SWYER SYNDROME WITH A GERM CELL TUMOUR

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ABSTRACT

Objective: A 23-year-old patient with primary amenorrhea and female external phenotype presented to the outpatient department of Mercy Teaching Hospital, Peshawar. On clinical examination, she was tall with under developed secondary sexual characteristics. Laboratory/ radiological investigations revealed Swyer syndrome. Karyotyping showed XY chromosomal pattern. On imaging, a left huge adnexal mass, 16x12cm with multiple solid and cystic areas, small infantile uterus and right streak ovary was detected. Alkaline phosphatase, LH and FSH levels were raised. Laparotomy followed by left salpingoophrectomy was done. Intra-operatively, there was a small infantile uterus and right streak ovary and right normal looking fallopian tube with no ascites, visceromegaly and enlarged abdominal lymph nodes. Histopathology revealed dysgerminoma.

Conclusion: Swyer syndrome also known as pure gonadal dysgenesis is a rare form of genetic disorder. Clinically, the patient has female external phenotype and XY chromosomal pattern. Early investigations and radiological imaging help in the proper diagnosis. Prophylactic gonadectomy reduces the risk of developing germ cell tumors.

Key words: Swyer syndrome, Germ cell tumors, Pure gonadal dysgenesis.

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CASE REPORT

A 23-year-old patient presented to Mercy teaching hospital, Peshawar with female external phenotype, primary amenorrhea and pain lower abdomen. She had no sexual problems, weight loss or altered bowel habits. Her family history was not significant. She was 70 kg, 6-feets, 2 inches, breasts developed Tanner 3, no axillary hair and pubic hair Tanner 1, normal vaginal length, small cervix, huge mass, regular margins, firm and limited mobility with no visceromegaly, ascites or enlarged lymph nodes.

Pelvic ultrasound revealed large solid and cystic mass inseparable from small infantile uterus. On CT abdomen, there was 3.3 ×1.6 cm size uterus and a hypo-dense 16.9 x 12x 11 cm mass with solid and cystic areas, reaching up to the level of L-5 vertebra. There was loss of surrounding structures interface. The mass was displacing gut loops with no local and regional lymphadenopathy noted. Ca-125, Beta HCG, Alpha feto-protein, testosterone and LDH levels were within normal range. Alkaline phosphatase was 410 U/L, while LH/FSH levels were raised (35.4 IU/L, FSH 104.5 IU/L). Ultrasound guided trucut biopsy revealed dysgerminoma. Intra-operative findings showed 16 x 12 cm left adnexal mass, multiple solid and cystic areas, irregular margins and adherent left fallopian tube, small infantile uterus, right streak ovary and normal looking fallopian tube. Left salpingoophrectomy was done and histopathology report revealed a poorly differentiated neoplasm composed of nests and sheets of neoplastic cells, brisk mitotic activity with ovarian capsule, omentum and vascular invasion.

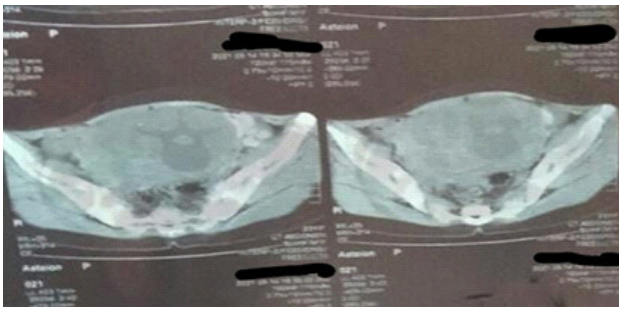

Fig 1: CT scan showing a huge hypo dense mass.

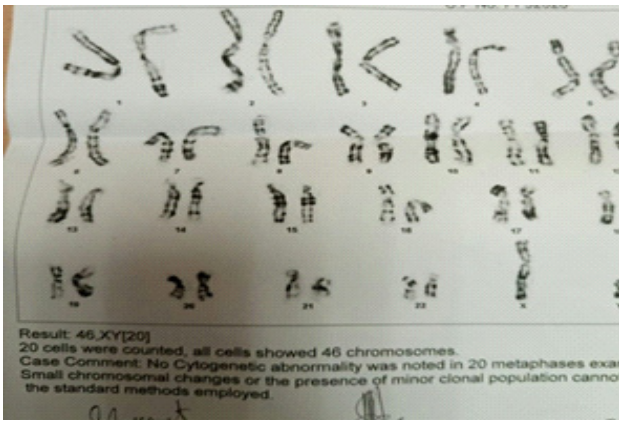

Fig 2: Karyotyping revealed a genotype of 46 XY.

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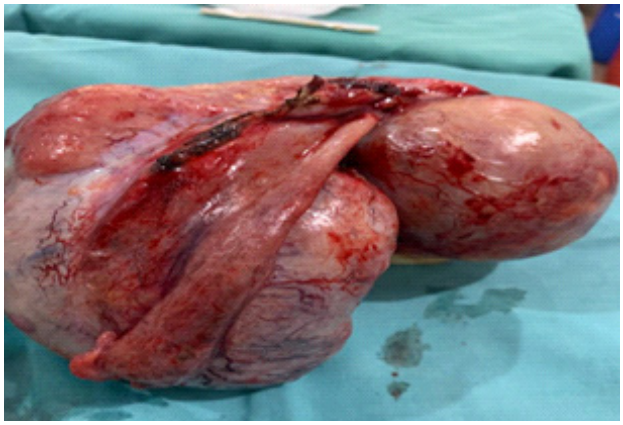

Fig 3: Left ovarian mass with adherent left fallopian tube.

DISCUSSION

Swyer syndrome is a very rare disorder of failure of sex organs development, first described in 1955 by Jim Swyer. [5] The incidence is 1 in 100,000. [2] Patients have XY chromosomal pattern, born as females, and present with a history of primary amenorrhea. [5,6] They have normal looking external genitalia, failure of development of secondary sexual characteristics, small infantile uterus, and streak gonads.

There may be molecular and genetic abnormalities (mutations) in different genes affecting SRY function, required for testicular formation. Some cases are not inherited and in some cases, exact cause remains unknown.[7] These patients have 46XY karyotype . The other differential diagnosis like Mayer–Rokitansky–Küster–Hauser syndrome, (XX) Turner syndrome, Congenital Androgenic Insensitivity Syndrome (XY) needs to be excluded.

These patients are at a high risk of developing germ cell tumors like gonadoblastomas and/or dysgerminomas due to presence of Y chromosome. The chance of Dysgerminoma to be malignant is high. [8,9] Treatment includes gonadectomy or pelvic clearance followed by chemotherapy in case of malignancy. Hormone replacement therapy may be considered for development of secondary sexual characteristics. These patients are infertile but may become pregnant by using donated eggs. Proper evaluation early diagnosis, counselling and psychological support is essential in the management of such patients. Prophylactic gonadectomy reduces the risk of developing germ cell tumor.

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