41. Chromosome Translocation \( t(1p-; 17q+) \) in a Boy with Gingival Fibromatosis

By Jun-ichi AZUMI, Motomasa SASAKI, Kazuo YUKI, Yoshio KOMATSU, Shin-ichi KITA, and Sajiro MAKINO

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Recently developed banding techniques involving quinacrine mustard and Giemsa staining procedures have facilitated to a considerable extent precise and reliable analyses of structural abnormalities of chromosomes in mammalian cells and information has recently been increasing on the structure and nature of aberrant chromosomes in clinical and pathologic subjects of man.

While working with chromosomal surveys in patients with congenital oral diseases, we have chanced to observe a case showing a chromosome translocation between no. 1 and no. 17, or \( t(1p-; 17p+) \), in a boy with gingival fibromatosis. In this report, some preliminary accounts are presented on cytogenetic and clinical features of this patient.

Case report. The patient is a boy born in April 1967, after 40 weeks of gestation, to a 36-year-old father and a 24-year-old mother, both being phenotypically normal. He is a product of the fourth pregnancy of this couple, and weighed 3000 g at birth. The second pregnancy was aborted artificially. His two elder sisters were clinically normal. There are neither history of drug therapy or radiation exposure during the pregnancy in the mother, nor the occurrence of the same anomaly in the family. At the age of 4-1/3 years, he received surgical operations due to ankyloedactyly of the right thumb and hydrocele testis. Unusual overgrowth of the gingiva was recognized for the first time while he was 8 months old.

On physical examination at the age of 6 years, he was specially noted by major signs of gingival fibromatosis (Fig. 4), excessive hair-growth of the whole body, and peculiar face. He weighed 15 kg and his IQ was 107 at this age. Blood tests, endocrine function tests, urinalysis, chest roentgenograms, and electrocardiogram were within...
normal limits. Results of dermatoglyphic examinations in this patient are shown in Table I.

Table I. Dermatoglyphic patterns of the hands of the patient and parents

| Finger pattern | \( \begin{array}{cccc} \text{V} & \text{IV} & \text{III} & \text{II} & \text{I} \\ \text{Patient} & RL & W & UL & UL & D \\ \text{Mother} & UL & UL & UL & UL & D \\ \text{Father} & UL & W & UL & W & W \end{array} \) | \( \begin{array}{cccc} \text{I} & \text{II} & \text{III} & \text{IV} & \text{V} \\ \text{Patient} & W & UL & UL & W & RL \\ \text{Mother} & D & UL & UL & UL & UL \\ \text{Father} & D & W & UL & W & UL \end{array} \) | \( \begin{array}{c} \text{Total ridge count} \\ \text{Patient} & 133 \\ \text{Mother} & 131 \\ \text{Father} & 162 \end{array} \) |

Palm pattern

| Simian crease | Atd angle | Hypothenar pattern | \( a-b \) ridge count (sum of both hand) | \( t \) (%) |
|---------------|-----------|--------------------|---------------------------------|-------|
| \( \begin{array}{cc} \text{Left} & \text{Right} \\ \text{Patient} & + & + \\ \text{Mother} & - & - \\ \text{Father} & - & - \end{array} \) | \( \begin{array}{cc} 56 & 55 \\ 41 & 45 \\ 36 & 36 \end{array} \) | \( \begin{array}{cc} \text{Arch} & \text{Loop} \\ \text{unlar} & \text{ulnar} \\ \text{unlar} & \text{unlar} \end{array} \) | \( \begin{array}{cc} 70 & 35.4 \\ 78 & 15.6 \\ 56 & 27.2 \end{array} \) | \( \begin{array}{cc} 39.8 & \text{(?)} \\ 20.8 & \text{(?)} \\ 25.4 & \text{(?)} \end{array} \) |

Cytogenetic findings. Chromosomal investigations were undertaken in the patient, his parents and two elder sisters following the routine blood-culture method involving the air-drying and Giemsa-staining procedures. Quinacrine fluorescence technique after Caspersson et al. (1970) was applied to identify the aberrant chromosomes and analyse their structure.

It was found that all family members except the patient had 46 normal chromosomes consistent with the phenotypic and chromosomal sex. In contrast, the chromosomes of the patient showed a remarkable divergence from a normal complement. It was shown in 30 metaphases from the routine Giemsa-slides that each one member of pair no. 1 and pair no. 17 was missing, while they were replaced by two unusual chromosomes, one by a B-like element and the other by a C-like one. The fluorescence banding analysis in this case disclosed a presumable reciprocal translocation occurring between the short arm of a no. 1 chromosome and the long arm of a no. 17 chromosome at \( t(1p--; 17q+) \) (Figs. 1–3). Mensural examinations on 15 metaphasic cells detected no appreciable difference between the amount of material deleted from no. 1 chromosome \( (1p--) \) and the additional segment translocated on no. 17 \( (17q+) \). Following the Paris nomenclature system, the translocation was found to occur at the points of
no. 1p31 and no. 17q24. Thus this patient was characterized by a karyotype of 46, XY, t(1; 17) (p31; q24).

Remarks. The cytogenetic literature indicated several cases of congenital anomalies with translocations between chromosome no. 1 and other elements. Maganias et al. (1967) described a mentally retarded girl with signs of Turner’s syndrome who had a balanced 1/G translocation with 46 chromosomes. They interpreted the mild phenotypic anomalies of this girl as being due either to a deletion of

Fig. 1. Quinacrine fluorescence karyotype of the patient, showing 1/17 translocation.

Figs. 2-3. Partial karyotypes of groups 1 and 17, illustrating t(1p−;17q+).
a few loci at the breaking points or to a position-effect phenomenon. Kontras et al. (1966) reported a family in which the transmission of a balanced 1/G translocation was observed during through 4 generations. The G-group chromosome involved in this family was evidently no. 21, because 4 children in this family were affected with Down’s syndrome. No confirmative studies have been done by means of fluorescence microscopy or of other banding techniques.

Recently, Neu and Gardner (1973) described a partial trisomy of chromosome no. 1 in a family with a t(1q−; 4q+) translocation. Berghe et al. (1973) mentioned a malformed newborn infant with a partial trisomy of a chromosome no. 1 due to a balanced familial translocation with a t(1q−; 12p+). Very recently, in an incidence study of chromosome abnormalities among 5049 consecutive liveborn children at a Danish maternity hospital, Friedrich and Nielsen (1974) found two familial cases with translocations between the long arm of no. 1 and the long arm of no. 17, or t(1q−; 17q+). In one family, the karyotype 46, XY, t(1:17) (q21; q21) was found in a boy, his father and a paternal relative, and in another the karyotype 46, XY, t(1;17) (q12; q25) was observed in two boys. None of the probands and their relatives with such translocation showed physical or mental anomalies. The translocation t(1p−; 17q+) in the present case seems to be the first record as a human chromosome error. Identification of the abnormal chromosome in the above cases was performed by fluorescence microscopy or Giemsa-banding techniques.

Gingival fibromatosis is an infrequent oral disease in which teeth were mostly covered by the hyperplastic gingival tissue. In familial studies it has been reported that the condition is transmitted as an autosomal dominant trait, and that the transmission is through an affected parent (Emerson 1965, Witkop 1965). Previously no cytogenetic report on gingival fibromatosis has been referred to in
the literature. The fact that both parents have a normal karyotype suggests a sporadic origin for the reciprocal t(1p-; 17q+) translocation in this patient. The implication of this chromosome aberration in the clinical condition has remained unclear.

Summary. This paper reports clinical and cytogenetic features of a presumable reciprocal translocation between no. 1 and no. 17, or t(1p-; 17q+), found in a boy with gingival fibromatosis. Karyotype 46, XY, t(1; 17) (p31; q24) was given to this patient. His parents and two sisters are clinically and karyotypically normal.

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