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FAMILIAL CONGENITAL MUSCULAR DYSTROPHY
WITH GONADAL DYSGENESIS

(Abstract)*

In the medical department, Hammerfest Hospital, seven individuals in one family were observed, suffering from cataract and generalized weakness in the striated musculature. Five of the patients were under ten years of age. The two eldest ones were sister and brother, 22 and 26 years of age, respectively. The woman could hardly walk even with support. She had clinical signs of ovarian agenesis, increased urinary excretion of gonadotrophic hormones, and high tolerance to insulin. The diagnosis was verified by laparotomy demonstrating thin, white, fibrous ovaries and infantile internal genitalia.

The brother could hardly walk, had fairly long extremities, cubitus valgus, and widely separated nipples. The pubic hair presented a feminine appearance and the testicles were infantile and soft. No increase in urinary gonadotrophic hormones were found. The amount of neutral 17-ketosteroids was low and the output of oestrogens increased in urine. The histological examination of testicular tissue showed a thick, fibrous capsule and scanty testicular canals with a few Sertolian cells. Most of the preparation, however, merely consisted of a hyaline substance. In the vascular stroma were seen large heaps of Leydig cells, similar to what is seen in Klinefelter’s syndrome.

The ovarian agenesis may either have been caused by a virus infection during early embryonic life or be due to heredity. The latter suggestion is supported by the presentation of two sibs with ovarian agenesis and Klinefelter’s syndrome.

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