117. Non-fluorescent Y Mosaicism in a Boy with Ambiguous Genitalia

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(Comm. by Sajiro MAKINO, M. J. A., Sept. 12, 1974)

The quinacrine fluorescence technique has demonstrated that the distal end of the long arm of the human Y chromosome is much more intensely fluorescent than any other chromosomal segment in the complement, which considerably facilitates the recognition of the Y (Zech 1969). Recently, however, there are reports of males having no such a fluorescent Y chromosome. Phenotypic manifestations of those patients have been known to be variable. An additional male patient with a non-fluorescent Y chromosome is preliminarily described in this paper.

Clinical findings. A 16-month-old male infant was referred to our clinic for ambiguous genitalia and bilateral hernias. The proband was the product of a normal pregnancy and delivery, with 2,300 g on birth, by a mother and a father aged 24 and 26 years, respectively. On examination, external genitalia were recognized as a penis-like phallus without orifice and urogeital sinus. Bifid scrotum contained no testicular element. He had no other physical anomalies.

Chromosome findings. Chromosome analyses performed on lymphocyte cultures disclosed the occurrence of 3 cell lines: a 45-cell line (16%) with a missing chromosome in the G-Y group; a 46-cell line (36%) with an apparently normal chromosome constitution; and a 47-cell line (44%) with a small extra acrocentric chromosome.

Differential staining procedures, G-, Q-, C-, and T-banding techniques (Seabright 1971, Caspersson et al. 1971a, Sumner 1972, Dutrillaux 1973), were applied in order to identify the missing and extra chromosomes.

The G-banding analysis showed the 4 small acrocentric chromosomes of the 45-cell line were nos. 21 and 22: the missing chromosome was the Y in this cell line. The 46-cell line showed a Y chromosome which had only one faint band on its long arm. Two small chromosomes of similar characteristics were seen in the 47-cell line (Fig. 1).

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Essentials of this article were presented before The First Asian Pediatric Congress held in Manila on April 30 to May 4, 1974.
The quinacrine fluorescence technique failed to demonstrate the Y chromosome having a characteristic bright fluorescent band in either 46- or 47-cell line. All the other chromosomes showed a normal fluorescent pattern; there was no overt evidence suggesting a translocation of the fluorescent segment of the Y chromosome (Fig. 2).

The C-banding method revealed no heterochromatic region on the long arm of the Y chromosome of the patient (Fig. 3).

The T-bands stained with acridine orange were normal in autosomes and the X. The Y chromosome had a medium bright fluorescent region on its short arm (Fig. 4).

In contrast to the findings in the patient, the Y chromosome of the father showed a brightly fluorescent Q-band on its long arm. This was confirmed by the C-banding method. In conformity with the above findings, the Y-chromatin of the father was positive in buccal cells and peripheral lymphocytes, while it was negative in cells of the patient.

The length of the Y chromosome relative to no. 14 was 0.605 in the patient, while 0.815 in the father. The ratio of euchromatic region of the Y to its whole length calculated in C-banded specimens of the father was 0.432.

It is evident, therefore, that the non-fluorescent segment of the
Fig. 2. Q-banding karyotype of the patient, from 47 cell line.

Fig. 3. A C-banding metaphase plate from the patient. Note the difference in heterochromatic region of the Y chromosomes between the patient (arrows) and his father (in insert).

Fig. 4. Partial metaphases showing Q-bands (a and c) and T-bands (b). a and b: from the patient. c: from his father.
Y was considerably longer in the patient than in his father, though the opposite is true with overall length. In the light of the above findings, it is assumed that the fluorescent abnormality of the Y chromosome of the patient can not be explained by a simple deletion of the long arm.

**Comments.** The human Y chromosome, though varies in length among different individuals, is usually transmitted unchanged from the father to his son. Since there is no evidence to suspect illegitimacy, we have to conclude that the abnormal Y had derived from the normal Y chromosome of the father. Unless we make a rather unlikely assumption that a structural rearrangement had turned the heterochromatic segment of the Y into a euchromatic one, a simple deletion or a post-zygotic translocation can not explain our cytogenetic findings in the present case.

Caspersson et al. (1971b) described 4 XO/XY patients with a non-fluorescent Y chromosome. A similar case has been reported by Lo Curto et al. (1972). As in the present case, the non-fluorescent Y of each patient was longer than the non-fluorescent part of the father's Y chromosome. In one of cases Caspersson et al. (1971b) found that the Y was associated with bivalent no. 2 in a considerable proportion of metaphase I cells, and interpreted that an unbalanced 2p/Yq translocation involving the whole brightly fluorescent segment of the Y had occurred in a germ-line cell of the father; fertilization of an ovum by a sperm bearing a normal no. 2 and a non-fluorescent Y may satisfactorily account for the karyotype status of this case. It follows then that a basically similar mechanism had been involved in the genesis of the present non-fluorescent Y chromosome, though the final conclusion must await meiotic studies in the patient and his father.

**In summary.** A 16-month-old male infant associated with a non-fluorescent Yq- mosaicism, 45, X/46, XYq-/47, XYq-Yq-, is described. The proband had ambiguous genitalia and bilateral inguinal hernias. The abnormal Y chromosome is analyzed by means of G-, Q-, C-, and T-bandling techniques. It is demonstrated that the Y chromosome of the patient is smaller than that of the father, and that the non-fluorescent part of the father's Y was considerably smaller than the whole non-fluorescent Y chromosome of the patient.

**Acknowledgments.** The author is grateful to Dr. Minoru Umeda for referring the case, Professor Shunzo Konishi for helpful advice, Emeritus Professor Sajiro Makino, Professor Motomichi Sasaki, and Dr. Nobuo Takagi for revising the findings.
References