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SHORT COMMUNICATION

CONSTITUTIONAL Y/15 TRANSLOCATION IN A WOMAN WITH LUNG CANCER

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ABSTRACT

In the course of fragile site investigations on 16 patients with leukemia, lymphoma and lung cancer, a 74-year-old woman with lung cancer was found to have additional heterochromatic material on 15p. G, C, Q and Ag-NOR banding suggested that an important part of the long arm of the Y chromosome was translocated to the short arm of chromosome 15.

INTRODUCTION

It has been known for some time that certain constitutional chromosome abnormalities are associated with high susceptibility to cancer. The best established examples are retinoblastoma with del (13) (q14) and aniridia-Wilms' tumor with del (11) (p13) (Dallapicola, 1987). Therefore the cytogenetics studies may be useful for delineating individuals with an increased tumor risk.

The purpose of the present report is to describe a case with a constitutional Y/15 translocation in a patient with lung cancer.

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CASE REPORT

A 74-year-old, markedly dyspneic, white woman underwent bronchoscopy with biopsy and the results were suggestive of squamous cell carcinoma. Examination revealed a normal female phenotype and normal intellect. There were neither toxic nor infectious antecedents and she had never smoked. She had received chemotherapy and died of respiratory insufficiency in January, 1988.

The patient had six sibs. Her brother died from colon cancer, and two sisters died from uterine carcinoma and pancreas cancer, respectively. Her husband is 78-years-old and healthy like all other family members. One maternal uncle and one nicce were sterile but could not be examined.

The patient had only one child who was given in adoption and the family has no information about him.

MATERIALS AND METHODS

Chromosome investigations were carried out on peripheral blood cultures. PHA stimulated lymphocytes were examined after GTG, QFQ, CBG and NGG banding. It was not possible to study the tumor cells from the patient and the chromosomes of other family members.

RESULTS

G banded metaphases showed a stained mass on 15p with two dark bands of distal Yq morphology (Figures 1A, B). Q, C and NOR banding showed that the extra material was Q and C band positive and NOR negative (Figures 1C, D, E, F). The brightly fluorescent Q positive segment indicated the presence of Yq on autosome 15 (Figures 1D, E). The karyotype was thus established as 46,XX, -15, +der (15), t (15;Y) (p11; q12).

DISCUSSION

Translocations between Y and acrocentric chromosomes have been reported and the most common type is clearly Y/15 (Fryns *et al.*, 1985). The incidence of these translocations in the general population was mentioned by Nielsen and Rasmussen (1976) as approximately 1 in 2,000. Smith *et al.* (1979) considered data from these authors and from other major newborn studies and found a rate of 0.1 per thousand in the population. Y/non-acrocentric autosome translocations must be extremely rare, since none were found in these newborn studies. The relatively high frequency of Y/15 translocation needs to be considered cautiously because some of these

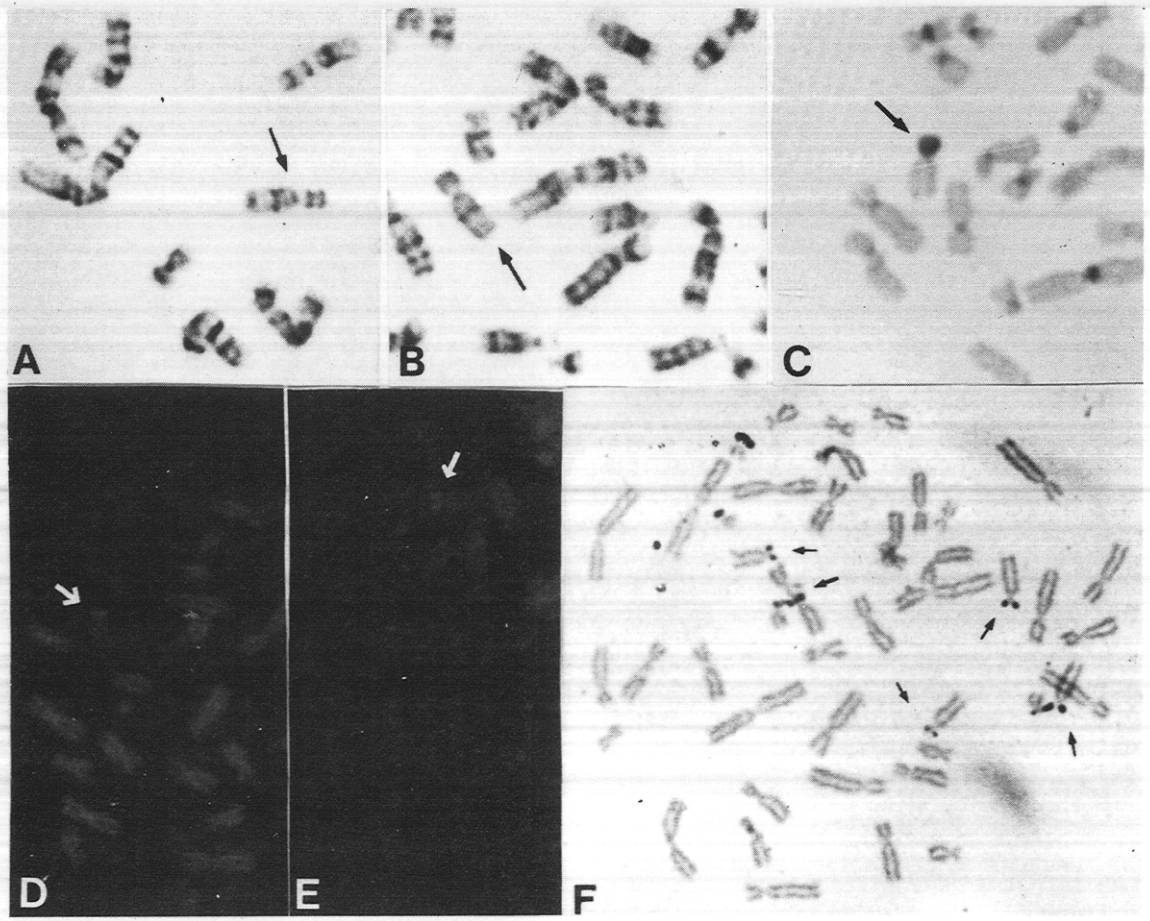

Figure 1 - G-banded (A, B), C-banded (C) and Q-banded (D, E) partial metaphases showing the Y/15 translocation. Ag-NOR stained metaphase (F) of the patient showing five group D NOR-bearing chromosomes.

cases may represent enlarged short arms or satellite variants and not Yq material (Spowart, 1979).

Most of the cases with Y/acrocentric translocation have been ascertained because of some phenotypic abnormalities but most of the carrier relatives of affected probands have normal phenotypes. Thus, these chromosomal aberrations appear not to affect the phenotype (Alitalo *et al.*, 1988).

At least five cases of Y/autosome translocations, including the present one, were observed in patients with non-gonadal neoplasia (Funderburk *et al.*, 1982; Alimena *et al.*, 1985; Benitez *et al.*, 1987; Moreau *et al.*, 1987). Alimena *et al.* (1985) found 21 patients with constitutional chromosome anomalies representing approximately 1.5% of their patients with malignant hematologic disorders. One of them showed a t (Y;17) which represents 0.07-0.08% of the sample. Benitez *et al.* (1987) found one patient with t (Y;15) (0.1%) in their series of 718 patients with different hematologic diseases. When these authors considered data from other series of patients with the same disorders, including Alimena's series, the frequency of Y/autosome translocations was 0.04%. If we add our series which also presents patients with solid tumors, the frequency is about 0.05%, slightly higher than in newborn studies.

The lung cancer observed in our patient may be coincidental to the Y/15 translocation but evidence for this would require more extensive investigations on families with similar chromosome aberrations.

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RESUMO

O presente trabalho descreve uma paciente portadora de câncer de pulmão que apresenta material heterocromático extra no braço curto do cromossomo 15. A análise em bandamento G, C, Q e Ag-NOR sugere uma translocação envolvendo o braço longo do cromossomo Y e o braço curto do cromossomo 15.

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