Testicular Feminization Syndrome

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Two sisters, aged 26 and 24 years, presented in the OPD on 8/2/1999 with history of primary amenorrhoea. They have three younger brothers and one elder sister married with two children.

Examination of both sisters revealed average uilt, absent axillary and pubic hair and Tanner Stage III breast development. Abdomen was soft with no organomegaly and normal hernial orifices. Both sisters had infantile external genitalia and in each case the vagina ended in a blind pouch of about 2-3 cms in length.

Investigations revealed normal blood urea and serum creatinine, negative buccal smear and 46XY Karyotype in both. Testosterone was 9.3 mg% in the elder and 5.3 mg% in the younger. Ultrasonography of both individuals revealed no uterus but both the gonads were seen and each were about 4 x 2 cms in size. Kidneys were normal in size, shape and echo texture.

Both of them underwent laparotomy and bilateral onadectomy on 16/2/99. Laparotomy revealed gonads were located in the ovarian fossae and were attached to ach other with pelvic peritoneal fold. The infundibulopelvic ligaments were found inferolaterally to the gonads.

Histopathologic examination of the gonads revealed immature testicular tissue and leydig cell hyperplasia.

Postoperatively T. Premarin 0.625 mg was administered once daily. The sisters were discharged on the 8th postoperative day with a final diagnosis of complete androgen insensitivity syndrome.

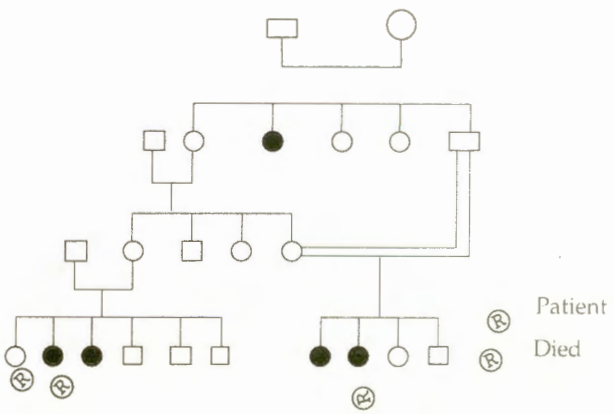

Fig. 1: Pedigree Analysis

Family history revealed that this condition had been observed in the family in two previous generation as shown in pedigree analysis (Fig. 1). The first was the maternal grandmother's sister. The second and third were daughters of their maternal aunt, who had married her maternal uncle.