TRISOMY SYNDROME

Report of a Case*

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The syndrome of trisomy 13-15 was originally described by Patau et al in April 1960, although it was Therman et al (1961) who designated the entity 'D: trisomy'. Marden et al (1964) came across 2 cases in their survey of 4412 neonates for congenital anomalies. It would appear that a similar frequency pattern obtains in this part of the country as well. Including the case under discussion, two infants with the condition were encountered among the 5234 infants born during a three-year-period at the Medical College Hospital, Calicut. The first of these forms the basis of a previous report (Nair 1965). To our knowledge, no other cases have been observed as yet outside Europe and America.

A wide spectrum of anomalies has been described in the cases reported in the literature to date, although no two cases have been identical. The major abnormalities associated with the syndrome have been sufficiently similar to make possible reliable clinical diagnosis in most cases. Despite individual patient variability, the most common abnormalities have been cleft lip, cleft palate, shallow supraorbital ridges with sloping forehead, low set ears, eye defects, micrognathia, flabbiness of the skin of the nape of the neck, polydactyly, rocker bottom feet, haemangiomas, heart defects, horizontal palmar creases, and renal anomalies. Smith (1963) emphasizes that the prognosis for survival is grim. He considers the variability in phenotypic expression to be due to differences in the genetic background plus maternal host environment. Townes et al (1964) attribute the variation in number, type and degree of anomalies in trisomy syndromes to the innumerable different combinations of genes that are possible for a given chromosome. Rhode and Berman (1963) have proposed some postulates to account for phenotypic variability as a function of environmentally governed factors. While the precise nature of the aetiology is obscure, the parents need to be told that the entity is sporadic in origin with no likelihood of succeeding progeny being affected.

Report of a Case

A 37-year-old pregnant woman was brought to the hospital on 11.12.64, eight hours after rupture
Fig. 1.—Front view of patient with D1 trisomy.

Fig. 2.—Side view of the same patient.