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TESTICULAR FEMINIZATION

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CASE REPORT:

The testicular feminization syndrome is characterized by the presence of the tests in phenotypic females — good breast development and completly female external genitalia. In most patients, sexual hair, both pubis and axillary, is completely absent, but in some it is more or less sparse (2). Two hundred cases have been reported in the world until 1967. (1). This is a communication to report another case of this interesting syndrome.

Case report: A 17 year-old girl was referred to me in July, 1971, for primary amenorrhea. Physical examination revealed a height of 175cm. and a weight of 68kg. The blood pressure was 110/70mm Hg. She was a tall girl with a masculine figure who appeared rather depressed and mentally sub-normal. The breasts were well developed and pubis hair was scanty. The hair in the pubis area was in a triangular growth pattern. The external genitalia, labia majora, labia minora and clitoris were normal. There were bilateral inguinal hernia consisting of two solid mobile tumors palpated in inguinal canals near the labia majora. The bimanual examination revealed no palpable uterus, neither tubes nor ovaries. The remainders of the physical examination were unremarkable.

Laboratory findings, routine hemogram, urine analysis, fasting blood sugar and blood urea were normal. The protein bound Iodine was 4.8ucg. per 100ml. The twenty-four hour urinary excretion of total 17 Ketosteroids

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was 10.07mg. and that of urinary F.S.H. 52.6MU. The buccal smear revealed a negative sex chromatin pattern and Karyotype of leukocyte culture was consistant of XY constitution. The histological examination of biopsies obtained from gonads in the inguinal canals in August 1971 revealed testicular tissue with Sertoli and Leydig Cells. Seminiferous tubes were seen, but no Spermatogenesis was found. (Fig. 1,2)

In September 1971, a laparatomy was performed. The pelvis was masculine type. No uterus, tubes or ovaries were found. The result of the histo-pathological examination of totally removed gonads showed the same aspect as mentioned above. Following the surgery the patient was started on a long term estrogen replacement. (Fig. 3,4)

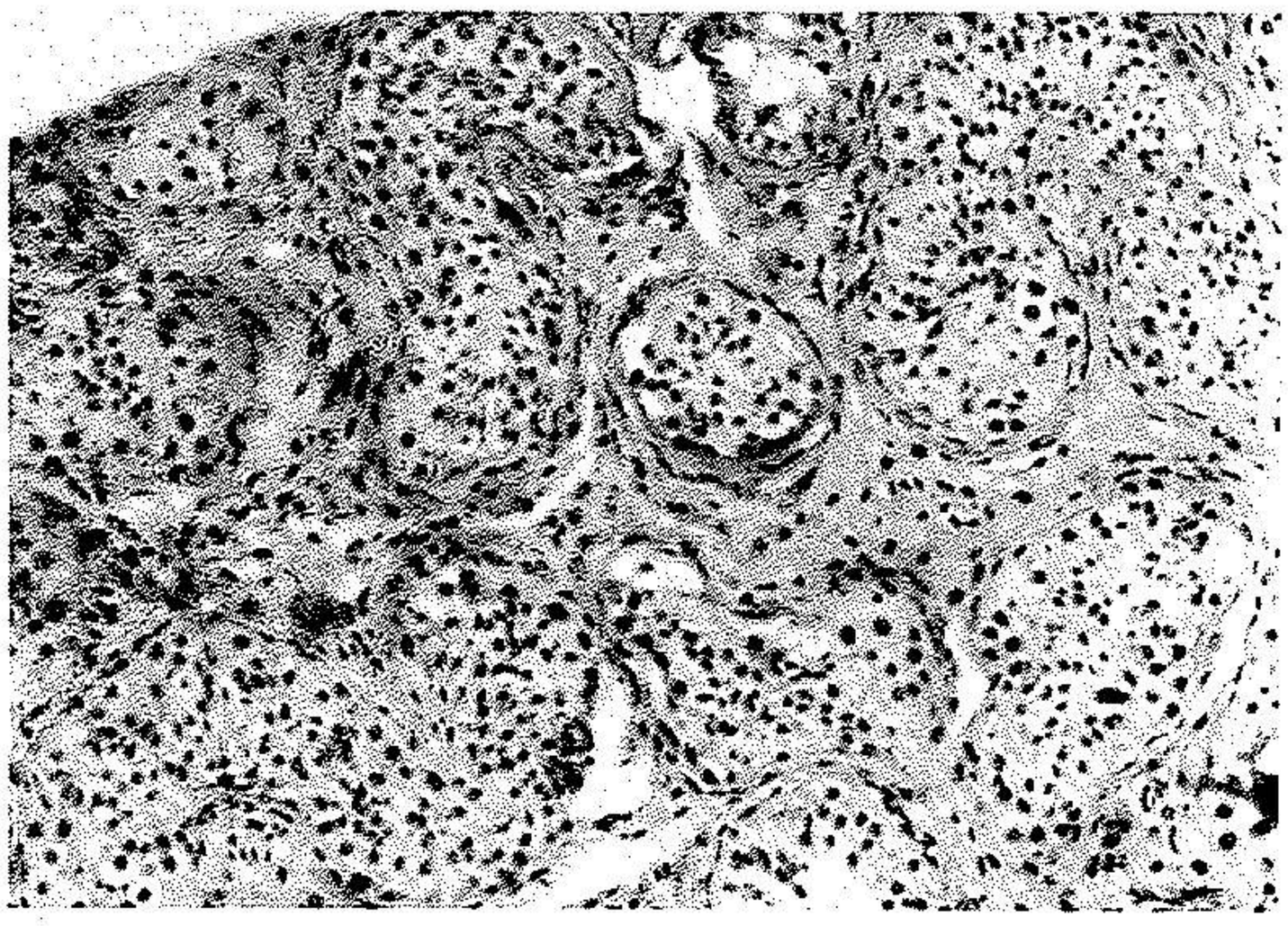

Fig. 1_2

The histological aspect of biopsies obtained from gonads
In the inguinal tissue.

Fig. 1

Seminiferous tubes, show sertoli and leydig cells, but No spermatogenesis is seem.

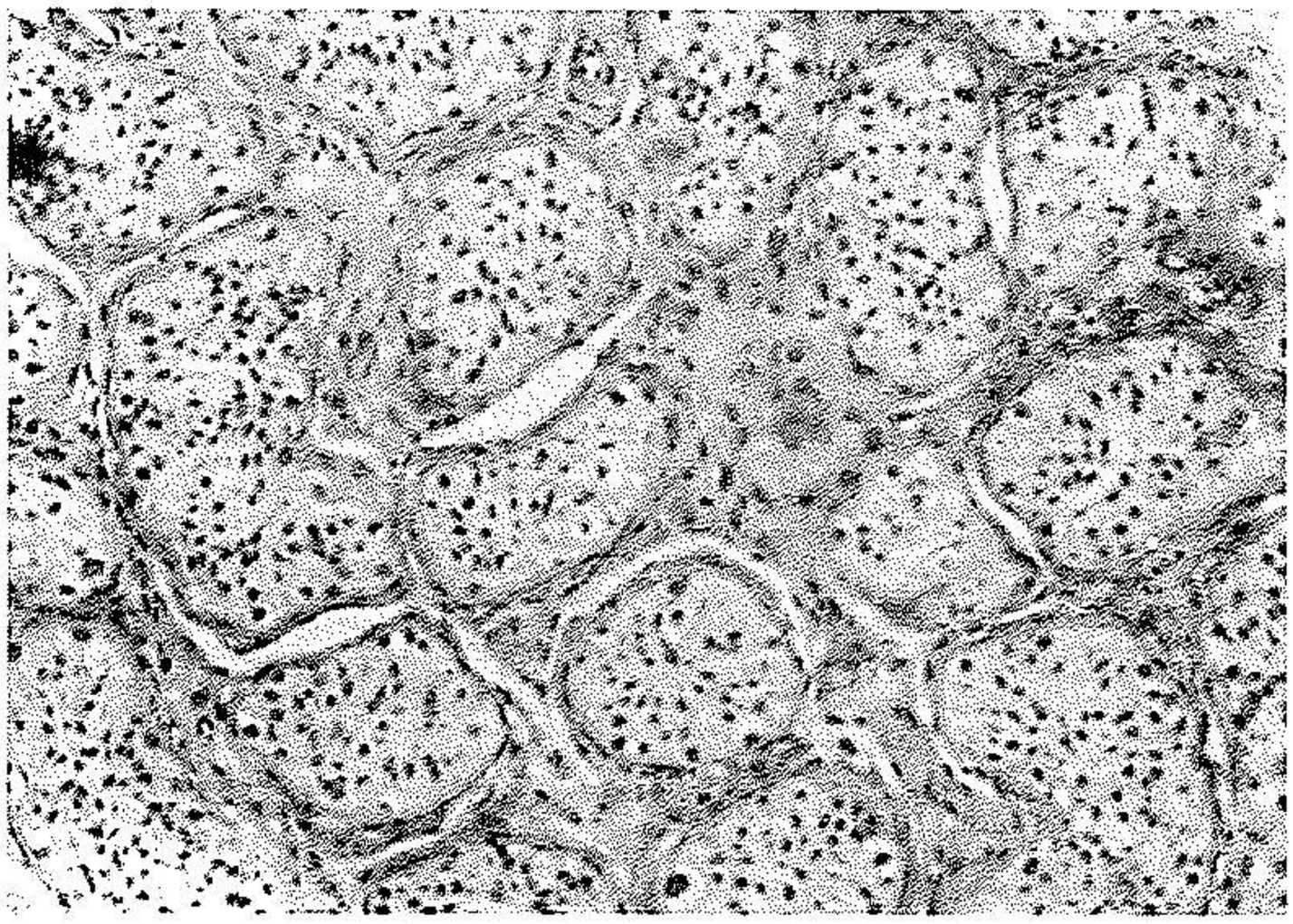

Fig. 2

Thickened tubular basement Membranes, lined with sertoli Cells.

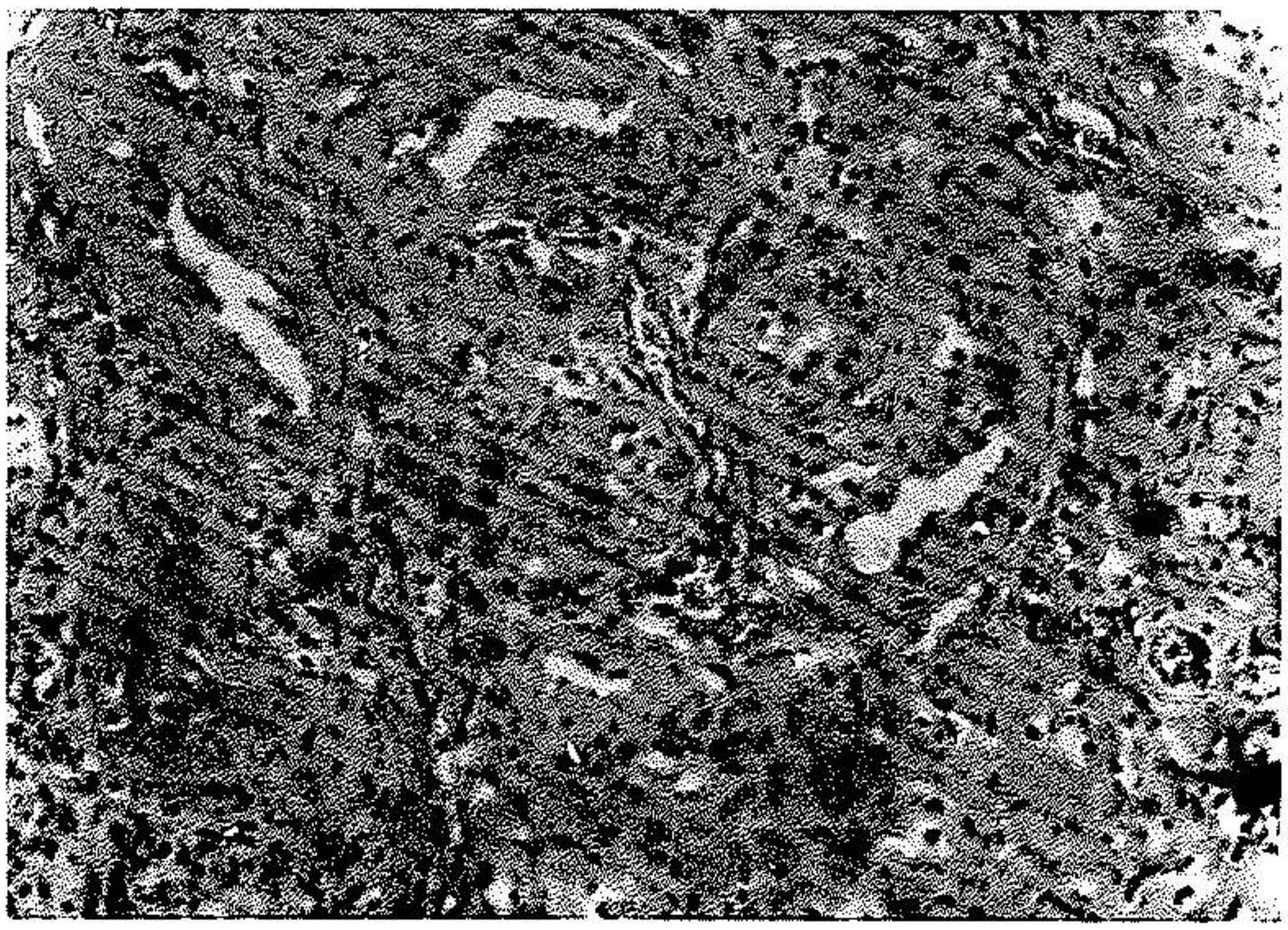

Fig. 3_4

The histological aspect of totally removed gonades.

Fig. 3

Tunica vaginalis, containing vessels, with thickened and Hyalinized wall, and infiltrated by inflammatory cells.

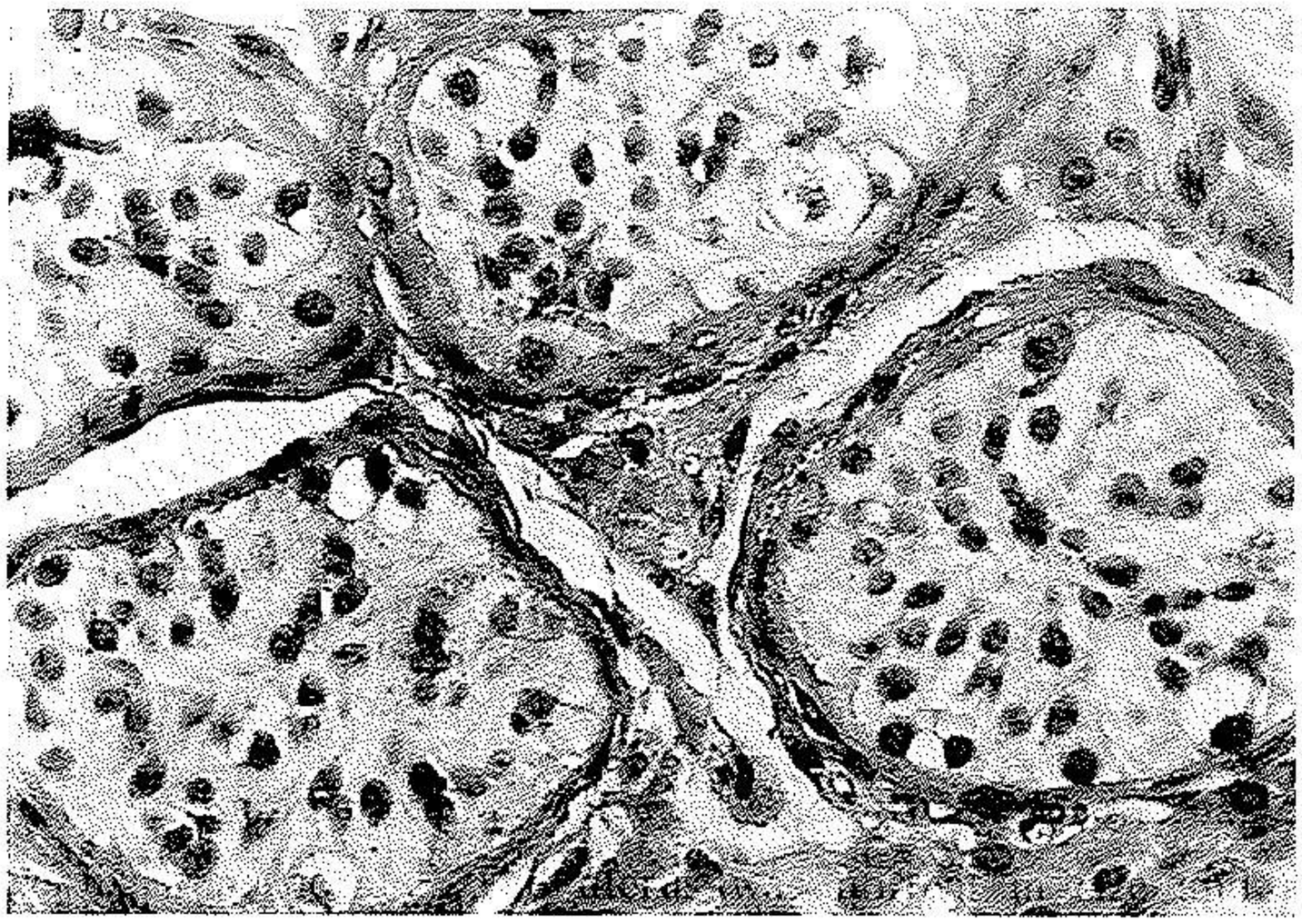

Fig. 4 Seminiferours tubes show a thickened and hyalinized basement Membrane, with sertoli and leydig gells.

Discussion:

Testicular feminization syndrome was first reported by Steglehner in 1817. It was then defined in 1948 by Goldberg and Maxwell (1) and was called Morris Syndrome (1). 200 cases have been reported in the world until 1967 (1).

The patient with primary amenorrhea caused by testicular feminization considers herself to be and appears as a normal female, having a feminine body contours, well developed breasts, proper external genitalia and a normal appearing introitus. The vagina may occasionally be absent or deformed, usually present, it ends in a blind pouch in as much as the uterus is absent. Upon closer inspection, both axillary and pubis hair are noted to be scanty.. The gonads morphologically tests, may or may not be palpable during bimanual examination. About half of these patients have their gonads in the inguinal canal. Most authorities prefer not to accept patients without spontaneous and usual development of the breasts during puberty as typical of testicular feminization although several have been reported.

The pre-operative diagnosis of testicular feminization rest upon the proof of Male-type chromosome pattern. The precise genetic cause of testicular feminization is not known.

Recognized as a female-carried hereditary disorder, its mode of transmission may be eventually prove to be that of either a sex linked recessive or a sex limited autosomal dominant, genetic entities now indistinguishable. The hypothestical pathogenesis of testicular feminization admitting it to be a wholly genetic aberration, embraces several possibilities; a variety of intra uterine testicular insufficiency. Some maternal interference with the fetal process of testicular induction and failure of genital end-organ responsiveness to fetal testosterone.

Each of these supposed mechanisms, representing deprivation of malegonad influence, would cause the genital organs to develop in female direction.

The most recent evidence from hormonal studies in patients with testicular feminization, demonstrating that their gonade produce both androgen and estrogen supports the thesis that there is a genetically produced failure of end-organ response to androgen.

If such nonresponsiveness of androgenic target organs is the basic defect in this condition, hairlessness and female type secondary sex characters become explicable.

It is then reasonable to compare testicular feminizarion to nephrogenic diabetes insipidus and pseudohypoparathyroidism, disorders in with hormonal insufficiency plays no role.

Résumé

Une observation de testicule felminisant est rapportie. Il s'agit d'une jeune fille de 17 ans qui nous consulte pour une amenorrhee primaire.

L'examen physique montre une grande jeune fille de 1,75m. pesant 68kg. avec une tension arterielle de 110/70. Le phenotype est feminin avec des seins bien developpes, la pilosite pubienne triangulaire est peu fournie, l'appareil genital externe est normal, le vagin bien developpe est largue. Il n'existe pas de trace d'element derivant des canaux de Muller.

-¸La palpation des regions inguinales montre une double hernie avec presence dans le sac de 2 formations dont la biopsie se revile etre du tissu testiculaire.

La sex chromatinien est negatif, le caryotype est XY.

La malade a ete castre est une hormonotherapie subsitiurive instauree.

References

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