# Intracranial hemorrhage in an infant with severe Factor V Deficiency

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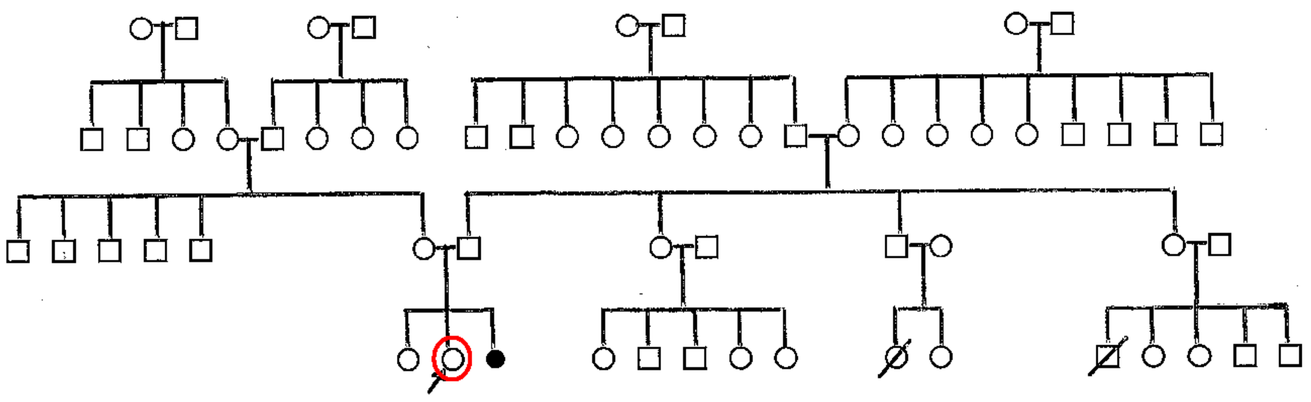
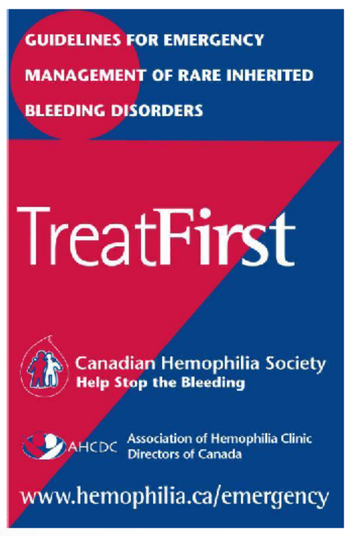
## Case presentation – A 5 month old with epistaxis

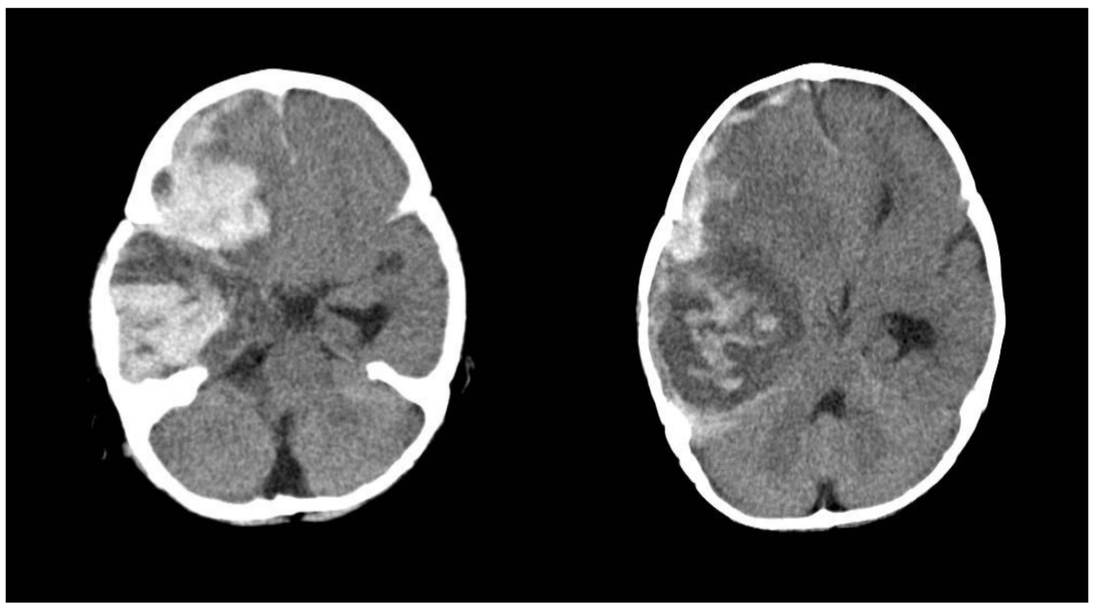
Our patient was an otherwise healthy 4-month-old infant from a remote rural community who presented with recurrent epistaxis resulting in clinically significant anemia. The activated partial thromboplastin time (aPTT) and international normalized ratio (INR) were markedly elevated at 158 seconds and 2.4, respectively. She was referred to the Pediatric Hematology program for further evaluation, where a Factor V level of <0.01 IU/mL was identified. The family history (shown in the pedigree below) did not identify relatives with significant bleeding, and Factor V levels are normal in both parents (Mom: 0.61 IU/mL, Dad: 0.6 IU/mL). Following the diagnosis, the family was educated to be alert to the early signs of bleeding, particularly intracranial hemorrhage, and was provided with a Treat First information card to present to the local hospital if she required emergency evaluation and care.






## Acute deterioration with intracranial hemorrhage

Two weeks after diagnosis, the patient began to experience increasing irritability and decreased activity. She was taken to the local Emergency Department on three occasions and discharged with a diagnosis of viral illness. On the fourth visit, she presented with a decreased level of consciousness, and after stabilization, including Fresh Frozen Plasma (FFP) and transfer to our tertiary care center, neuro-imaging demonstrated intracranial hemorrhage. The patient underwent emergency neurosurgical decompression under coverage with FFP. Regular transfusions of FFP continued daily for the first five days post-operatively to maintain near normal aPTT and INR.



## Long-term management and education

Based on a single case report[1] of intracranial hemorrhage in another infant with Factor V deficiency, prophylaxis with FFP, three times weekly, was initiated and will continue for the foreseeable future. No clinically significant hemorrhage has occurred since the initiation of prophylaxis. Factor V genotyping revealed homozygosity for a novel missense mutation (p.Cys2193Tyr) located three amino acid positions away from the C-terminus of Factor V.

Factor V deficiency is a rare congenital bleeding disorder. In Canada, 50 cases have been registered in the Canadian Rare Inherited Bleeding Disorders Registry; six are classified as severe. A major challenge for patients with rare bleeding disorders is the lack of awareness among medical personnel. Despite her caregiver's persistence and the information card we had provided, early signs and symptoms of intracranial hemorrhage were not appreciated. Education for parents and professionals is a crucial component of the therapeutic plan for patients with rare bleeding disorders.

[1]Salooja N, Martin P, Khair K, Liesner R, Hann I. Severe Factor V deficiency and neonatal intracranial haemorrhage: a case report. *Haemophilia* 2000: 6, 44-46.

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