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CASE REPORT

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Pregnancy in a true hermaphrodite

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A unmarried apparently male patient, aged 17 years, attired in a shirt and waistcloth was seen on 13th September, 1997 with his hair clipped. He complained of menarche 1 year back, with 3 months amenorrhea and a painful suprapubic lump. He had a history of hernia operation done at the age of 8 years. A smooth testicle like structure was removed from the sac. He was having sex as a male and also as a female. On examination his skin was smooth, there was no moustache or beard, breasts were well developed and a suprapubic lump of about 12 weeks size was felt. On examining the genitals a well developed penis with well formed scrotum united in the midline was seen. The urethra was under the scrotal folds and a long narrow vagina admitting one finger was present. There was a linear scar in the left groin and a smooth swelling like a testicle in the right groin. On palpating it, the patient experienced testicular sensation.

Buccal smear showed a mosaic pattern with only 10% cells positive for sex chromatin. Luteinising hormone was 0.58 IU/L and follical stimulating hormone 0.75 IU/L (normal value in follicular phase 0.5 0- 181 IU/L)

Fine needle aspiration cytology of the groin mass showed spermatogonia and few spermatids; the impression being undescended testis with maturation arrest at spermatid level.

Sonography showed a 17 weeks macerated fetus in the uterus (Figure 1). The case was differentiated from hypospadias, testicular ferminization and congenital adrenal hyperplasia (CAH), and diagnosed as a true hermaphrodite (Figure 2).

D and C was done on 15th September, 1999. The patient refused corrective plastic surgery, karyotyping and even removal of the right testicle.

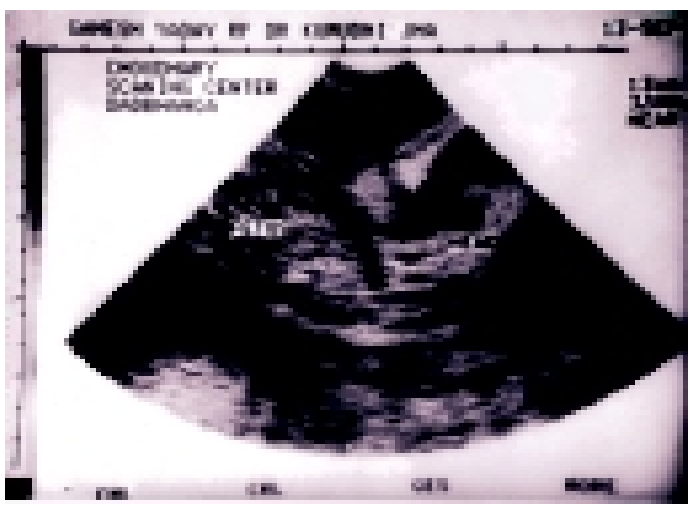

Figure 1. Ultrasonography showing macerated fetus

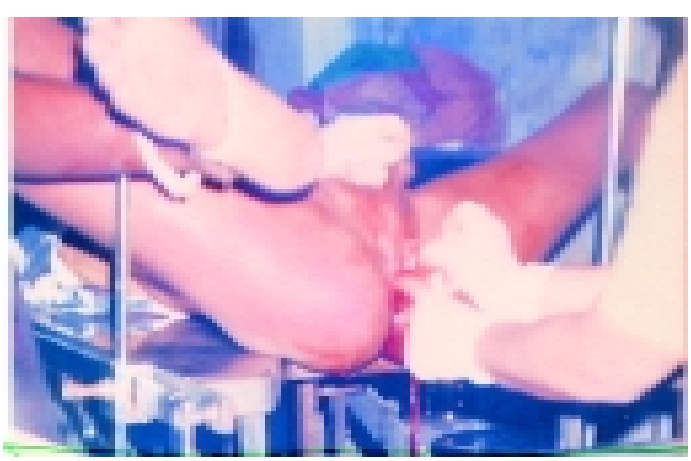

Figure 2. Testis in the right gooin, penis, cathter in the urethra, and finger in the vagina.

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Discussion

Abnormal sexual development may result from

1. Sex chromosomal abnormalities interfering with testicular differenciation – 45X/46XY mosaicism.
2. Biosynthethtic failure (enzymatic failure) –5-alpha reductase definiency leading to inadequate utilization of testosterone.
3. Mullerian inhibiting factor deficiency – mullerian defects.
4. Masculinisation of 46 XX patients in congenital adrenal hyperplasia (CAH).
5. Genes capable of producing H-Y antigen may be seen on an autosome, leading to 46 XX female.

True hermaphrodites are rare in Europe but commener in Africa and majority have a uterus and vagina, the karyotype mostly being 46 XX. This occurred in 58% of 172 cases reviewed by van Niekerk [1]. The next most common karyotype was 46 XX/XY seen in 13% , followed by 46 XY in 11%, 46 XY/47 XXY in 6%, and other mosaics in 10%. True hermaphodites are said to be 60% 46 XX, 20% 46 XY, and 20% mosaics and chimeras. The testicular cells must have a Y component. If both gonads have 46 XY cell lines, individuals with 46 XY/46 X karyotypes may possess normal testes and may be completely masculine.

H-Y antigen studies resulted in positive findings although Y chromosome specific DNA probes excluded a simple inheritance pattern and a nonenviornmental factor could be determined [2]. The most common distribution of gonads is an ovotestes on one side and an ovary on the other. Ovotestes may be bilateral or combined with a testes – such a case has to be differentiated from CAH (detected by urinary 17 ketosteroids) and from 46 XY androgen insensitivity cases. In these cases, despite normal breast development, the patient may come with primary amenorrhea, and scanty axillary and pubic hair, but a short blind vagina with no cervix. On laparoscopy the uterus is absent and the testes may be in the abdomen, inguinal canal or in the labia. Karyotype will show normal XY pattern. These patients lack androgen receptors and the gene located on X chromosome between xp 11 and xq 13 [3]. The child usually presents with a testes in a hernial sac, as in our case. Reductase deficiency is due to an autosomal recessive gene with familial presentation.

If CAH patients are treated with cortisol they may menstruate and therefore a hysterectomy with removal of the ovaries will be required. Changing of the sex entails plastic correction of the phallus, and may lead to severe psychological problems.

Kim et al [4] have reported a pregnancy in a true hemaphrodite. In phenotype female true hermaphrodites, 12 living offsprings have resulted from 14 individual pregnancies. Uterine maldevelopment and stenosis of the cervix or fallopian tubes however renders normal childbirth improbable. A true hermaphrodite may show good breast development and menstruate as in our case. In such cases adjustment to masculinity may entail hysterectomy and oophorectomy. Our patient had a functional uterus and ovary as he had conceived. He refused any adjustment of sex or a gonadectomy to prevent malignancy. Autofertilisation could not have been possible in this case as there was maturation arrest at the spermatid stage.

References

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