Haemophilia B case report
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INTRODUCTION Haemophilia B (Christmas disease) is a rare genetic coagulopathy. It is caused by
deficiency of coagulation factor IX due to inherited mutation of gene located on X chromosome.
Disease commonly affects male population and its presentation depends on severity of clotting protein
shortage. It is usually diagnosed in early age and manifests itself with easy bruising and delayed after-
trauma bleeding in joints, muscle or other tissues.

CASE REPORT One-year-old boy was admitted into emergency unit for bleeding from mouth cavity
and febrile state. He was born on term, hypotonic with parietal cephalohaematoma and Apgar score 9/9.
In first few months, he had suffered intraventricular haemorrhage (IVH) grade II and his tests had shown
prolonged activated partial thromboplastin time (aPTT). At admission he became somnolent with poor
reflexes (GCS 10), irritable and crying. He had sunset eyes sign and his heart rate intermittently
decreased (75 bpm). Pupils were isochoric and normally reactive. Emergency head CT showed
intracranial suprasellar and prepontine haemorrhage, and hydrocephalus. CT angiography showed no
vascular abnormalities. External ventricular drain was implanted. Heteroanamnesis revealed that patient
had slight head trauma one day before hospitalization. He had knee bruises and his post-vaccination
bleedings were long-lasting. Blood tests showed increased thrombocyte count and prolonged aPTT. He
had a low concentration of factor IX and haemophilia B was diagnosed.

DISCUSSION AND CONCLUSION Bleeding diatheses are due to vascular, platelet or coagulation
factor defects. Although rare, haemophilia B should be considered as cause of bleeding in differential
diagnosis.