TURNER'S SYNDROME IN THE NEWBORN

(Report of a Case)

by

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and

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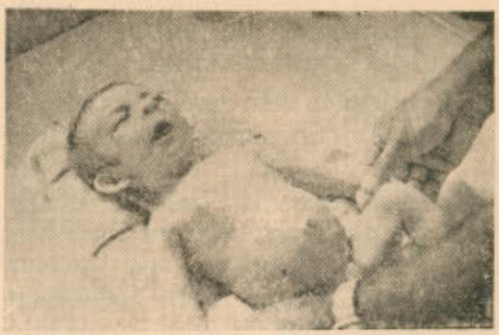
In 1938, Turner described the syndrome which bears his name. The designation 'Ovarian Agenesis' was subsequently suggested with reference to the state of the ovaries. Sex chromatin and cytogenetic studies have thrown light on the basic chromosomal aberrations in these cases, and the term 'XO chromosomal anomaly' would appear to be the most logical. In view of its distinctive clinical features, the condition lends itself to recognition at all ages. The present communication relates to a neonate with Turner's Syndrome met with at the Medical College Hospital, Calicut. The entity in the neonatal period has not been reported in India.

Case Report

A female infant was born normally at term of a 25 year old primigravid woman. The pregnancy had been uneventful except for the presence of maternal hydramnios towards term. The birth weight was 5 lbs. 10 oz., the length being 17.3". An interesting combination of congenital anomalies was present. These included bilateral pitting oedema involving the feet, legs and hands, marked webbing of the neck, broad chest, a low hair line at the nape of the neck, and prominent low set ears. The infant was hypognathic, the palatal arch being narrow. Simian creases and distally placed triaxial radii were observed in both palms, and the finger nails were small. Cubitus valgus was not present. There was a short Grade III systolic murmur along the left sternal border. The femoral pulses were markedly diminished. The external genitalia were normal, although the clitoris was slightly enlarged. Buccal smear examination revealed a negative chromatin pattern. Facilities for karyotype analysis were not available.



Discussion

It is evident that the infant had the cardinal features of Turner's Syndrome, like congenital lymphoedema of the extremities, webbing of the

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neck, coarctation of the aorta etc. Franklin (1963) has drawn attention to the low birth weight at term. The average birth weight locally has been observed to be 6 lbs. 5.8 oz. (Nair et el., 1963). Lemli and Smith (1963) have stressed that the abnormality common to all their patients was shortening of stature present from birth. Dermal configurations described by Uchida and Soltan (1963) in Turner's Syndrome include distal displacement of axial triradii, simian creases, single crease on the fifth finger, and bridged upper palmar creases. Male sex chromatin pattern can be detected in the large majority of cases, the female pattern being present in as few as 20 per cent. XX/XO mosaicism has been reported in these cases. Sohval (1963) has suggested that the extragonadal features may be due to one or more genes having pleomorphic manifestations. Since laparotomy was not done, no opinion can be expressed as regards the state of the gonads.

Kaul et al. (1964) reported a case where congenital somatic abnormalities characteristic of Turner's Syndrome were associated with the presence of ovotestes rather than ovarian agenesis. It is interesting to recall that Turner (1944) made no mention of the condition of the gonads.

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