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An Interesting Case of DSD

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Case Presentation

We present a child with 46 XX SRY-negative maleness syndrome. phenotypically male, and genotypically female. The child was born at 33 weeks gestation, birth weight 1.5kg to consanguineous parents, and was noted to have ambiguous genitalia with hyponatremia and hyperkalemia (K 10mmol/L). Hydrocortisone and fludrocortisone was started with biochemical improvement; however as the hormone profile was atypical the medication was discontinued. The baby was assigned as female. At 2 months she was reviewed in national diabetes and endocrine centre where she was noted to have a phallic structure measuring 3.3cm, severe chordee with penoscrotal hypospadias and no vaginal opening. The scrotum was bifid and well developed with testicles palpable bilaterally in the inguinal area.

Investigations

Pelvic ultrasound confirmed bilateral inguinal testicles and suggested the presence of uterus and vagina. Laboratory investigations demonstrated elevated DHEAS and Testosterone with undetectable Estradiol and high AMH. Karyotype was 46XX, SRY gene negative. Mild hyponatremia and hyperkalemia were noted but an ACTH stimulation test was not suggestive of CAH. MRI at 4 months confirmed bilateral testicles in the inguinal region, with possible rudimentary vagina and uterus. The child was reassigned to male at 7 months. Diagnostic laparoscopy and bilateral gonadal biopsy confirmed testes, although the testicular appearance was unusual. The patient was doing well at last review and is now 2 years old.

Discussion

46 XX-SRY negative male is a rare disorder with only few cases being reported. Some of the reported cases virilise at puberty however all cases are infertile.