Swyer syndrome with dysgerminoma: A case report

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ABSTRACT

Introduction: Swyer syndrome is a 46, XY karyotype but phenotypically female condition with primordial Mullerian structures, and it usually presents with primary amenorrhea in outpatient settings. The risk of gonadal neoplasia is high, necessitating early prophylactic removal of these dysgenetic gonads. Approximately 5% of dysgerminomas have been reported to be associated with Swyer syndrome. **Case Description:** We present a case of a 15-year-old girl diagnosed with Swyer syndrome associated with left ovarian dysgerminoma. She sought assistance for primary amenorrhea at the gynaecological clinic of Hospital Tuanku Fauziah, Kangar. Physical examination revealed that her secondary sexual characteristics were at Tanner stage 3. The abdominal examination noted the presence of a vague pelvic mass. There were no facial dysmorphisms or signs of hyperandrogenism. An ultrasound showed a solid pelvic mass with a small uterus. Her tumour marker CA 125 was elevated, and hormonal profiling revealed hypergonadotrophic hypogonadism with XY karyotyping. Primary surgery was performed, and histopathological examination reported a left ovarian dysgerminoma at FIGO stage 1A. **Discussion:** Swyer syndrome belongs to a group of pure gonadal dysgenesis disorders. Early diagnosis is crucial as prophylactic gonadectomy in these cases reduces the risk of developing germ cell tumours..

Ovotesticular disorder of sex development with 46 XY – Rare and out of the ordinary

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ABSTRACT

Introduction: Ovotesticular disorders of sex development (ovotesticular DSD) is a very rare condition that an individual has both ovarian and testicular tissue. It is among the rarest disorders of sex development in humans. **Case Description:** We would like to share a case of ovotesticular DSD patient who presented to Hospital Taiping with the chief complaint of primary amenorrhea. There was no abdominal mass palpable during examination and she had normal female external genitalia. Abdominal ultrasound was not able to visualise a uterus or adnexal masses but able to visualise a vaginal plate. Abdomen and pelvic MRI reported the absence of uterus with hypoplastic ovaries and a blind end vaginal plate. Biochemical investigations showed there was raised FSH and estradiol. Karyotyping was reported 46, XY. Tumour markers were normal. Diagnostic laparoscopy and bilateral gonadectomy were performed. Intra-operatively, the absence of a uterus with the presence of bilateral gonads was noted. Her post-operative recovery was uneventful. Histopathology reported the presence of unencapsulated nodules composed of immature infantile seminiferous tubules, with Sertoli cells and Leydig cells in between the tubules. The report also showed ovarian parenchyma but no cellular atypia or malignancy. **Discussion:** The presence of both ovarian and testicular tissue in an individual with a karyotype of 46 XY is very rare even among the cases of ovotesticular DSD. A multidisciplinary team should be involved in the management of patients with ovotesticular DSD.