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Case Report

The hidden uterus in Swyer syndrome with gonadoblastoma: a diagnostic dilemma

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ABSTRACT

Disorders of sex development (DSD) comprise a rare group of disorders in which genotype and phenotype are discordant. In 46 XY DSD, also known as Swyer syndrome, patients present a complete female phenotype and the 46 XY karyotype is usually identified during investigation for primary amenorrhea and/or delayed puberty. The risk of developing a gonadal tumor can be as high as 30%-40%. A 27-year-old phenotypically female with the complaints of primary amenorrhea with a pelvic mass presented to our setup. The case was approached applying the role of clinical, radiological and laboratory analyses. The final diagnosis of Swyer syndrome with gonadoblastoma was made. Karyotyping was done which revealed 46 XY set of chromosomes. Her FSH and LH levels were found to be elevated. Patient was explained about streak gonads. After surgical resection of tumor and gonadectomy, HRT was started. Primary amenorrhea is a common diagnostic challenge, wherein there are numerous causes that need to be approached in a systematic manner. However, when a case with a pelvic lump or a solid pelvic mass presents to a clinician, the approach becomes difficult to justify amenorrhea and pelvic mass as a single entity.

Keywords: DSD, Swyer syndrome, Gonadoblastoma, Pure gonadal dysgenesis

INTRODUCTION

Pure gonadal dysgenesis is a condition with a normal set of chromosomes, that is, 46 XX or 46 XY, and the latter is better known as Swyer syndrome.[1] Condition usually presents as primary amenorrhea due to the fact that gonads have no hormonal or reproductive potential. Incidence of the condition being 1 in 100,000 whereas its combination with gonadoblastoma is rarer and reported to be present in 5% of the cases.[2] Owing to the condition, lack of secretion of Anti-Mullerian hormone leads to the normal development of mullerian structures. The cause for lack of AMH has been detected as mutation in the DNA-binding region of the SRY gene as depicted in Figure 1 in 10%-20% of cases.[3] The risk of developing a gonadal tumor can be as high as 30%-40% and can occur during childhood or adolescence. However, in majority of the cases, cause for the condition is still unknown.[4]

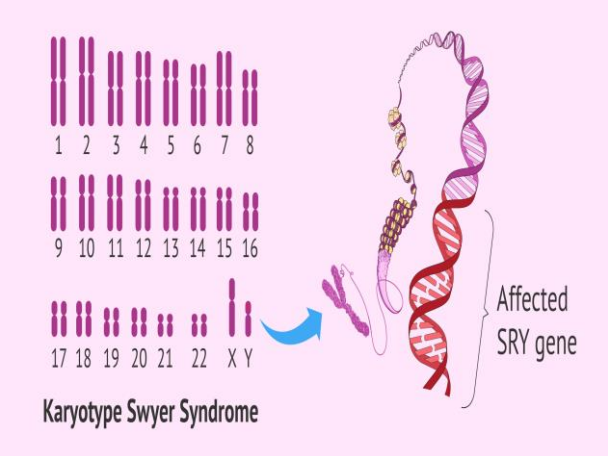

Figure 1: Mutation in the DNA-binding region of the SRY gene in 10%-20% of cases.

CASE REPORT

A 27-year-old, married female presented with the complaints of primary amenorrhea, pelvic pain, and a feeling of lump on the right side of the lower abdomen and abdominal distension for the last 4 months. The background knowledge of the patient revealed that she had no uterus based on the previous workup by a local clinician. The clinical examination showed the female phenotype with relatively underdeveloped breasts (Tanner stage III), sparse pubic hair, and normal external female genitalia. The urine pregnancy test was negative. She revealed the history of hormonal pill intake since past 5 months as prescribed to her by the local clinician.

On examination, patient was hemodynamically stable. She weighed 64 kg and her height was 164 cm. She did not have increased facial hair or acne. On per abdomen examination, she had 22-24 weeks size hard fixed abdominal lump, mobile, non-tender. No inguinal mass was palpable. No lymph nodes palpable. On local examination, external genitalia was normal. On per speculum examination, cervix and vagina were found to be healthy. Examining bimanually, 22-24 weeks size mass was felt; cervix was deviated to left side; uterus could not be felt separately and right side fornix was obliterated.

USG and MRI were done which revealed hypoplastic uterus and cervix with vagina. Bilateral ovaries were not visualised separately. Solid mass lesion was noted arising from right adnexa with multiple internal septations, calcific foci, hemorrhage; features suggestive of solid abdomino-pelvic mass arising from right adnexa.

Her LH=300 and FSH=150 mIU/ml values were too high resembling pseudomenopause, with serum estradiol=10 pg/ml and Prolactin, TSH levels were within normal limits. Her serum testosterone levels (0.45 ng/dl) were found to be low. Patient was advised karyotyping which revealed 46 XY pattern genotype. Her tumor markers such as serum beta HCG, LDH, AFP and CA-125 were within normal limits. Based on the clinical, biochemical and radiological findings, a provisional diagnosis of pure gonadal dysgenesis with gonadal tumour was made.

Patient and her family were counselled regarding disorder of gonadal dysgenesis with probability of tumor and need for exploratory staging laparotomy with gonadectomy and preservation of uterus for reproductive function with assissted reproductive technique. She was explained regarding post-operative need for Hormone replacement therapy; its risks and benefits.

With due consent of patient and family, patient underwent hysteroscopy followed by exploratory laparotomy and surgical staging. Hysteroscopy findings revealed long cervix with tubular uterine cavity and thin endometrium and bilateral ostia seen. On laparotomy, grossly the mass was seen arising from right adnexa, 15×15 cm solid, cystic with disrupted capsule (Figure 2). Uterus was measuring-3×3×1 cm with right ovary and tube not seen separately from the mass and left sided streak gonadal tissue appearing to be ovary. Infra-colic omentectomy was done and biopsy from rectal, bladder and uterine surfaces was sent for histopathological examination as shown in Figure 3. However, no tumour deposits were noted. Right-sided tumor and the left-sided streak gonad were extirpated with peritoneal wash cytology. The tumor margins were freed from the adjacent structures. Postoperative combination of estrogen and progesterone was prescribed with regular follow-up. HPE showed the right-sided gonadoblastoma and the left-sided streak gonad of ovarian origin. All other organs, including omentum, were free of tumour.

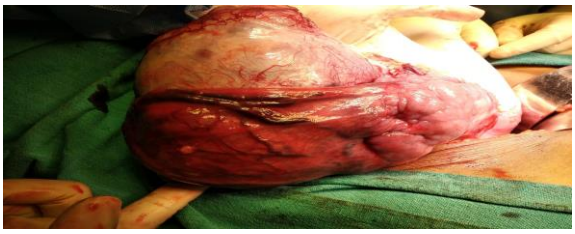

Figure 2: Gross appearance of gonadoblastoma as seen intra-operatively.

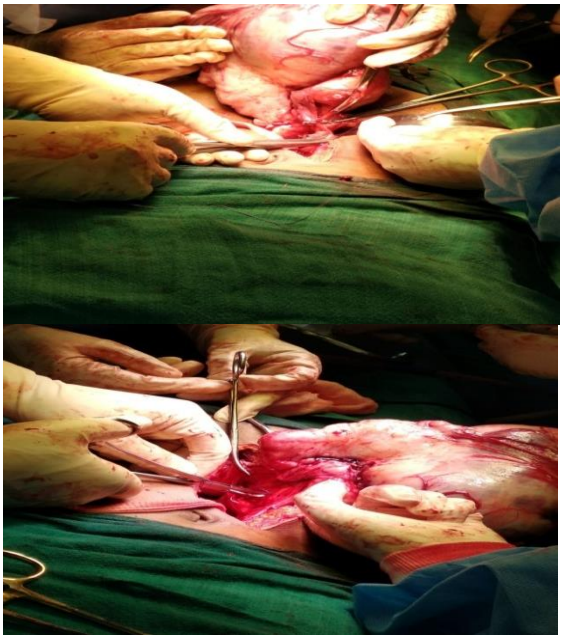

Figure 3: Infra-colic omentectomy done along with gonadectomy.

DISCUSSION

Patient suspected to suffer from Swyer syndrome are first subjected to laboratory testing for measurement of FSH, LH, TSH, free T4, estradiol, testosterone. In the described case, FSH and LH levels were elevated and estradiol levels were low; these findings were suggestive of hypergonadotropic hypogonadism.[5] Differential diagnosis of MRKH syndrome, complete androgen insensitivity syndrome (Morris syndrome) must also be considered.[6]

Karyotyping should be done in case of any pubertal delay and elevated gonadotropins.[7]

Once gonadal dysgenesis is confirmed, tumor markers should be done.[8] Transabdominal USG is the first choice diagnostic imaging. MRI is restricted to cases not diagnosed by USG. After surgical treatment, HRT is indicated.[9] Estrogen should be induced as soon as possible to ensure adequate bone mass formation and prevent reduction in bone mineral density that lead to osteopenia and osteoporosis.[10]

CONCLUSION

Patients with Swyer syndrome should be subjected to gonadectomy as soon as diagnosed because of their high risk for tumors such as gonadoblastoma. In cases of high risk of malignancy, bilateral gonadectomy with adjuvant chemotherapy should be considered. Cyclic estrogen and progesterone is indicated till the age of 50 years. The accurate and early diagnosis of these abnormalities would allow for conservative treatment, which can ensure the preservation of fertility, reduce emotional trauma, and improve patient survival.

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