MATERIALS
MATERIALS

Clinical, cytogenetical and biochemical studies were carried out on 346 individuals examined in the Pathology Department of the Institute of Postgraduate Medical Education and Research, Calcutta. All the individuals were screened for sex chromatin and cases that were studied karyotypically were divided into patients and the controls.

Control.

Control was maintained from the study of ten individuals, seven females and three males referred for various reasons not involving sex chromosome anomalies. Some cases showed symptoms of mental deficiency, growth retardation and skeletal abnormalities but the rest were clinically normal. Screening of the sex chromatin and karyotypic analyses served as the cytogenetic control in this study.

Patients.

Majority of the patients had decided upon a hospital investigation mainly because of primary amenorrhoea or infertility. They were referred from the various Out Patients Departments, especially the Departments of Gynaecology and Urology, of the Institute of Postgraduate Medical Education and Research, Calcutta and even from other hospitals in the city and state. Complaints of the patients ranged from short stature, primary or secondary amenorrhoea in females to cases of ambiguous external genitalia, hypogonadism and cryptorchidism in males.

Data initially collected from the patients included details of the complaint and its duration, the nature of labour, medication or diseases, if any of the mother during pregnancy, general appearance of the proband, type of facies,
disposition of the limbs, presence or absence of secondary sex characters, measurements from the crown to the symphysis, arm span and total height. Family data collected included the number of siblings arranged according to birth sequence, position of the proband among the sibs and a history of diseases, if any, among members of the family or near relatives. In cases of primary amenorrhoea care was taken to eliminate cases of heredo familial delayed menarche and those with certain diseases (e.g., tuberculosis) which could account for the failure to menstruate.

Diagnosis of the cases was made on the basis of clinical features before any cytogenetic investigations were carried out. Buccal smears were taken for sex chromatin analysis from a total of 346 individuals, 276 females and 70 males. Detailed karyotype was studied in 33 selected phenotypic females and seven males and glucose 6-phosphate dehydrogenase analysis was carried out on the haemolysate of 37 females and six males.