True hermaphrodite: 46, XX/46, XY, clinical, cytogenetic and histopathological studies

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A case of unilateral ovo-testicular intersex in a five year old phenotypically male child referred for ambiguous genitalia is reported. General abnormalities included a prominent phallus, fusion of labia, bifid scrotum, absence of urethral opening and corpus. Gonads were not palpable. X and Y chromatin was found to be positive. Chromosome studies revealed 46, XX/46, XY karyotype. Laparotomy findings confirmed true hermaphroditism. The findings are compared and discussed with cases reported in the literature.

Key words: ambiguous genitalia; inter sex; hermaphrodite; mosaicism

The occurrence of sex chromosomal abnormalities in human is estimated to be 0.22% in newborns.1 The most common sex chromosomal abnormalities include XO (Turner's syndrome), XXY (Klinefelter's syndrome), XYY, XXX etc. due to chromosome non-disjunction during mitotic or meiotic cell division. Though 10-20% of all sex chromosomal aneuploids have been reported to be mosaics (XO/XX), the incidence of XX/XY mosaic or mosaic/chimera is very low. Relatively very few cases of true hermaphrodites (46, XX/46, XY) have been reported in the literature. So far no case of true hermaphrodite confirmed by detailed studies has been documented in our population.2 During the course of a survey on the cytogenetic studies in the human population of Ahmedabad, a patient with ambiguous external genitalia was referred. The investigations confirmed true hermaphroditism in the propositus.

Case history:

A five year old male child (Fig. 1) was referred for ambiguous external genitalia. The mother was 40 years and father 50 years at the time of birth of the proband. Genital abnormalities include a well developed phallus, absence of urethral opening, corpus, fusion of labia, bifid scrotum, and testes were not palpable. The presence of a vaginal canal was confirmed by dye test. At laparotomy a well developed unicorne uterus, fallopian tubes (bilateral), a left gonadovary and a right gonad-ovotestis were found to be present (Fig. 2). Histopathological study of the biopsy of the gonads was done.

As the clinical findings were suggestive of XX/XY karyotype, buccal
Fig. 1. (a) Clinical features of the propositus.
(b) External ambiguous genitalia.