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.