51. A Case of XO-Male Newborn with Multiple Malformations

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The case which had testicles, normal male external genitalia and a 45,X karyotype was referred to as "XO-male". The only two cases of "XO-male" have been reported in the literature previous to this case (Lo Curto *et al.*, 1974; Foraboscol *et al.*, 1977). The present paper describes an additional case of the XO-male.

Case reports. The patient was a male newborn as the first child between a 25-year-old mother and a 26-year-old father. There was no history of abortion, still birth, congenital malformations, the exposure to the atomic bomb, and consanguinity in this couple. The clinical examinations revealed that his parents were phenotypically normal.

The birth weight of this patient was 1960 g at 38 weeks of gestation. His remarkable clinical signs were hypotonicity, respiratory disturbance, macrocephaly, antimongoloid slant, long palpebral fissures, hypotelorism, blepharophimosis, anophthalmos, saddle nose, low nasal bridge, small nose, single nostril, micrognathia, high arched palate, low-set ears, malformed auricles, short and web neck, shield chest with increased intermammary distance, normally rounded abdomen, normal male external genitalia with small penis and cryptorchism, anal atresia, abnormal palmar creases, and nail hypoplasia. Computed tomography revealed that his brain was a holoprosencephaly (Fig. 1).

Fig. 1. External pictures of the patient.

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Cytological findings. The chromosome preparations were made on the leucocyte cultures from the patient. The G- and Q-banding differential stainings were also applied for the chromosome identification. The chromosome counts were made with 61 well-delineated metaphases. Karyotypes were analysed in 32 cells by the conventional Giemsa staining, in 12 cells by G-banding, and in 17 cells by Q-banding analyses. The chromosome constitution of the patient based on the conventional Giemsa specimen showed 45 chromosomes. The G- and Q-banding analyses have revealed that the missing chromosome was one of the X chromosomes (Fig. 2). The Y-chromatin was not observed in the examination of 100 cells stained with the quinacrine mustard (Fig. 3, a, b). Then his chromosome formula was 45,XO. The chromosomal examination of his parents was not co-operated.

Fig. 2. G-banding karyotype of the patient (45,XO).

Fig. 3. (a) The chromosome of the patient, and (b) the cell nuclei showing without Y-chromatin stained with quinacrine mustard.

Remarks and conclusion. Remarkable clinical features of our case are the normal male external genitalia having a small penis and cryptorchism, and also some Turner-like stigmata showing short and web neck, shield chest with in-

creased intermamillary distance, low-set ears, abnormal nose, high arched palate, and micrognathia. This combination of the clinical features is very similar to that of previous reports (Lo Curto *et al.*, 1974; Forabosco *et al.*, 1977). The development of the clinical male features depends on the testicular differentiation of the embryonic gonad (Jost, 1970) and this testicular differentiation trigger with the Y chromosome (Hamerton, 1968). The only exception of this rule seems to be the male having XX-, or XO-karyotype. There are reviewed four theories to explain this exception by Bauch *et al.* (1980): The male differentiation in those cases is caused (1) by an undetected mosaic of cells with the XX and cells with the XY or the XXY gonosomes, (2) by an undetected translocation of a part of the Y-chromosome to an autosome, (3) by a crossover of Y- and X-chromosome leading to a transfer of Y-chromosomal information to the X-chromosome or (4) by a sex reversal mutation of an autosomal gene.

In our case it has not been possible to discover the Y chromosome and Y chromatin and one of the X chromosomes even by the use of the G- and Q-banding techniques, and also the mosaic cells in the leucocyte cultures.

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