Testicular Feminisation with Atypical Karyotyping

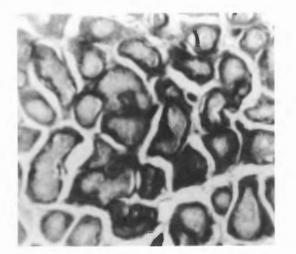
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Mrs. M. K., 20 yrs. H/F attended the GOPD of CNMC&H on 16.5.2000 with the C/O primary amenorrhoea. She was married for 1 ½ yr. And had no coital problems. O/E – tall and good looking, secondary sexual characters pubic hair – well developed; axillary hair – scanty; breasts – well developed, no galactorrhoea; P/A – NAD; External genitalia – normal. On coughing two oblong shaped mass each measuring 3 cm/2 cm were seen to be herniating through superficial inguinal ring into the upper part of labia majora. P/V and P/R – well developed blind vagina.

Routine investigations, carried out on 20-5-2000 were within normal limits, USG both from outside as well as from our institution showed the absence of uterus and poorly delineated ovaries. Karyotyping – numerical and structural abnormality of chromosome in the form of presence of double cell line with one 46xx in 40% cells and the other with 47 chromosomal counts. The extra chromosomal big sized chromosome appear to have been found by Robertsonian translocation of two chromosomes which appear to be x/x. GTG banding – a double cell line i.e. 50% cell line – 46xx and 50% of cell line is 46XY – Suggesting true hermaphroditism.

Diagnostic Laparoscopy which was done on 28-5-2000 showed absence of uterus and adnexa, Gonadectomy was done at the same sitting. Histological examination of removed gonads showed testicular structure only.



Microphotograph of the gonadal tissue HP shows structure of testis