Structural Abnormalities of the Y Chromosome and Abnormal External Genitals

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Summary. Three infants with different types of Y-chromosome anomalies, including short- and/or long-arm deletion and mosaicism, are reported. The karyotypes of these patients were: 45,X/46,X,del(Y)/47,X,del(Y), del(Y) on peripheral lymphocytes and 45,X/46,X, del(Y) on gonadal tissue (case 1), 45,X/46,X,del(Y) (case 2), and 45,X/46,X,r(Y) (case 3). In case 1 the euchromatic segment on the deleted Y was distinctly larger than that of the father’s Y.

The three infants had no gross phenotypic anomalies except ambiguous genitals and low birth weight, and they were small for date. The histologic diagnosis in two of them was mixed gonadal dysgenesis (cases 1 and 2).

The relationship between structural abnormalities of the Y chromosome and ambiguous genitals as well as male-determining factors is discussed.

Introduction

The Y chromosome in human beings belongs to the group of small chromosomes, its length being between that of F-group and G-group chromosomes. The distal two-thirds of the long arm of the Y chromosome shows characteristic quinacrine fluorescence patterns, and its length varies from individual to individual. This segment has been considered genetically inactive with regard to sex or testicular determination, but its function remains obscure.

Since the development of banding techniques, many authors have published studies of structural Y-chromosome abnormalities and of mixoploids with lack of a Y chromosome (Caspersson et al., 1971; Chandley and Edmond, 1971; Armendares et al., 1972; Meisner and Inhorn, 1972; Nielsen et al., 1972; German et al., 1973; Khudr and Benirschke, 1973; Neu et al., 1973; Berger et al., 1974; Bühler et al., 1974; Magnelli et al., 1974; Yanagisawa and Yokoyama, 1974; Fried et al., 1975; Winters et al., 1975; Wilson et al., 1976; Fonatsch et al., 1977; Lønberg et al., 1977; Yunis et al., 1977; Fass et al., 1978; Gaal et al., 1978).

We studied three patients with ambiguous genitals and structural Y chromosome anomalies with short- and/or long-arm deletion.

Case Reports

Case 1. A 16-month-old male infant was the first child of healthy and nonconsanguineous parents. The pregnancy was uncomplicated and delivery vaginal at term. At birth he weighed 2300g and was a normal baby except for bilateral inguinal hernias and ambiguous genitals. The external genitals were recognized as a small phallus without a urethral orifice in its center, a urogenital sinus, and a bifid scrotum, which contained no testicular elements (Fig. 1). Preliminary findings in this patient were reported elsewhere (Yanagisawa and Yokoyama, 1974).

Case 2. This 7-month-old female infant was born after 38 weeks gestation of an uncomplicated pregnancy to healthy parents who were second cousins. Her birth weight was 2400g. The external genitals were similar to those in case 1: there was a urogenital sinus, and a bifid scrotum, which contained no testicular elements (Fig. 1). Preliminary findings in this patient were reported elsewhere (Yanagisawa and Yokoyama, 1974).

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Case 3. This 2-month-old male infant was the first child of healthy nonconsanguineous parents. His birth weight was 1654g after 38 weeks gestation. No birth defects were seen except for abnormal external genitals: a bifid scrotum, cryptorchidism on the left side, a small phallus without a urethral orifice, and urogenital sinus (Fig. 3).
Fig. 1. External genitals; case 1

Fig. 2. External genitals; case 2

Fig. 3. External genitals; case 3

Fig. 4. Histology of gonadal tissues from case 1. A: infantile seminiferous tubules (right gonad, 100 x, H.E.); B: mesonephric remnants (left gonad, 100 x, H.E.); C: Fallopian tube (left gonad, 100 x, H.E.)

Fig. 5. Histology of gonadal tissues from case 2. A and B: seminiferous tubules in ovarian struma (right gonad, 100 x and 200 x, H.E.); C: infantile seminiferous tubules (left gonad, 100 x, H.E.)