A Rare Case of Ovotesticular Disorder of Sexual Differentiation

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INTRODUCTION

- Disorders of sexual differentiation (DSD) are congenital conditions characterized by atypical development of chromosomal, gonadal, or phenotypic sex.
- Among these DSD, only 5 % of cases have both mullerian and wolffian derived internal structures that depict corresponding adjacent gonad. This is known as True Hermaphrodite or Ovotesticular Disorder of sexual differentiation. Hence, the rarest among intersex disorders.
- Having both organs in one body yields to confusion of one's identity. Being born as well with ambiguous genitalia, assigned and reared as female however with male sex preference like the index case; is of great impact once left undiagnosed until age 23. The patient has regular menstrual period with associated left labial bulge. Genitalia is grossly female with a small phallus. There is normal breast and pubic hair development.
- Diverse phenotypes and karyotypes are evident to this special population. Thus, they are only distinguished by histologic diagnosis. This case is a mosaic karyotype of 47,XXY/46,XX hence suggestive of True hermaphrodite with Klinefelter variant. Biopsy likewise confirmed a diagnosis of Ovotestis.
- There were two cases of True hermaphrodite with Klinefelter variant published in the country but in contrast, both have 47,XXY/ 46, XY karyotype. No worldwide estimate was ever published.
- Approach may be limited to these conditions given a few number of cases. However, varieties of sexual orientation nowadays present a challenge to conquer these limits.

CASE

- This is a 23-year-old, born as the first of twin with ambiguous genitalia. The second of twin is male with no abnormalities. Index case has male sex preference when pubertal age was reached. No other symptoms noted such as excessive hair growth, excessive weight gain, abdominal pain or any discomfort.

Until 4 months prior to hospital admission
- There was lateralized lower quadrant pain but with concomitant bulging of left labia during her menstrual period.
- She self-medicated with analgesics

- Progression of pain occurred even during premenstrual periods 3 months prior hospitalization.
- There was associated nausea and vomiting.

Intractable pain for the succeeding couple of months prompted patient to seek consultation.

PHYSICAL EXAMINATION

- Ectomorph with normal vital signs. (Fig 1)
- Tanner stage 3 for both breast and pubic hair development but sparse axillary hair distribution. (Fig 2)
- Direct tenderness on the left lower quadrant area.
- Ambiguous female genitalia with small phallus (5 cm), swollen left labia and intact female urethra located inferior to the base of phallus. (Fig 3)
- No adnexal masses and no vaginal discharges. The rest of the examinations were normal.

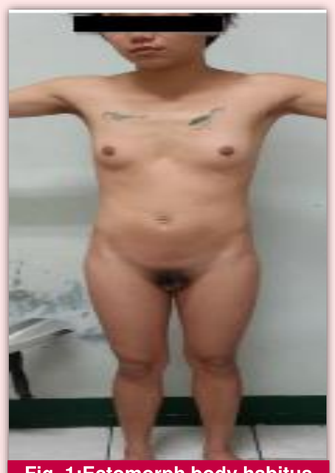

Fig. 1:Ectomorph body habitus with breast and pubic hair development

Fig. 2: Axillary hair distribution

Fig. 3: Ambiguous genitalia

DIAGNOSTICS

- Hematologic and blood chemistry examinations were all normal.
- Ultrasonographic findings revealed normal retroverted uterus, normal right ovary (Fig 4a) and a hypoechoic mass measuring 2.5 x 1.9 x 1.2 cm on the left inguinal area (Fig 4b)
- Karyotyping was requested with a result of 47, XXY/46, XX (Fig 5) indicative of mosaic karyotype of True hermaphrodite with Klinefelter variant.

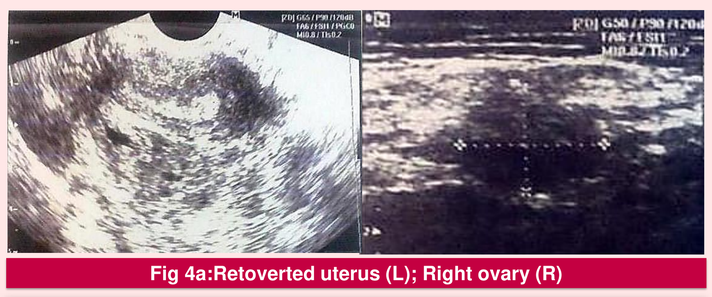

Fig 4a:Retoverted uterus (L); Right ovary (R)

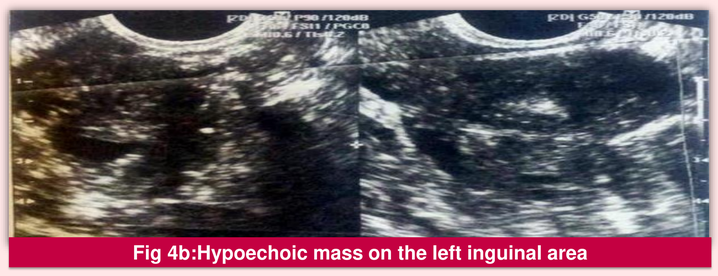

Fig 4b:Hypoechoic mass on the left inguinal area

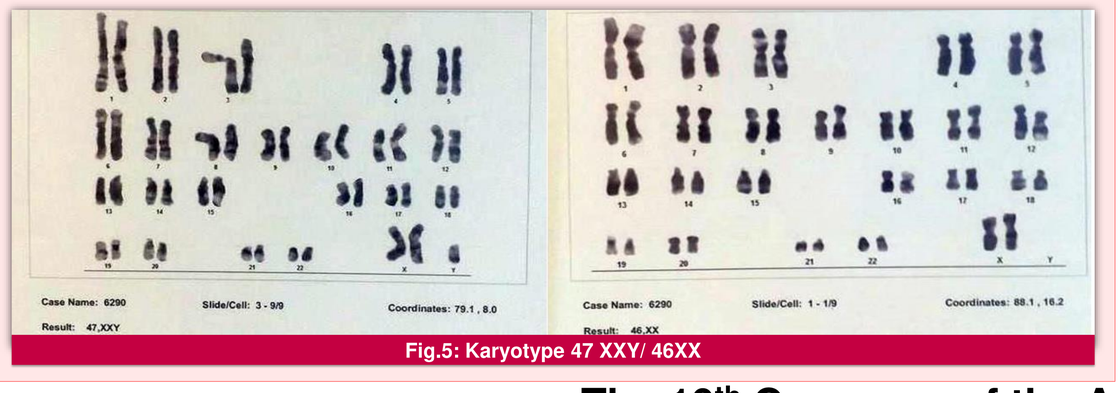

Fig.5: Karyotype 47 XXY/ 46XX

- Interdepartmental referral was made to Urosurgery, Internal Medicine-Endocrinology and Psychiatry and concurred with same diagnosis of G0, True hermaphroditism with Klinefelter variant.
- She underwent Exploratory Laparotomy, biopsy of the right ovary (Fig 6), left gonadectomy (Fig 8a& 8b) orthoplasty (Fig 9 &10) with left herniotomy (Fig10) under regional anesthesia.
- Intraopertive findings of unenlarged uterus, normal right ovary and fallopian tube but absent contralateral adnexa. Left gonad is located at the left inguinal area.

Fig 6: normal right ovary

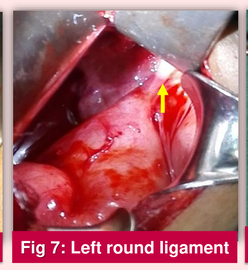

Fig 7: Left round ligament

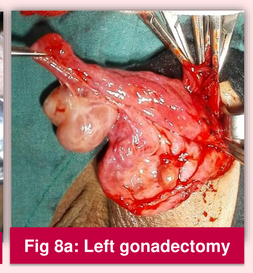

Fig 8a: Left gonadectomy

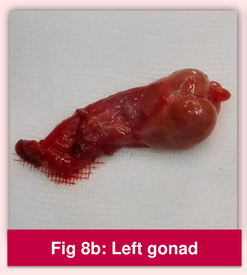

Fig 8b: Left gonad

Fig 9: Orthoplasty

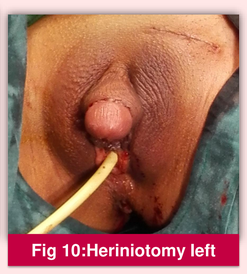

Fig 10:Heriniotomy left

DISCUSSION

- True hermaphrodite is a rare condition characterized by mixed ovarian and testicular tissues which may present as ambiguous genitalia. The index case is the first of twin while the second of twin is a male. Mechanism in this type of twinning is the occurrence of parthenogenetic or premature activation of an egg wherein diagnosis can be made from the time of delivery using DNA samples.
- Development of external genitalia reflects the androgen production level and exposure hence this can range from ambiguous genitalia to isolated hypospadias.
  - The index case had a small phallus measuring 5 cm in flaccid state. This is considered as phallus given the evident anatomical structure compared to mere hypertrophy of clitoris. Distinctive comparison between the two is accurately done in neonates wherein most standard deviation of measurements are studied and recorded based on these ages.
- Internal gonadal structures such as testicular development is explained by the mechanism of translocation of testis determining genes of Y to X chromosome or an autosome and autosomal dominant mutations that promote testis development in the absence of a Y chromosome. This may manifest as bilateral ovotestes or an ovotestis and contralateral ovary or testis.

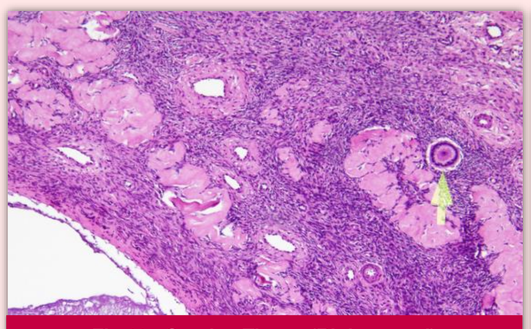

Fig 11: Ovarian Tissue (Right gonad)

Fig 12:Testicular (T) and Ovarian (O) tissues (Left gonad)

  - Documented in this case is a normal right ovary (Fig 11) and an ovotestis in the contralateral gonad (Fig 12).
- Descent and position of the gonad depend on the amount of testicular tissue present.
  - 50% of ovotestes are located abdominally.
  - 25 % are positioned in the inguinal region like the index case and the other 25% are labioscrotal position.
- The patient underwent Gonadectomy with Herniotomy considering a hypoechoic mass which was highly suspicious of gonad. This is abnormally located at the inguinal area which is the source of intractable pain and labial bulge. Moreover, this is also to prevent possible sequela of malignancy. It is recommended that gonadal biopsies for confirmation should be taken to differentiate from other intersex disorders.
- With male sex preference, the patient underwent Orthoplasty to completely mobilize and straighten phallus. It is claimed that it is being used as a normal male sexual organ with a female partner. Moreover, patient planned for preservation of female reproductive organs but no disclosure if for possible future gestation.

CONCLUSION

- Management of this condition is a Multidisciplinary approach.
- Early recognition and careful assessment upon delivery should be done to conduct proper diagnostics and appropriate interventions.
- However, sex preference of an individual should be taken into consideration prior to any procedure especially for patients who are diagnosed during pubertal or adult stage.
- This is the first case of True Hermaphrodite with Klinefelter variant in the country who has female reproductive organs. Regardless of their choice of sex preference or if there is preservation of both sexes; guidelines and advance interventions as to various age groups may be constructed through further studies to promote self-fulfilment and higher chances of fertility.

  In addition to the hormonal therapies that are usually administered, this case report may contribute to possible surgical approaches to these special population.

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