiency in factor IX affecting 1 in 25,000 males. Most patients with severe Hemophilia B (levels <1%) manifest clinically at the time of circumcision; the remaining present by age two with sporadic or excessive hemorrhage. Early diagnosis and treatment are essential for prophylaxis and preoperative planning.

Clinical Presentation: We report a 5-month old male with a fast-flow intracranial arteriovenous malformation (AVM) in addition to a RASA 1 mutation and cutaneous capillary lesions, known as Capillary Malformation Arteriovenous Malformation Syndrome (CM-AVM). He developed symptomatic hydrocephalus at 3 months of age, and severe hemophilia B was incidentally discovered in his work up for treatment planning. In consultation with a Pediatric Hematologist, a tunneled Broviac catheter was placed with perioperative factor IX concentrate dosing. Because patients with severe deficiency may produce antibodies to exogenous factor IX, kinetic post-treatment studies were performed to assess for factor inhibition. He then received his first embolization therapy with perioperative factor IX concentrate dosing followed by prolonged manual pressure at the puncture site without adverse event. A second treatment is scheduled. The treatment of a high flow AVM in a patient with CM-AVM and severe hemophilia B to our knowledge has not yet been reported.

Conclusion: This case describes the feasibility and safety of endovascular embolization on a fast-flow intracranial AVM with perioperative Factor IX concentrate replacement.