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A case of 22-trisomy mosaic

By

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An extra small acrocentric chromosome was found in 50% of the cultured blood cells of a somato-mentally retarded girl with congenital heart failure and different dysmorphic symptoms. The supernumerary chromosome proved to be chromosome No. 22.

Report of a Case

This female child was born from the second pregnancy of a 30-year-old mother and a 34-year-old father. There were no spontaneous abortions and the family history was uninformative. After an uncomplicatad pregnancy the baby was delivered at term, with 3225 g weight and 52 cm length. Physical and mental development was delayed and therefore the patient was referred to us for chromosome investigation at the age of 18 months. At that time her weight was 8950 g; the length 73 cm; and the head circumference 43.5 cm. She was not able to stand without help.

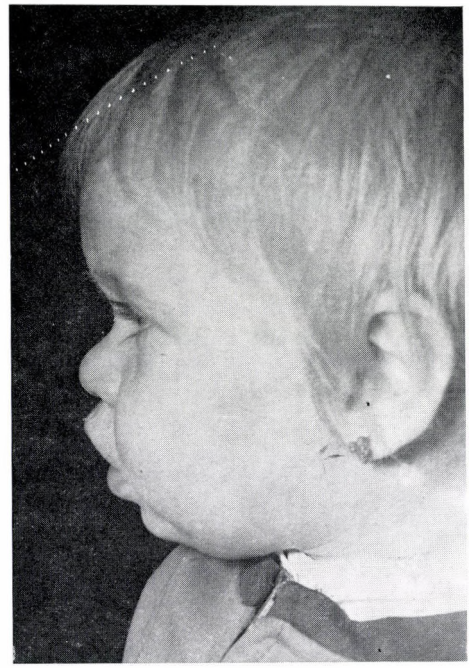
Except for a congenital stenosis of the aorta, she had no visceral malformation. Dysmorphic signs were microcephaly, asymmetry of head with left frontal and parietal prominence, strabismus, beaked nose with low nasal bridge, long philtrum, slight micrognathia, gothic palate, asymmetry of the ears and hypoplastic nails (Fig. 1).

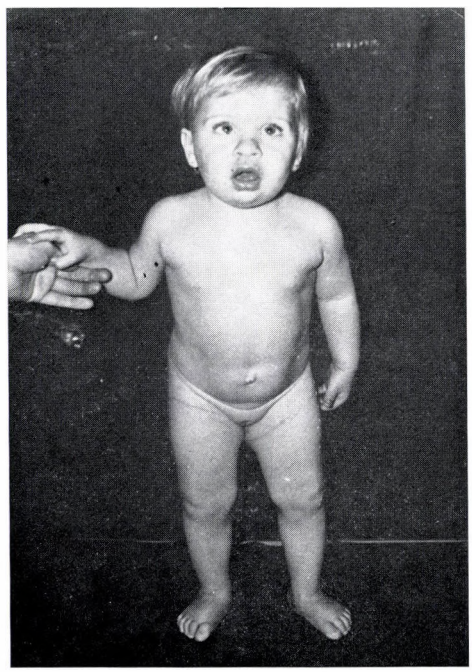
Chromosome Study

Chromosome study revealed 46, XX karyotype in 23 investigated peripheral blood cells of the patient, while in 21 cells a 47, XX, +22 karyotype was found (Fig. 2). The parents and their first child have normal karyotypes.

Discussion

Trisomy 22 seems to be a rare anomaly and the question frequently arose whether it was connected with the cat eye syndrome. Before the banding era, 22-trisomy was considered a clinical entity [6]. Its cardinal symptoms consisting of mental and growth retardation, microcephaly, micrognathia, preauricular skin tag, appendage and sinus, congenital heart defect, cleft palate, were found also in cases where chromosome identification was carried out by new chromosome techniques [1, 2, 3, 5, 8, 9, 10]. The clinical features of 22-trisomy have recently been reviewed by Begleiter et al. [3].





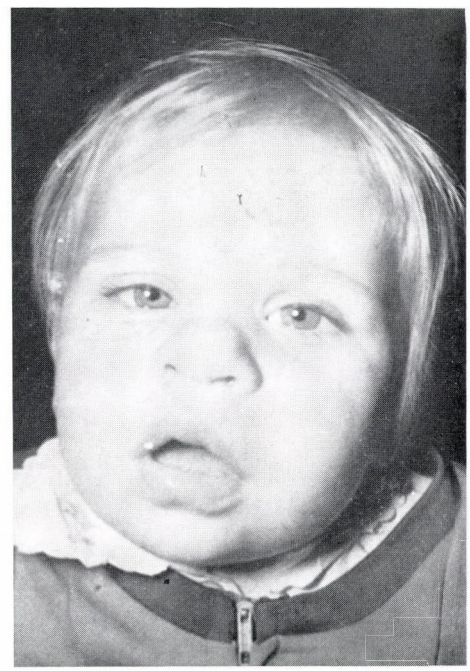


FIG. 1. The patient at 18 months of age

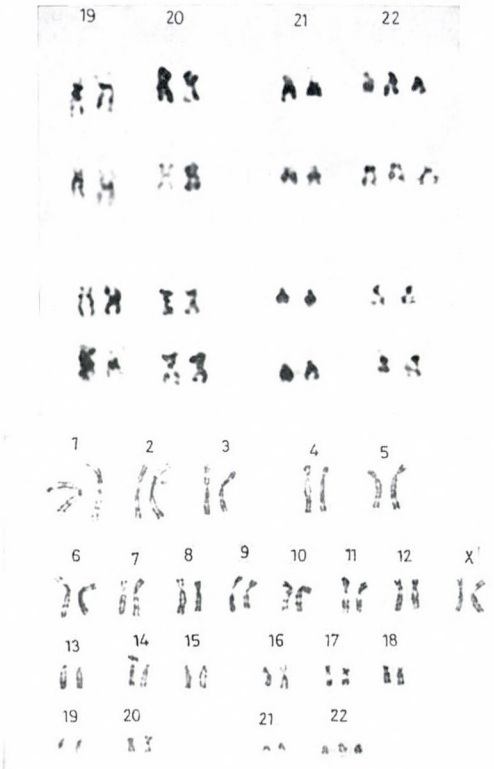


FIG. 2. (a) Chromosomes No. 19—22 in trisomic and normal cells; (b) karyotype in a cell with 22-trisomy. Identification by GAG technique

In a comprehensive study [7] the inconsistency of the phenotypic manifestation of the cat eye syndrome has been emphasized there having been one single case only where the supernumerary chromosome was identified as No. 22 [4]. All the other supernumerary chromosomes found in the cat eye syndrome remained unrevealed even when identification was attempted by several banding methods [7].

It has been assumed [10] that 22-trisomy and the cat eye syndrome are variants of the same entity. If this is true, the supernumerary chromosome must be a deleted chromosome No. 22 due in some cases to a pericentric inversion or translocation. This would explain the divergence in the clinical manifestations of the cat eye and 22-trisomy syndromes and also the experience that mental retardation is more serious in patients with 22-trisomy.

In spite of the fact that in our patient the 22-trisomy involved only 50% of the blood cells, she is severely retarded mentally and shows many symptoms of those common cases reported previously. In external appearance she resembles especially the

patients reported by Penchaszadeh and Coco [8].

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