

## Acquired Haemophilia A in pregnancy: A case report

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### ABSTRACT

**Introduction:** Haemophilia is a genetic disorder caused by a mutation in one of the genes responsible for blood clotting factors production. There are several types of haemophilia, the most common ones being Haemophilia A and Haemophilia B. Haemophilia A is caused by a mutation in the F8 gene, which is responsible for producing a protein called factor VIII. These clotting factors are essential for the coagulation cascade. In rare cases, haemophilia can occur due to spontaneous genetic mutations. Acquired haemophilia can affect reproductive-aged women during pregnancy and the postpartum period. Patients with acquired factor VIII deficiency, experience severe or life-threatening bleeding episodes, even in the absence of a previous bleeding predisposition. **Case Presentation:** A 26-year-old, gravida 3 para 2 initially presented with sudden onset of iliopsoas hematoma at 6 weeks of pregnancy. A series of investigations was performed and confirmed the diagnosis of acquired haemophilia. She was referred to our tertiary hospital for delivery. **Discussion:** The cause of acquired factor VIII deficiency during pregnancy is unclear, but it is speculated to be related to the complex immunological changes that occur during pregnancy. Sensitization of the mother's immune system to fetal factor VIII during previous pregnancies has been suggested as a possible cause, although inhibitors can also develop in the first pregnancy. Determining the severity of bleeding in patients with factor inhibitors based solely on titre levels is challenging, as inhibitors to factor VIII are cleared in a non-linear manner, underestimating bleeding risk in some cases.

## 46 XY partial gonadal dysgenesis with gonadoblastoma and dysgerminoma following laparoscopy prophylactic bilateral gonadectomy: A case report

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### ABSTRACT

**Introduction:** 46 XY partial gonadal dysgenesis is a disorder categorized under differences in sex development (DSD) which occurs following abnormal gonadal development. There is a disproportion between one's genotype, phenotype, and gonadal development which influences the wide range of presentation and clinical appearance of 46 XY females. **Case Description:** This is a case of a 46 XY female who presented with primary amenorrhea and delayed puberty at the age of 18 years old. She has ambiguous genitalia with both breast and pubic hair at Tanner stage 2. Transabdominal ultrasound found a small uterus with a vagina but was unable to locate the gonads. MRI abdomen and pelvis showed a 20 mm right gonad located extra-pelvis near the right iliac vessels while the 9 mm streak left gonad was at the normal location next to the sigmoid colon. All her tumour markers were normal except LDH (332). She was given estrogen therapy for pubertal induction and underwent laparoscopy prophylactic bilateral gonadectomy after three years of postponement due to financial restrictions and the COVID-19 pandemic. The intraoperative findings were similar to the MRI findings and the histopathology results showed left gonadoblastoma and right dysgerminoma FIGO stage 1A. She did not require any further surgery or adjuvant chemotherapy based on a discussion with the gynae-oncology team. She continued taking estrogen therapy until she had withdrawal bleeding and oral progesterone was added. **Discussion:** 46 XY DSDs are at high risk of developing germ cell tumour and require prophylactic gonadectomy as soon as the diagnosis is established. However, delay in presentation and surgery may affect the outcomes and prognosis.