ACTA MEDICA IRANICA
BVol. XIII; 1970, P. 41-45

CHROMOSOMAL & CYTOGENETIC

Studies in a case of Klinefelter's Syndrome

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Case Report:

Sh., A., a twenty one year old man was first seen in June 1967, and admitted to the Razi Hospital for the first time because of gynecomastia, hypogonadisme and underdeveloped penis and testes.

There was no family history of consanguinity, congenital abnormalities and hypogonadisme.

Physical Examination:

His height was 186 Cm., weight 96 kg., blood pressure 138/80 mm. Hg., and pulse rate was 70 per minute.

The head circumference was 55 cm., and extremities were thin and delicate, and the subcutaneous fat tissue was increased in chest and abdomen (Fig. I.). There was no beard and axillary hair. Pubic hair, present only at the penis, was black and straight. Gynecomastia was present.

The penis was small and the testis measured less than 2 Cm., the prostate gland was small and soft.

Special Labratory Findings :

Routine blood counts, urinanalysis and blood chemistry determinations were with in normal limits. The radio-active iodide uptake was 20% in 24 hours.

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Fig. 1

Urinary 17_Ketostroides were 12 mgs., per day. Urinary gonadotr_opin excretion was 150 mouse units per day on two occasions.

Tissue Studies :

Buccal smears showed positive sex chromatin similar to those seen in females. The karyotype determined from culture of leucocyts from peripheral blood was 47 (XXY), (fig. 2) .

A biopsy specimen from the testes revealed atrophy and hyalinization of the seminiferous tubules in which Leydig Cells were preservd. Spermatogenic activity was absent.

Comments :

The association of small testes, hyalinization of the seminiferous tubules with spermatogenic arrest, gynecomastia, increased excretion of gonadotropin in the urine and XXY karyotype clearly established klinefelter's Syndrome.

In 1942 Klinefelter, Reifenstein and Albright described a clinical syndrome of hypogonadism and gynecomastia. Barr demonstrated that many of these patients were chromatin positive.

They have 47 chromosomes instead of 46, 2X chromosomes and 1Y.

Some patients with Klinefelter's syndrome, however, lack of the Clinical fiindings of gynecomastia, hypoplastic testes, and eunuchoidisme and may be diseovered only when they appear in a fertility clinic with a finding of aspermia; thu spectrum of Klinefelter's syndrome is quite broad.

For the first time in 1966 Grand and Rossen suggest an unique combination of hypogonadism, a leukocyte karyocyte of 47 XXY chronic lung disease, dwarfisme, microcephaly, hypothyroidisme, deafness and pancreatic insufficiency.

Treatment :

The testicular lesions is irreversible. The use of Gonadotropin therapy, unfortunately is not consistently effective.

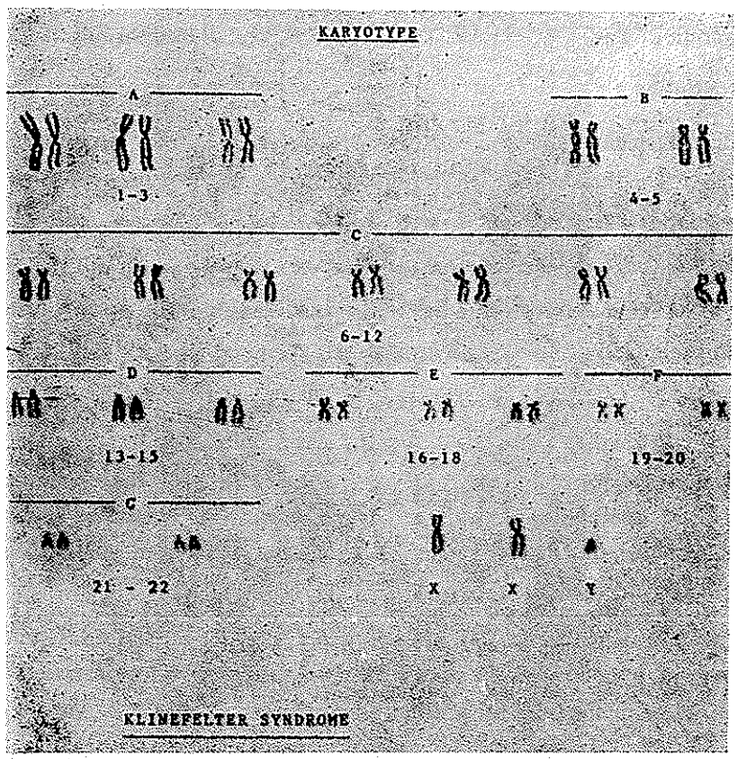

Fig. 2.

If androgen deficiency is present, treatment with malesex hormone is effective. The gynectomastia is not affected by hormonal treatment and mastectomy may be necessary in some patients for cosmetic reasons.

Summary

A 21 year old boy was first seen because of Gynecomastia and primary hypogonadisme.

A biopsy specimen from the testes revealed hyalinization of the seminiferous tubules with spermatogenic arrest, and tissue studies showed positive sex chromatin and the Karyotype was 47 (XYY).

Resume

Le Biopsie testiculaire et l étude de "Sex chromatin" chez un jeun homme agé de 2I ans avec gynécomastie et hypogonadisme nous á montré une hyalinisation des tubes seminiferes et un sex chromatin positive à type (XXY)

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