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Quick Response Code:

A rare case of 46XX ovotesticular disorder of sex development

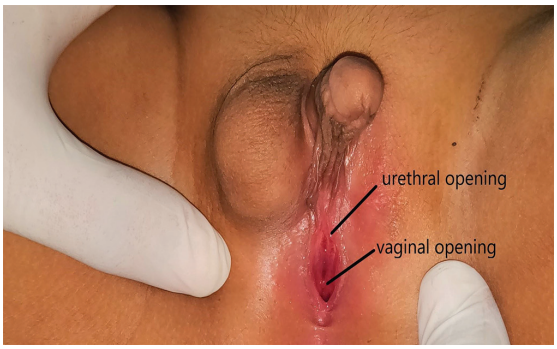

Fig. 1. Phallus with right hemiscrotum with two opening - urethral opening (open arrows) and vaginal opening (solid arrows).

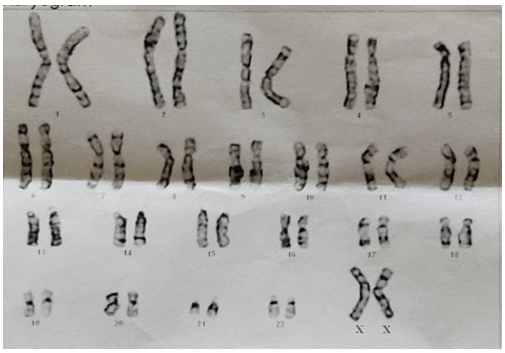

Fig. 2. Karyotyping of the child showing 46XX.

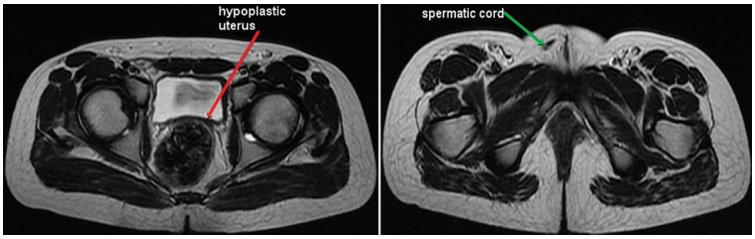

Fig. 3. Magnetic resonance imaging showing rudimentary uterus (red arrow) and right side spermatic cord (green arrow).

A 10 yr old male child[†] was brought to the department of Endocrinology, Andhra Medical College, Visakhapatnam, India, in October 2019, in view of ambiguous genitalia with non-palpable left gonad and passing urine beneath phallus since birth. Normal anthropometry was noted, and the child was brought up as male since birth. Phallic length was 3.5 cm and right scrotal gonad of 3 ml with no palpable gonad in the left labial fold was seen. Urethral and vaginal openings were noted (Fig. 1) and a karyotype of 46XX was observed (Fig. 2). Magnetic resonance imaging revealed severe hypoplastic uterus with the left fallopian tube and ovary of 35 × 10 mm (Fig. 3). Pre-pubertal hormonal profile was observed with greater than two times elevation of testosterone upon human chorionic gonadotrophin stimulation. As parents wanted to raise the child as a male, rudimentary uterus and intra-abdominal gonad removal and correction of perineal hypospadias was planned after re-evaluation at 14 yr of age.

***Conflicts of Interest*:** None.

Sagar S.L. Reddy[*] & K.A.V. Subrahmanyam
Department of Endocrinology, Andhra Medical College, Visakhapatnam 530 002, Andhra Pradesh, India
[*]*For correspondence*: kavsendo@yahoo.co.in

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[†]Consent to publish clinical information and images obtained from patients parents.