FAMILIAL X;Y TRANSLOCATION IN A MALFORMED MALE INFANT AND HIS MOTHER

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Summary A male infant, the proband, with 46,Y,der(X),t(X;Y)(p22.3; q11.1), and his mother with 46,X,der(X),t(X;Y)(p22.3; q11.1) are presented. The proband was involved with a peculiar face, congenital heart disease, dry and scaly skin, and growth and psychomotor retardation. He died on the 111th day after birth. At necropsy a congenital heart disease was found, but there was no other major visceral malformation. The mother of the proband was healthy except for her short stature associated with disproportionately short limbs. Steroid sulfatase activity in her skin fibroblasts and lymphocytes was only half that of normal females.

INTRODUCTION

Translocation between X and Y chromosome is relatively rare. The majority of these translocations are those between Xp and Y. In cases having these translocations, life prognosis is generally good. In spite of this trend, however, the proband was involved with a marked growth and psychomotor retardation and congenital heart disease, and died on the 111th day after birth. No reports of X;Y translocation patients complicated with congenital heart disease or who died in infancy have been found.

CLINICAL AND CYTOGENETIC FINDINGS

Case 1

A male infant, the proband, was born on August 26, 1986 after 42 weeks of gestation as the second child to unrelated parents. The mother of the proband...
(Case 2) had had no experience of spontaneous abortion or stillbirth. When the patient was born, the father was 27, and the mother 23. Both his father and brother were healthy, and had normal karyotypes.

Fig. 1. Facial appearance of the proband at 15 days after birth.

Fig. 2. Complete G banded karyotype and partial Q banded karyotype of the proband.