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Deletion 13q12.1 in a child with Coats disease

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Cytogenetic study of a child with the presumed clinical diagnosis of retinoblastoma of the right eye revealed del. 13q12.1. Histological examination of the removed eye showed changes which were characteristic of Coats disease. This finding is discussed.

The aetiology of Coats disease is unknown. It results from vascular anomalies in the retina (teleangiectasies, microaneurisms, etc.) and is fatal for the vision despite the benign character of the process. Other authors suggested the role of vascular degenerative lesions [2]. So far, genetic studies were not undertaken regardless of data showing the hereditary nature of the disease with predominant affliction of males in about 80% of all cases.

The aim of the present study was to explain the cytogenetic finding of del. 13q12.1 found in a child with Coats disease.

Report of a Case

The propositus (L.S.H.), a three years old male was referred to the Children's Eye Clinic because of squinting of the right eye and glistening of the pupil. The presumed clinical diagnosis was retinoblastoma of the right eye and the child was sent to us for cytogenetic examination.

Enucleation was performed. Histological examination of the nerve cells revealed total retinal detachment with subretinal exudate containing cholesterol crystals, pigment clusters and single foam cells.

The final diagnosis was Coats disease.

Chromosomal study of the patient and his parents was performed by means of peripheral blood leukocyte cultures. For high-resolution G-banding prometaphase analysis, chromosomes were obtained by a slight modification of the technique of Yunis [4, 5].

Results

At least 50 well-spread early metaphase and prometaphase plates were analysed.

Cytogenetic study revealed normal karyotypes of the parents. High resolution analysis of the patient's karyotype showed shortening of the white band 13q12 (Figs 1, 2). At the stage of 600 G-band chromosomal

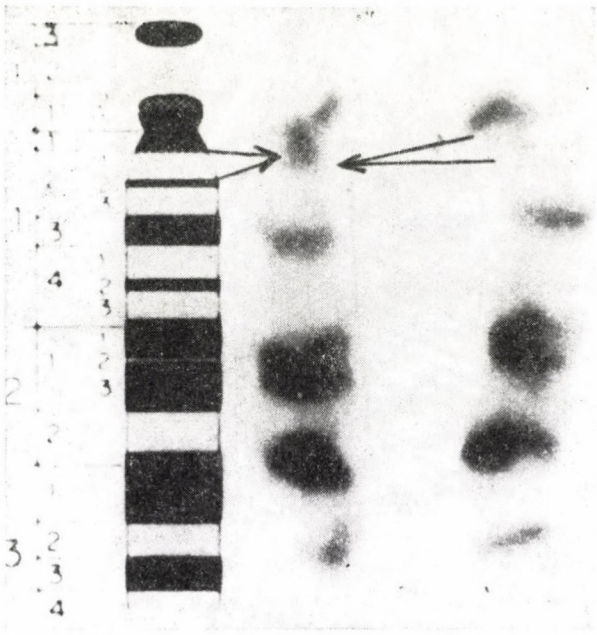

FIG. 1. The patient's chromosomes 13

FIG. 2. Chromosome 13 pair from patient with deleted chromosome 13 (del 13q12.1) on the left of the pair

differentiation it was identified as del. 13q12.1.

Discussion

To the best of our knowledge, del. 13q12.1 has never been reported in Coats disease. The present finding was casual and we have not studied other patients with Coats disease. For this reason it is difficult to comment upon the finding. Nevertheless, we suppose that the chromosomal abnormality 46,XY, del. 13q12.1 could be related to the child's disease. Chromosome 13 was namely found a major pathogenetic factor in blastomogenic processes: acute myelogenous leukaemia (del 13q12–14); myelofibrosis; Ph(—) chronic myelogenous leukaemia [3], and lymphoma and meningeoma [1], and deletion 13q14 has been accepted as a specific genetic marker for retinoblastoma.

We assume that the segment 13q12 might somehow occur in the eye's morphogenesis. New data over a large body of material and other kinds of eye tumours will help to establish the proper importance of chromosome polymorphism on which the clinical varieties of the ocular tumours are based.

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