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INCREASED NUMBER OF CHROMOSOME ABERRATIONS IN THE PERIPHERAL BLOOD CULTURE OF A RETINOBLASTOMA PATIENT

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Chromosome studies were performed on blood lymphocytes from an eight-year-old patient whose left eye had been enucleated earlier because of retinoblastoma. GTG-banded karyotypes showed both numerical and structural chromosome aberrations, and the number of the patient's lymphocytes with chromosome aberrations increased. It was concluded that retinoblastoma survivors need continuous control because of the increased risk of second primary tumors.

INTRODUCTION

Retinoblastoma is the most common eye tumor in childhood /3/ with an incidence of one new case per 15.000 - 18.000 live-born children /10,17/. This type of tumor can be unilateral or bilateral, and hereditary transmission can be detected in some cases /3,11/. The conventional treatment of retinoblastoma is the enucleation of eyes with large tumors, and the radiotherapy of eyes with small tumors /17/. The survival rate of children with retinoblastoma is better than 90 % /17/. Cytogenetic analyses of retinoblastomas have shown that the most characteristic abnormality is deletion of the long arm(q14) of chromosome 13. /4-6, 13-16, 18, 19/.

Other changes have also been found as trisomy of 1q isochromosome (6p), del (6q), monosomy of chromosome 16, 1p+ marker chromosome and the presence of homogeneously staining regions (HSR) and double minutes (DMs) /4,13/. The retinoblastoma survivors have an enhanced risk of second

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primary cancers /7,8,12/.

In this work, we have investigated the chromosome content of a peripheral blood culture from an eight-year-old girl, one of whose eyes had earlier been enucleated because of retinoblastoma.

An increased number of the patient's lymphocytes with chromosome aberrations was detected.

MATERIALS AND METHODS

Case report

The patient's (K.Sz.; 10.09. 1978.) left eye was enucleated because of retinoblastoma at 2 and half years of age. No other treatment was performed. Later control investigations (two examinations per year) showed no signs of metastasis or a second tumor. The present status of the patient is good and her hematological picture is without abnormalities.

Chromosome examination

Chromosome studies were performed on blood lymphocytes from the patient by the routine microculture technique of Autio and Schröder /2/. The mitoses were examined under a microscope at a magnification of 1600 X. GTG-banded karyotypes were analysed according to the ISCN /9/.

RESULTS

Table I shows the chromosome examination data. It can be seen that seventy-three (91.2 %) of the eighty cells examined contained 46 chromosomes, while seven mitoses (8.8 %) were hypodiploid.

Table II relates to the karyotype analysis data. Twenty metaphases were examined: eight metaphases were normal, while twelve karyotypes contained numerical or structural aberrations. Three karyotypes showed numerical aberrations; all of them contained 45 chromosomes, and one of the chromosomes 6, 7, 8, and X was lost in these cases. The characteristic structural aberration in retinoblastoma, del (13) (q14) was found only in four mitoses. Other deletions, such as 1q-; 5p-; 6p-; 10q- and 16p-, were present too. The most frequent translocation

TABLE I

Chromosome examination data of the patient's lymphocytes

Number of cells examined	Number of cells with numerical aberrations	Chromosome number in the metaphases examined				
80	7	41	42	44	45	46
		1	1	1	4	73

TABLE II

Numerical and structural chromosomal aberrations

Type of aberration	Number of mitoses
45,XX,-6	1
45,XX,-7,del/5//p14→pter/, t/9;?//q34;?/, t/12;12//p13;q24/	1
45X,-X,-8,+mar der/8/ t/1;8//q24;p23/	1
46,XX,del/1//q41→ter/	1
46,XX,del/13//q14/	2
46,XX,del/13//q14/,t/9;?//q34;?/	1
46,XX,del/16//p13→pter/	1
46,XX,t/9;?//q34;?/	2
46,XX,del/6//p11→pter/,del/10//q25→ter/, del/13//q14/,t/9;?//q34;?/	1
46,XX,t/11;12//p13;q24/	1

(in five mitoses) was t(9;?) (q34;?), which resulted a 9q+ chromosome. The translocations t(1;8) (q24;p23); t(11;12) (p13;q24) and t(12;12) (p13;q24) were each found in a single case.

DISCUSSION

Deletion of chromosome band 13q14 is the characteristic aberration for retinoblastoma /4-6, 13-16, 18, 19/, the most common intraocular tumor in childhood /1/. Retinoblastoma occurs unilaterally in about 65 % of the cases and the majority of patients have a negative family history for this tumor /1/. Our patient had an unilateral retinoblastoma without a hereditary basis.

Because of the increased risk of the development of second primary malignancies in patients with retinoblastoma /1,7,8,12/, chromosomal examinations were performed on blood lymphocytes from the patient. Only seven (8.8 %) of the eighty cells examined under the microscope were hypodiploid. The remaining seventy-three cells (91.2 %) contained a normal human chromosome number. From the twenty mitoses karyotyped, the characteristic deletion 13q14 was seen in four karyotypes.

Twelve karyotypes showed numerical or structural chromosome aberrations, which represents an increased number of the patient's lymphocytes with chromosome aberrations.

In five karyotypes, an unidentified 9q+ marker chromosome was found (Fig.1).

It can be concluded that retinoblastoma survivors need continuous control for the early diagnosis of second primary tumors, to which they have an increased risk.

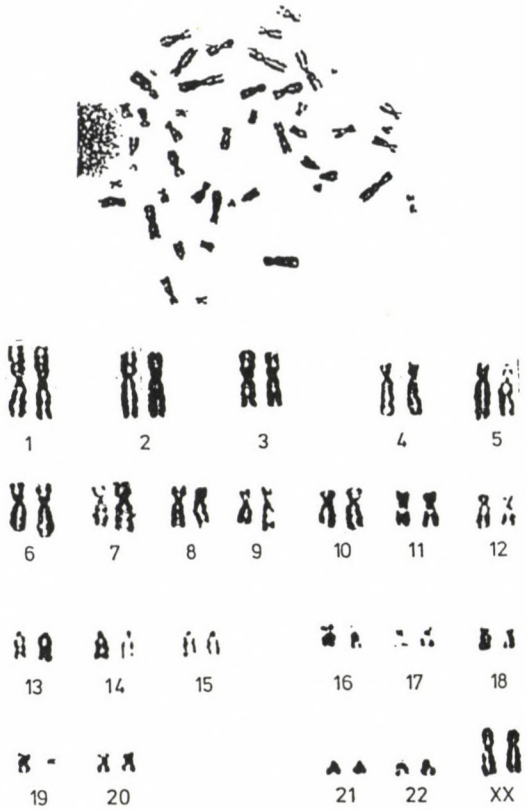

Fig 1. The karyotype shows the characteristic translocation t/9;?//q34;?/ was found in five mitoses of a retinoblastoma patient.

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