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# *DE NOVO* COMPLEX CHROMOSOMAL REARRANGEMENT IN A PHENOTYPICALLY ABNORMAL GIRL[1]

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Summary. Complex chromosomal rearrangements occur rarely in humans. We found them only once among 9000 patients karyotyped with banding in our Department.

The present report describes a malformed girl with *de novo* complex chromosomal rearrangement of autosomes 1, 4, 5, and 20.

## CASE REPORT

A. W., a 4-month-old girl (Fig. 1) was referred to the Genetic Counselling Unit for comprehensive evaluation of her mental retardation and associated dysmorphic features. The girl was born to a healthy, non-consanguineous parents. The pregnancy was full-term and uneventful.

Birth weight was 2350 g. At the girl's birth the father and mother were both 25 years old. There was no history of exposure to know mutagens. Another child of this mother with a different partner was in good health.

When seen at 4 months of age, her height, weight and head circumference were less than the 3rd percentile. Physical examination revealed the following malformation: microcephaly, prominent frontal suture, hypertelorism, slightly mongoloid palpebral fissures, medial strabismus, prominent nose with a broad base, pronounced philtrum, microretrogeny, bilateral clinodactyly of the 5th fingers, bilateral transverse palmar creases, nails hypoplasia, systolic murmur in the heart region made us think about congenital heart disease.

## CYTOGENETIC INVESTIGATION

Chromosome studies were performed on peripheral blood lymphocytes. 40 metaphases were analysed using G-banding. The banding studies showed the kariotype

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46, XX, 1p+, 4q−, 5q−, 20p+, q+. The interpretation of her kariotype as shown in Figure 2 is as follows: 46, XX, t(1; 5) (p 36; q31), t(4; 20) (q 31; q 13), ins(4; 20) (q27 q31; p11). The parents have normal kariotypes.

## DISCUSSION

Complex chromosomal rearrangements occur rarely in humans (Kleczkowska et al., 1982). Up to now about 23 cases of complex chromosomal rearrangements were described (Fryns et al. 1984, Bogart et al. 1986).

According to the way of transmission all complex chromosomal rearrangements may be divided in two subgroups:

1. Complex chromosomal rearrangements with familial occurrence,
2. Complex chromosomal rearrangements arising *de novo.*

Partial trisomies resulting in congenital malformations in the offspring seem to be the most frequent consequences of familial complex chromosomal rearrangements (Palmer et al., 1976, Mattei et al., 1979). Less frequent consequences of complex chromosomal rearrangements seem to be partial monosomies. They occur mainly as the consequences of *de novo* complex chromosomal rearrangements [2]. Congenital malformation may appear even without visible loss of chromosomal material (Pai et al., 1980, Fryns et al., 1984). Possibly they are due to the positional effect in the rearranged chromosomes. In our case complex chromosomal rearrangement arose *de novo* and the applied banding techniques revealed apparently balanced complex chromosomal rearrangement involving four autosomes although some slightly deletions resulting in monosomies could not be excluded.

Mental retardation and phenotypic abnormalities in our patient could be dependent on the position effect or subtle deletions. The use of subbanding techniques (high resolution banding techniques) can be very useful. Unfortunately, our patient died before the high resolution banding was performed.

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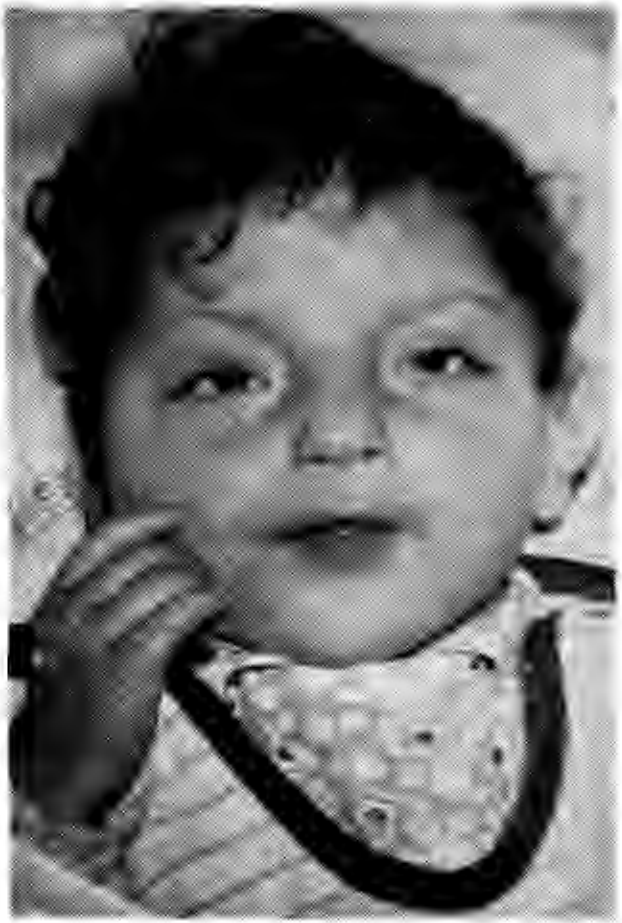

Fig. 1. The face of the patient A. W.

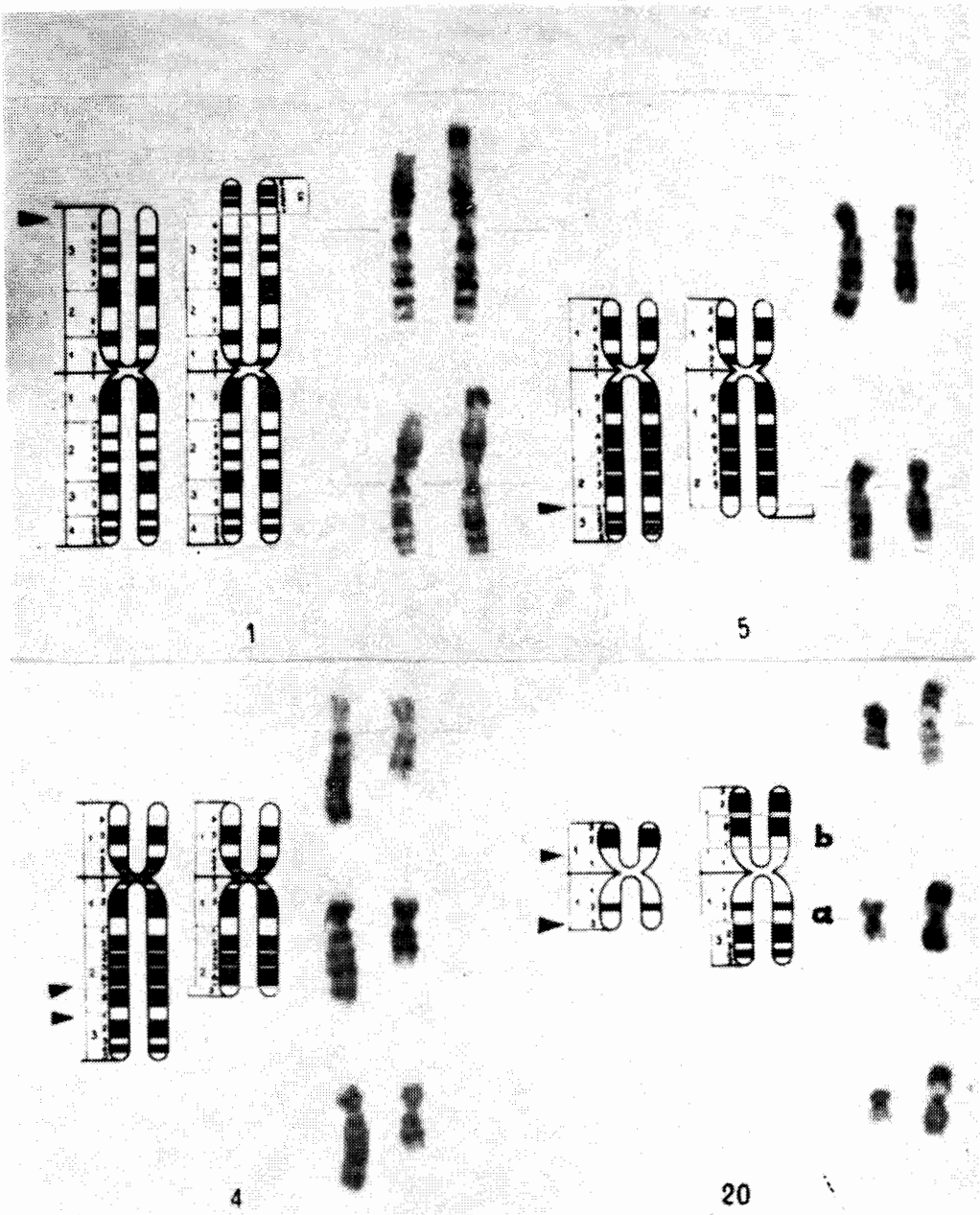


Fig. 2. G-banding diagram with the breakpoints in the patient's karyotype (according to the Paris Conference 1971)

## PRZEGRUPOWANIE CHROMOSOMÓW POWSTAŁE *DE NOVO* U DZIEWCZYNKI Z NIEPRAWIDŁOWYM FENOTYPEM

Streszczenie

Przeprowadzono badania cytogenetyczne czteromiesięcznej dziewczynki, u której zaobserwowano opóźnienie rozwoju psychoruchowego oraz obecność wad wrodzonych (małogłowie dysmorfia twarzoczaszki, brak płytek paznokciowych obu stóp, wada serca). We wszystkich badanych komórkach stwierdzono zmiany powstałe *de novo* w obrębie czterech chromosomów 46, XX, t(1; 5) (p36; q31), t(4; 20) (q31; q13), ins (4; 20) (q27 q31; p11). Na podstawie analizy prążków G wydaje się, że w rozważanym przypadku mamy do czynienia z translokacjami zrównoważonymi. Obserwowane nieprawidłowości fenotypu mogą być zależne od wpływu „efektu pozycji" w przegrupowanych chromosomach bądź od niewielkich delecji, niemożliwych do wykrycia standardowymi technikami cytogenetycznymi.

## ПЕРЕГРУППИРОВКА ХРОМОСОМ, ПОЯВИВШИХСЯ *DE NOVO* У ДЕВОЧКИ С НЕПРАВИЛЬНЫМ ФЕНОТИПОМ

Резюме

Аномалии относительно нескольких хромосом редко наблюдаются у человека. У 4-х-месячной девочки были произведены цитогенетические исследования ввиду отсталого психомоторного развития и наличия врождённых дефектов (микроцефалия, дисморфия лицечерепа, отсутствие ногтевых пластинок на пальцах обеих ног, порок сердца). Цитогенетические исследования во всех исследуемых клетках показали изменения, возникшие *de novo* в пределах четырёх хромосом: 46, XX, t(1; 5) (p36; q31), t (4; 20) (q31; q13), ins (4; 20) (q27 q31; p11).

На основании анализа полосок G кажется, что в данном случае это были уравновешенные транслокации. Наблюдаемые неправильности фенотипа могут зависить от влияния „эффекта позиции" в перегруппированных хромосомах или от небольших делеций, которые нельзя обнаружить с помощью стандартных цитогенетических методов.