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Turner's and Stein-Leventhal's Syndrome in Two Sisters*

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The Turner's syndrome is a well established entity. There is however little knowledge on the exact pathogenesis of the Stein-Leventhal's syndrome (S. L. S.) despite the efforts of many investigators.

In this communication we present two sisters, one of whom had cardinal manifestations of S. L. S. while the other showed many of the clinical and laboratory findings of the Turner's Syndrome. We hope that our report will prompt others to record their similar experiences and hence will result in a better understanding of the possible interrelations between these and other sexual developmental disorders.

Case I : A 20-year-old female was referred to us with the chief complaint of amenorrhea (Figure 1). Physical examination on admission revealed her height to be 144 cm (< 3 percentile) and a weight of 53 kg. Other pertinent findings included short neck, low hair line, a shield-like chest with very little breast development, multiple pigmented nevi, cubitus valgus and short 4th metacarpal bones bilaterally. The examination of the external genitalia revealed no abnormalities. Likewise pubic and axillary hair were normal in appearance. Intravenous pyelogram showed bilateral double collecting systems draining into separate pelves and ureters. Sex chromatine was 1% (control 16%). Chromosome analysis using peripheral leucocytes revealed two kinds of cell populations. One cell line showed 45 chromosome in which the missing chromosome was from the C group. In the other the model chromosome

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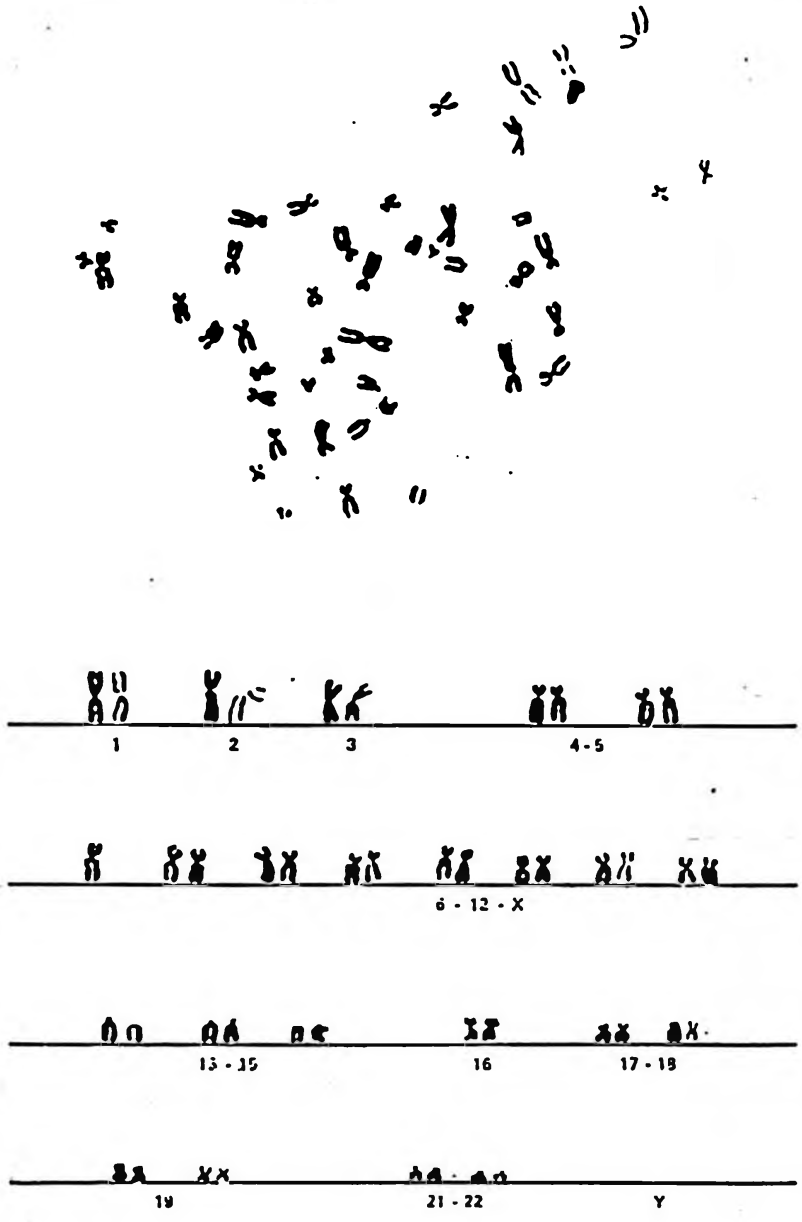

Figure 1

number was 46. However, karyotyping from this cell line showed that one of the chromosomes (45,X0/46,XXr), which was missing from the C group was replaced with a ring chromosome (Figures 1 and 2).

Dermatoglyphic findings were not unusual except that she had ulnar loops on all fingers.

Case II : The 23-year-old sister of the previous case was also seen with similar complaints. She stated that she had had her first menstruation at the age of 13 which had stopped after a few months. During the past few years, she had occasional menstruation following hormonal therapy. Two years ago, hirsutism, mostly over the face and legs, and obesity were noticed.

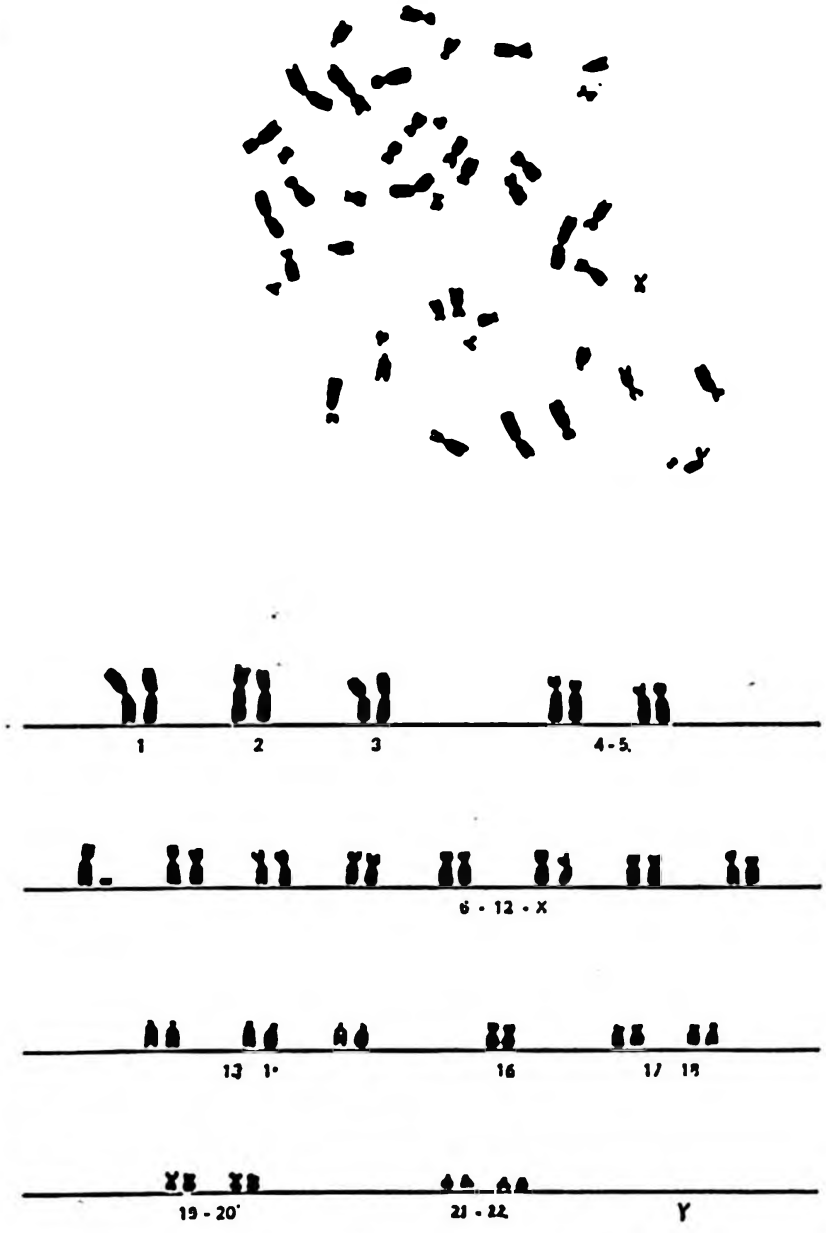

Figure 2

On admission, her height was 150 cm. (< 3 percentile) and weight 66.5 kg. No other abnormal findings could be found with the exception of marked hirsutism. Sex chromatine was 12% (control 16%) and chromosome studies using peripheral leucocytes revealed normal female pattern (46, XX). A laparatomy was performed which revealed a thickened tunica albuginea, grayish white in color, and multiple small cysts in both ovaries. A bilateral wedge resection was done. Biopsy material studied disclosed multiple follicule cysts (0.2 - 0.3 cm. in diameter) in addition to slight fibrotic thickening of the cortex (Figure 3). Her post-operative period was satisfactory and she began to have menstruation shortly after operation.

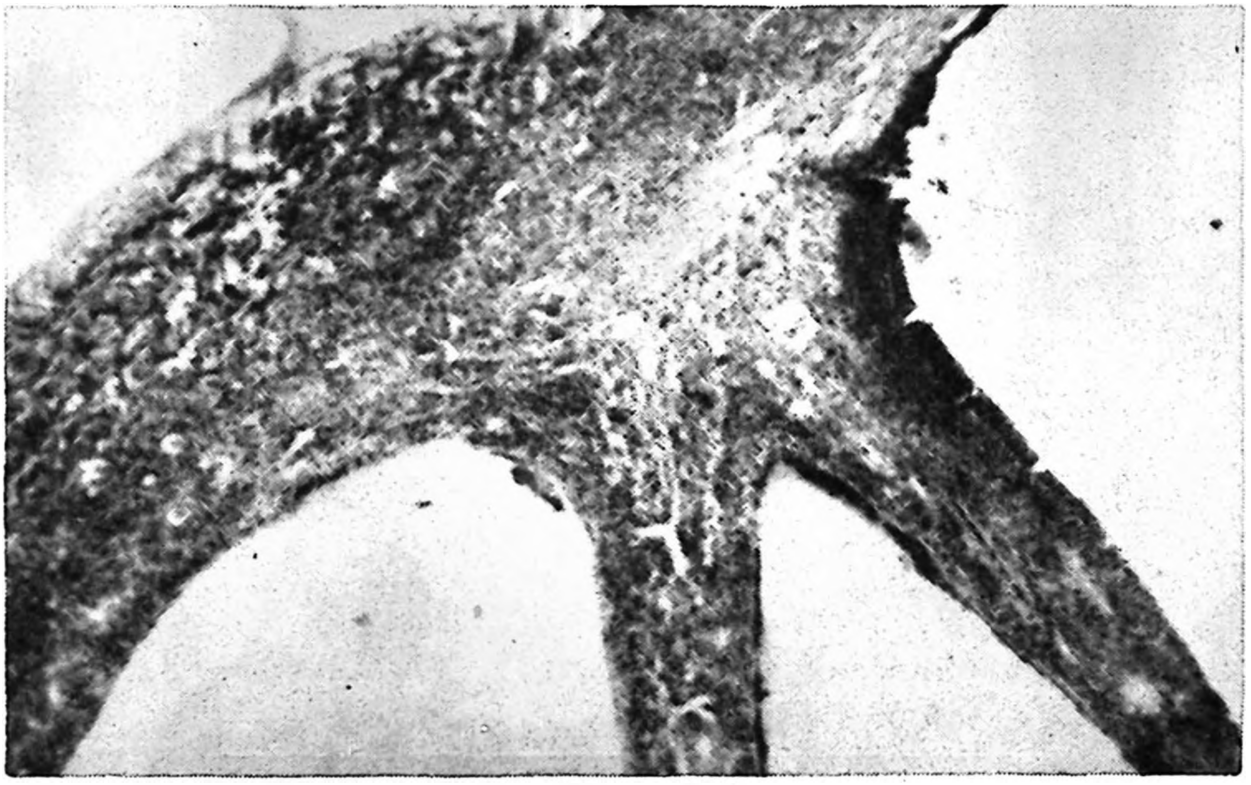

Figure 3

Discussion

As the clinical findings and karyotype indicate, the first case is a good example of the Turner's syndrome. The manifestations of her sister, however, are more typical of Stein-Leventhal's Syndrome. Perhaps the occurrence of two cases with sexual developmental disorders of different type in the same family is no more than coincidental. On the other hand, it should be noted that one case has been reported by Grouchy et al in which the authors could not definitely identify one syndrome to the exclusion of the other. This reported case, with a partial deletion of one of the X chromosomes also disclosed small spindle-shaped, nacreous ovaries.[1]

In view of the findings in this particular Case I, it may be speculated that our second patient also has partial chromosomal mosaicism (e. g. XX/XO) involving other tissues such as the ovaries. It is of interest, in this respect, to note that, there are other reports in the literature indicating chromosomal mosaicism in some, but admittedly few cases of Stein Leventhal's Syndrome. We found two cases in the literature with this syndrome who also had 46, XX/45, XO mosaicism.[2 3] In addition to a third case with 46, XX/46, XXq.[4] Although during the same survey we found 23 cases of S. L. S. who had normal chromosomes,[3 5-7] one should take note that in all these patients only peripheral leucocytes were used for chromosome analysis. It may be of further interest with

regard to our patients to mention the report by Williams et al in which a patient with normal female karyotype and usual stigmata of the Turner's syndrome, disclosed polycystic ovaries at the exploratory laparatomy.[8]

There are other reports in the literature about the families, various members of which had different sexual developmental abnormalities. In this respect it may be of interest to mention the family in which one sib had pure gonadal dysgenesis while her sister had ovarian dysplasia.[9] In both of these patients the chromosome studies revealed a female karyotype. Likewise we are aware of other reports of familial male pseudohermaphroditism in which the affected members had different sexual developmental abnormalities.[10] We believe all these findings point to the possibility that there may be certain relations between the various entities of sexual developmental abnormalities which have yet to be clarified.

Summary

Although the Turner's syndrome is a well established entity, there is little knowledge of the pathogenesis of the Stein-Leventhal's syndrome. In this communication two sisters, one of them having Stein-Leventhal's syndrome and the other Turner's syndrome, are presented. In view of the finding of families with members with various sexual developmental disorders, it is suggested that there may be some relation between such entities.

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