3rd centile. In our case height lay between the 3rd and 10th centiles and weight was below the 3rd centile. In previous cases height has usually been between the 25th and 50th centiles with 3 exceptions: the cases of Ricci *et al* (1968) and Kesaree and Woolley (1963) being less than the 3rd centile for height while the patient of de Grouchy *et al* (1968) was above average height. The weights showed no consistent features. In every case (where given) except our own (in which the ratio was of a child aged 3½ years) and that of Ricci *et al* (1968), the ratios of upper to lower segment were less than 1·00 showing that the legs were abnormally long.

The skin creases on the palms and soles were not markedly abnormal in our case, but in Kesaree and Woolley's case (1963) there was a simian crease, which is not necessarily of much significance. In view of the findings of a murmur in the present case it is of interest that a patent ductus arteriosus was found by Brody *et al* (1967) and Kesaree and Woolley (1963).

In conclusion, it would appear that no characteristic clinical picture can be defined, but mental subnormality and dwarfism are to be expected, together with a few abnormalities of the feet and hands; a congenital cardiac abnormality may be present in about a third of the cases.

Summary

A child is described with approximately 50:50 mosaicism 48,XXXX/49,XXXXX. The clinical features are compared with those of previously published cases with multiple X complements.

Examination of the frequency distribution of sex chromatin bodies and 'hot' Xs shows that each supernumerary X appears to be acting independently in respect of interphase condensation and DNA synthesis.

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Triple X Female and a Down's Syndrome Offspring

The triple X female has been characterized by the variability of her physical findings (Harnden and Jacobs, 1961; Telfer *et al*, 1970). Less attention has been given to the variability of her mental abilities (Day, Larson, and Wright, 1964). Most of these females have been purported to have reduced mental abilities. The following report deals with a triple X female who has normal intelligence and who is functioning as a quite adequate housewife and mother. She came under our observation after she produced a child with Down's syndrome (mongolism).

Case Report

Physical Findings. The patient is 30 years of age. She is a housewife who completed 2 years of college. Her height is 157·6 cm and weight is 69 kg. There was nothing unusual about her physical features that would have called attention to her abnormal chromosomal pattern. She has had 4 pregnancies. The first 2 re-

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sulted in 2 normal living children. The 3rd pregnancy resulted in a spontaneous abortion. Her last pregnancy at the age of 29 years resulted in a child with Down's syndrome. The child had 47 chromosomes with trisomy 21. The patient also has 47 chromosomes with a triple X karyotype. About 20% of her buccal cells contain 2 chromatin bodies. The patient's husband is physically and mentally normal. He has 46 chromosomes with a normal XY karyotype.

Discussion

Triple X females have shown a marked phenotypic variability. It is this variability of the physical findings that has made screening for these females difficult without examining their buccal cells for 2 chromatin bodies or doing a chromosome evaluation. The present triple X female was drawn to our attention after she gave birth to a child with Down's syndrome. There was nothing about this woman's physical appearance that would have indicated that she possessed an extra X chromosome.

While many of the reported patients with the triple X syndrome have had reduced intelligence, this finding is probably due to a biased selection of patients (Day *et al*, 1964). Many of the initial reports about these females came from institutions for the mentally retarded where mass screening was performed. Our patient is one of the few observed with normal intelligence; however, it is suspected that there are many more like her in the general population. Incidence at birth of this trisomic state is between 1.4 and 1.8 per 1000 live births (Maclean, Harnden, and Court Brown, 1961). Such a high frequency of this trisomic condition would indicate that there are many more triple X females in the general population than can be accounted for in institutions for the mentally retarded.

To our knowledge this is the first example of a triple X female giving birth to a child with a chromosomal abnormality. While a number of triple X females have given birth, the offspring have been chromosomally normal. It is not possible to determine if this triple X mother was at an increased risk of giving birth to a child with Down's syndrome. Examples of 2 chromosomal abnormalities in the same individual are well documented (Smith, 1970). There is, however, a lack of information about females with chromosomal abnormalities being at risk to produce an entirely different chromosome abnormality since most of these individuals are physically or mentally handicapped or fail to procreate.

It would be of interest to speculate that there is a causal relationship in the fact that this triple X

mother produced a trisomy 21 child. By now many mothers of patients with Down's syndrome have had chromosomal studies done and this is the first instance where a triple X mother has been identified. If there is a cause and effect relationship between this triple X mother and the trisomic 21 offspring, it is difficult to propose a mechanism at this time. Whether or not this was a chance occurrence will be determined as more children of triple X females are examined.

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It is with great regret that I have to report the death of Professor Hsia on 27 January 1972—Ed.

A Human Ring C Chromosome Associated with Multiple Congenital Abnormalities

Studies over the past several years have demonstrated a number of different chromosome abnormalities in patients with retardation and congenital defects. One of the less common structural abnormalities is a ring chromosome, and we have found only 5 instances of a ring C chromosome recorded in the literature. We have recently evaluated another patient with this chromosome abnormality and report the cytogenetic and phenotypic changes in this patient.

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