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Trisomy for Short Arm of Chromosome 20

I. Šubrt and V. Brychnáč

Department of Pediatrics, Faculty of Medical Hygiene, Charles University Prague

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Summary. In a patient with a peculiar face and with a severe spondylar dysplasia partial trisomy of the short arm of chromosome 20 was detected. The trisomy resulted from maternal reciprocal translocation t(20p—;21p+). We believe this case to be the first example of a confirmed partial trisomy 20.

Zusammenfassung. Bei einem Patienten mit einem seltsamen Gesicht und schwerer spondylärer Dysplasie wurde eine partielle Trisomie des kurzen Armes vom Chromosom 20 entdeckt. Diese Trisomie ging auf eine reziproke Translokation t(20p—;21p+) bei der Mutter zurück. Es scheint sich um den ersten gesicherten Fall einer partiellen Trisomie 20 zu handeln.

The abnormalities in the F-group chromosomes are rare, and only a few cases of the confirmed chromosome 20 heve been reported. The purpose of this paper is to report a case of a trisomy of the short arm of chromosome 20. This abnormality resulted from a maternal balanced translocation t(20p—;21p+). This partial trisomy could be recovered only by careful examination of the banding patterns of maternal chromosomes. The conventionally stained mitoses revealed an apparently normal karyotype.

Case Report

The patient (JH180966) is the only child of unrelated parents. At the birth of the child the mother was 21 and the father 29 years old. Labor was at term and the birth weight was 4300 g, the birth length 54 cm.

The child was examined in the age of 5 years. The findings showed a timid girl with a very mild mental subnormality. Physical features included prominent bilateral epicanthus, gothic palate, hypoplasia of the maxillar region with a consequent protrusive occlusion. Her hair was coarse. On the X-ray examination there was a severe spondylar dysplasia, a thoracal kyphosis, and lumbal hyperlordosis. The alcalic phosphatases were elevated and the T_3-test suggested thyreoidal hypofunction. Other clinical and biochemical examinations, including the prolin-hydroxyprolin level and acid mucopolysaccharides in urine were normal.

Dermatoglyphics

The mother and the child have additional triradii on the palms. The mother has triradii $(a—b)'$ and d' on the right and $(c—d)'$ on the left palm; the patient a' on the right and $(a—b)'$ on the left palm.

The axial triradii in the proposita are elevated and thus the total $at'd$ angle is 98°. All three members of the family have an increased total ridge count. The patient and her father have an increased number of whorls on fingertips 9 and 8, respectively.

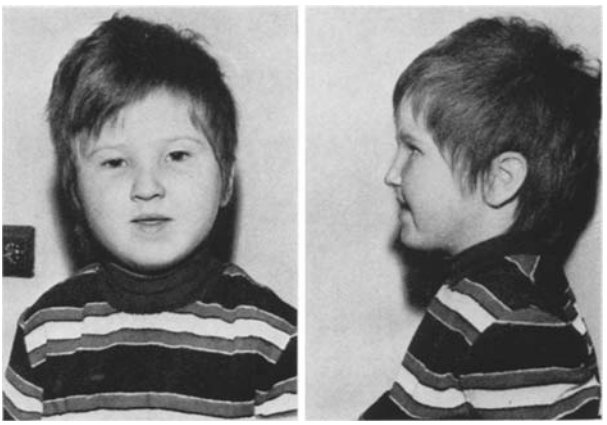

Fig. 1. The patient at the age of 7 years

Table 1

		I	II	III	IV	V	TRC	atd
Proposita	right	16/14	17/13	18/22	19/17	15/0	189	98°
	left	18/13	19/17	18/22	22/17	19/4		
Mother	right	22/20	17/0	14/0	17/11	15/3	167	69°
	left	19/21	17/0	13/0	16/10	15/9		
Father	right	18/22	18/14	15/21	25/24	17/11	194	82°
	left	23/13	16/0	20/21	20/12	15/0		

Cytogenetic Studies

The first examination of the Giemsa-stained slides of the child's blood culture showed 46 chromosomes. In the G group an abnormal chromosome with an enlarged short arm was found. The parental karyotypes were apparently normal, so we were not able to trace the origin of the child's extrachromosomal material.

After the introduction of the banding techniques, we reexamined this case and could only identify the abnormal chromosome to be chromosome 21 with an enlarged short arm. In spite of the apparently normal karyotypes in both parents, we decided to reexamine them too.

The G bands were achieved with the aid of our own modification of the ASG method of Sumner *et al.* (1971) in combination with the trypsin treatment according to Seabright (1971). The slides were incubated for 60 min in 2 × SSC at 60°C and, after a brief rinsing in saline (37°C), were treated for 1 min with 0.25% solution of trypsin. After careful washing in saline (37°C), the slides were stained with 2% solution of Giemsa.

According to the banding patterns, apparently three chromosomes 22 were found in the mother's mitoses. One of the small mediocentric chromosomes was similar to that present in our patient, i.e. chromosome 21 with an enlarged short

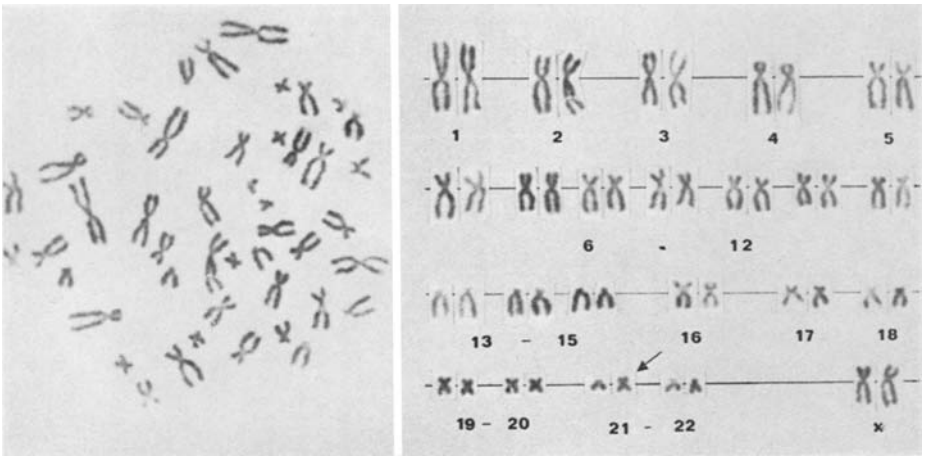

Fig. 2. The karyotype of the patient; arrow points to the abnormal chromosome

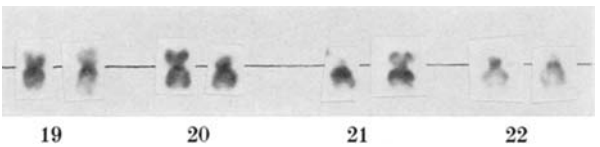

Fig. 3. Partial karyotype of the mother shows the translocation between chromosomes 20 and 21 (G banding)

arm. This portion corresponded to the short arm of chromosome 20. Therefore it was reasonable to suggest that one of the apparent chromosomes 22 in fact represents the long arm of the chromosome 20. These findings permit us to conclude that the mother is a carrier of a balanced translocation t(20p—;21p+) and that the extra-chromosomal material in our patient resulted from an unbalanced condition — the trisomy of the short arm of chromosome 20. The formula for the unbalanced karyotype is to be 46,XX,—21,+t(20p;21q)mat., or 46,XX,—21,t(20;21)(20pter→20p11::21p11→21qter) derived from the maternal karyotype with the formula 46,XX,—20,—21,t(20;21) (20pter→20p11::21p11 →21qter; 21pter→21p11::20p11→20qter).

Discussion

The unbalanced reciprocal translocations always represent the condition of duplication deficiency. However, in our case the deficiency includes the short arm of chromosome 21 which is known to be genetically silent. From this point of view the unbalanced karyotype found in our proposita may be regarded as a "pure" trisomy for the short arm of chromosome 20. Unfortunately there are no other cases for comparing the phenotypes. As has been mentioned above, there are only a few cases dealing with the abnormal chromosome 20. The deficiency

resulted from a ring chromosome 20 was reported in two epileptic patients (Atkins *et al.*, 1972; Faed *et al.*, 1972). The possibility of the trisomic state for the part of a chromosome 20 suggested Allderdice *et al.*, (1971). However, in their case there was a complex translocation involving chromosomes 6, 14, and 20; and the main product of the unbalanced condition is shown to be the partial trisomy 14. Carrel *et al.* (1971) performed an autoradiographic study of a familial translocation 13/F. The partial trisomy F was found in three sibs, but the chromosome of the F group is not specified. To our knowledge, the other cases in the literature deal only with F-group chromosomes, without the identification between both pairs of this group, or only with the F-like chromosomes. We therefore suppose that our case is the first example of a confirmed trisomy of the short arm of the chromosome 20.

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Dr. Ivan Šubrt, CSc
Department of Pediatrics
Faculty Hospital
Vinohradská 159
100 81 Praha 10
Czechoslovakia