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A multidisciplinary approach on managing Haemophilia A patient: A case report

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ABSTRACT

Introduction: Hemophilia is an inherited disorder and is caused by the deficiency of clotting factors in the blood. Both Hemophilia A and B are inherited via an X-linked recessive pattern, therefore Haemophiliac carrier mothers have a 50% chance of having affected males and a 50% chance of having carrier females. **Case Description:** We describe the case of a 32-year-old, female Haemophilia carrier in her first pregnancy. With strong family history of Haemophilia A in the wife's family with her father, paternal uncles and maternal cousins affected, the couple was well informed, empowered and determined to seek prenatal diagnosis for their fetus. We performed focus genome examination for genetic mutation testing to identify pathogenic gene in this patient. This then allowed prenatal diagnosis for Haemophilia A to be carried out with amniocentesis and results showed that the male fetus did not carry the gene mutation. Pregnancy was monitored as per protocol and a healthy baby boy was delivered. **Discussion:** Recent advancement in genome testing has helped to diagnose the fetus free of genetic mutation and was reassuring to the parents. Prenatal diagnosis has enabled parents to make informed decision and prepared them mentally as well as to what to expect. The importance of multidisciplinary approach including ethics consultation cannot be stressed enough as there was concern regarding quality of life should the fetus be Haemophiliac A, with possible issue of possible termination of pregnancy. We believed this was a first prenatal diagnosis for Haemophilia A in a fetus in Malaysia.

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Post-menopausal lady with huge symptomatic uterine leiomyoma: A case report

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ABSTRACT

Introduction: Uterine leiomyoma is the most common benign gynaecological condition that arises from the overgrowth of smooth muscle and connective tissue in the uterus. It is estimated to be present in 20-40 % of women over the age of 30. The prevalence increases during the reproductive age and decreases after menopause. **Case Description:** We describe a case of a 51-year-old nulliparous, who attained menopause three years prior to presentation. She presented with progressive distension of the abdomen associated with weight loss of 11 kg in a year. The Computed Tomography of Thorax, Abdomen, and Pelvis reported as the uterus and both ovaries are not visualized and large ill-defined heterogeneous enhancing mass with cystic regions noted within the central abdomen measuring 23.2 x 30.0 x 26.7 cm (AP x W x H) suggesting a mass of ovarian origin. The ultrasound-guided biopsy and histopathological examination of the mass confirmed benign leiomyoma. Total abdominal hysterectomy and bilateral salphingo-oophorectomy was done. The histological report of the post-operative specimen confirmed the diagnosis of uterine leiomyoma. **Discussion:** The exact cause of uterine leiomyoma is not clearly understood. It is estrogen-dependent and associated with low parity, obesity, family history of a first-degree relative and reproductive age. In this case, the patient is nulliparous but not obese and in a state of menopause. Therefore, the huge symptomatic uterine leiomyoma is a rare occurrence after menopause. The size of the mass at presentation posed a diagnostic dilemma because of the possibility of intra-abdominal malignancy.