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Psychoinfantilism and Chromosome Abnormalities

By JOHANNES NIELSEN and NIELS THOMSEN

The following study has been made on the background of the finding by Kramp and Nielsen (1968) of enlarged short arms of a chromosome in the 21–22 group in four out of five psychoinfantile members of a family with a case of Down's syndrome.

Material and Method

All female patients with psychoinfantilism in a psychiatric hospital with 340 beds for female patients were chosen as probands; 21 such patients were found, giving a frequency of 6·2 per cent. We got a blood sample for chromosome analysis from 20 patients; one patient was discharged before chromosome analysis could be made. The mean age of the patients was 34·9 years. Twenty age-matched female controls with no mental disorder were chosen for chromosome analysis among the staff of the hospital; the mean age of this group was 34·5 years.

Sex-chromatin analysis was made on Feulgen stained buccal smear, and chromosome analysis was made of leucocytes cultured according to the method described by Moorhead *et al.*, slightly modified; 30 to 65 metaphases were counted, and 15 metaphases with the modal figure as well as all metaphases with aneuploid cells were analysed. We looked for minor abnormalities in all chromosomes, but we were

Table I

Chromosome analysis in patients with psychoinfantilism

Distribution by chromosome number, total number of cells counted, Karyotype and enlarged short arms of a chromosome in group 21–22

Case no.	Age	Chromosome number					Total number of cells	Karyotype	Enlarged short arms of a 21–22 chromosome
		44	45	46	47	48			
1.	60	—	6	51	2	—	59	46,XX	
2.	35	2	1	30	—	—	33	46,XX	
3.	25	1	1	49	—	—	51	46,XX	+
4.	32	1	—	32	—	—	33	46,XX	
5.	20	—	—	57	1	—	58	46,XX	
6.	29	—	—	30	—	—	30	46,XX	
7.	27	—	3	58	—	—	61	46,XX	
8.	28	—	—	63	—	—	63	46,XX	
9.	19	1	2	28	1	—	32	46,XX	
10.	35	1	2	58	2	—	63	46,XX	+
11.	31	—	2	29	—	—	31	46,XX	
12.	29	—	—	32	—	—	32	46,XX	
13.	43	—	—	32	—	—	32	46,XX	
14.	53	1	42	2	—	—	45	45,XO	
15.	35	—	—	30	—	—	30	46,XX	
16.	40	—	—	32	—	—	32	46,XX	
17.	22	—	1	31	—	—	32	46,XX	
18.	57	—	3	29	—	—	32	46,XX	
19.	33	—	—	32	—	—	32	46,XX	
20.	44	—	2	59	1	—	62	46,XX	+
Total		7	23	764	7	0	843		
Percentage		0·8	2·7	90·6	0·8	0			

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TABLE II

Chromosome analysis in controls

Distribution by chromosome number, total number of cells counted, Karyotype and enlarged short arms of a chromosome in group 21–22

Case no.	Age	Chromosome number					Total number of cells	Karyotype	Enlarged short arms of a 21–22 chromosome
		44	45	46	47	48			
21.	55	1	1	29	—	—	31	46,XX	+
22.	38	—	2	30	—	—	32	46,XX	
23	25	—	1	29	—	—	30	46,XX	
24.	33	—	2	47	1	—	50	46,XX	
25.	24	—	—	32	—	—	32	46,XX	
26.	29	—	1	29	—	—	30	46,XX	
27.	25	—	—	32	—	—	32	46,XX	
28.	27	—	1	45	—	—	46	46,XX	+
29.	20	—	—	32	—	—	32	46,XX	
30.	32	—	—	30	—	—	30	46,XX	
31.	31	—	1	29	—	—	30	46,XX	
32.	25	—	—	31	—	—	31	46,XX	
33.	44	—	1	29	—	—	30	46,XX	
34.	53	—	3	58	1	—	62	46,XX	
35.	34	—	—	31	—	—	31	46,XX	
36.	40	—	—	32	—	—	32	46,XX	
37.	22	—	—	32	—	—	32	46,XX	
38.	57	—	1	30	—	—	31	46,XX	+
39.	31	—	—	51	1	—	52	46,XX	
40.	44	—	—	31	—	—	31	46,XX	
Total		1	14	689	3	0	707		
Percentage		0·1	2·0	97·5	0·4	0			

specially looking for enlarged short arms of the chromosomes in group 21–22.

RESULTS

The result of the chromosome analysis in the 20 patients with psychoinfantilism and the 20 controls is shown in Tables I and II. One patient, No. 14, had a chromosome constitution of 45,X (Turner's syndrome) and chromatin-negative cells with only 1 per cent chromatin-positive cells in buccal smear. Three patients, Nos. 3, 10 and 20, and three controls, Nos. 21, 28 and 38, had enlarged short arms of a chromosome in the 21–22 group. Patient No. 14 with Turner's syndrome has previously been described (Nielsen, 1970).

DISCUSSION

Our findings of an equal number of psychoinfantile patients and controls with enlarged short arms of a 21–22 chromosome indicate that there is no connection between psychoinfantilism and enlarged short arms of a 21–22 chromosome as indicated by the study by Kramp and Nielsen (1968).

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Johannes Nielsen, M.D.,
Niels Thomsen, M.D.,

The Cytogenetic Laboratory, Arhus State Hospital, Risskov, Denmark

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