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Hemihypertrophy: Incidence and Chromosomal Examinations

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INTRODUCTION

It was shown previously that, according to the literature and our own observations [27, 28, 29] hemihypertrophy has become increasingly frequent in the last decades (Table I). Although several authors have mentioned chromosomal aberrations in this connection, the aetiology of the anomaly is still unelucidated. Since 1965, we have made chromosomal examinations in six further cases of hemihypertrophy, and their results are reported below.

METHOD

In 6 apparently healthy children displaying hemihypertrophy, the chromosomes were examined in 72-hour cultures of peripheral blood by a slightly modified MOORHEAD method [43, 59].

RESULTS

Results of clinical observations and of chromosomal examinations in the present material are listed in Table II. It can be seen that no chromosomal anomaly was found in any of the six cases of congenital hemihypertrophy.

DISCUSSION

Congenital hemihypertrophy appears in early intrauterine life; it has been described in a 10-mm embryo [50] and may be supposed to occur even earlier. Such observations and the fact that the anomaly may involve several organs, the lack of other aetiological factors suggest that chromosomal changes in the germ cell or rather in the zygote may be found. Reports are contradictory in this respect (Table III). In six cases chromosomal aberrations were detected, while in 34 cases no chromosomal abnormality was found [5, 8, 19, 22, 24, 38, 47, 51, 64, 67]. Of these negative cases it was in nine that fibroblasts were cultured [38] and in one case that blood as well as fibroblasts were examined [67], while only peripheral blood cells were cultured in the remaining 24 cases. Our previous investigations have left the questions undecided whether the observed chromosomal irregularities were of a pathol-

TABLE I

Year		Number of cases	Own cases	Average number of cases reported per year
1839—1947		107*		1
(108 years)				
1947—1961	1947—1954	55**		4
(14 years)	1954—1961		11([28])	
1961—1968	1961—1964	71***	10([27])	10
(7 years)	1964—1968		14([30])	

References: * 56, 66

** 1—3, 7, 10—12, 21, 28, 33, 35, 40, 42, 44, 45, 48, 49, 52, 55, 57, 58, 60, 62, 65, 68.

*** 4—6, 13—18, 22, 24—27, 30—32, 37, 39, 41, 46, 47, 53, 54, 61, 63.

TABLE II

Material of present study

No.	Initials, sex	Age	Hemihypertrophy				Number of chromosomes				Chromosomal aberration
			Time of detection	Side	Difference in extremital length UE	LE	Associated disorders	45	46	47	total	
1	I. D. ♂	5 days	Birth	left	25	30	Klippel–Trenaunay syndrome; haemangiomatosis	—	12	—	12	∅
2	V. M. ♀	14 yrs	Birth	left	35	30	Renal dystrophy (left)	1	18	1	20	∅
3	M. J. ♀*	5 yrs	2 years	left		5	Congenital heart defect	—	19	1	20	∅
4	M. T. ♂	1 year	Birth	left	2	3	—	3	17	—	20	∅
5	E. B. ♀	10 yrs	5 years	left	2	10**	Exostosis on left tibia	3	17	—	20	∅
6	Z. R. ♂*	4 mths	2 months	left	3	4	—	3	27	—	30	∅

UE = upper extremity (mm) * = twins

LE = lower extremity (mm) ** = associated with facial asymmetry

TABLE III

Occurrence of chromosomal aberrations in hemihypertrophy

Age (yrs) sex	Tissue examined	Chromosomal aberration	Diagnosis	Published by
6 ♀	Fibroblast (b).	Mosaicism (diploid-triploid)	Hemihypertrophy; slight mental retardation; zygodactyly	ELLIS et al. [20]
	Blood	Normal		
10 ♂	Fibroblast (b) and fascia lata	Mosaicism (diploid-triploid) and large satellite on a large acrocentric chromosome	Hemihypertrophy; mental retardation; cryptorchidism	FERRIER et al. [23]
	Blood	Normal		
1 ♀	Fibroblast (b)	Mosaicism (trisomy 18 and normal)	Hemihypertrophy; slight microcephaly; mental retardation; torticollis; ventricular septal defect; double ureter and calices on left side	HOOK and YUNIS [36]
11½ ♀	Fibroblast (b)	4—5/21—22 translocation; besides, trisomy 2 and monosomy C on right side (XO ?)	Hemihypertrophy; atrophy of skin on left hand; mental retardation; congenital heart defect	BROGGER et al. [9]
10 ♀	Blood	Mosaicism (one of chromosomes 3 longer and normal)	Hemihypertrophy	GERLÓCZY et al. [29]
11½	Blood	Chromosomes 16 elongated	Hemihypertrophy; Wilms' tumour; increased gonadotrophin secretion	FRAUMENI et al. [24]

b = skin on both sides of the body

ogic nature and whether they were correlated with the hemihypertrophy. The chromosomal analysis performed in the present six cases failed to reveal any change in either the morphology or the number of chromosomes. It is improbable therefore, that there should exist a correlation between the morphology of the chromosomes and the syndrome. Of course, examination of peripheral blood leaves the possibility open that chromosomal malformations may exist in other tissues or organs. The 46 cases reported by other authors, with chromosomal changes only in six and our own observations lead nevertheless to the conclusion that a causal connection between chromosomal aberrations and hemihypertrophy is improbable. The

condition is frequently associated with other congenital anomalies, e.g. with Wilm's tumour [24]. It is therefore only natural that chromosomal aberrations may occur in patients with hemihypertrophy without necessarily constituting an aetiologic factor in the condition.

SUMMARY

In six cases of hemihypertrophy no chromosomal aberrations were found. The incidence of hemihypertrophy is increasing. Literary data have been reviewed concerning chromosomal examinations in hemihypertrophy.

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