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# Coincidence of paternal 13pYq translocation and maternal increased 13p NOR activity in a child with arthrogryposis and other malformations

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Cytogenetic examination of a boy with congenital multiple arthrogryposis, VSD and dysmorphic facies revealed a probable t(Y; 13)(q?; p1) translocation and three NOR-positive dots on one of the chromosomes 13. The latter variant could be followed in the family of the mother, the 13/Y translocation was found in the relatives of the father. Since all the family members affected by one or the other cytogenetic anomaly were healthy, the abnormal phenotype of the propositus was interpreted as coincidence by chance.

Translocations of a Y chromosome to an autosome are rare, and involve most frequently a genetically inactive part of the Y chromosome translocated to chromosomes 15 or 22. Here we report a familial 13pYq translocation combined with a peculiar NOR variant of chromosome 13.

## REPORT OF A CASE

K. G., a male infant was born at term weighing 3550 g after an uneventful pregnancy. At birth club foot, stiff joints, and a systolic murmur were recorded, and congenital multiple arthrogryposis and VSD were diagnosed.

At 8 months of age he was referred to our department because of failure to thrive and increased susceptibility to infections.

At admission he weighed 4220 g, his length was 58 cm, with a head circumference of 40 cm. He gave the impression of severe physical and mental retardation. His main symptoms were, a broad nasal bridge, epicanthic folds, strabism with exophthalmus on the right side and optic coloboma, high arched palate, large ears (Fig. 1).

Both arms and the right lower extremity were stiff with extension contractures of the elbows and the right knee, and with flexion of the hands and the right foot (Fig. 2). The testicles were not palpable. Cardiology revealed a ventricular septal defect. No characteristic bone anomalies were seen at radiologic examination. Blood and urine chemistry proved to be normal.

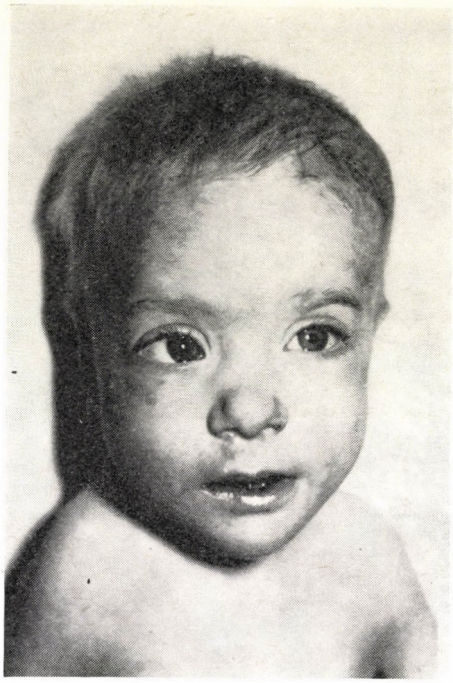

FIG. 1. The face of the propositus

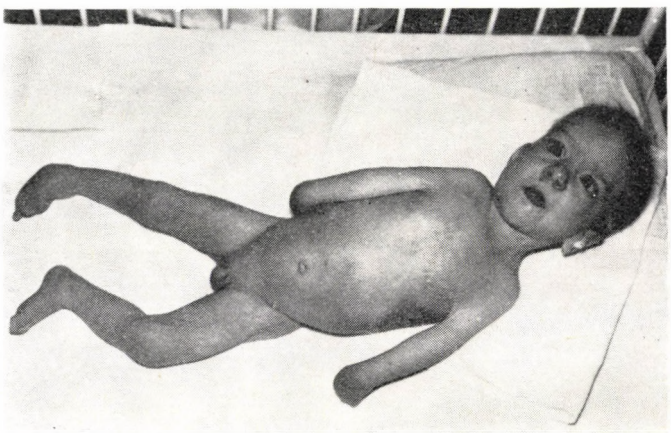

FIG. 2. Arthrogryposis and dystrophy of the patient

*Cytogenetic analysis*

In routine G-banded karyotypes of the proband the chromosome number was consistently 46, but one of the chromosomes 13 had longer short arms in each of the examined mitoses (Fig. 3). With Q- and C-banding the plus material on 13p proved to be positive with the intensity of Yq

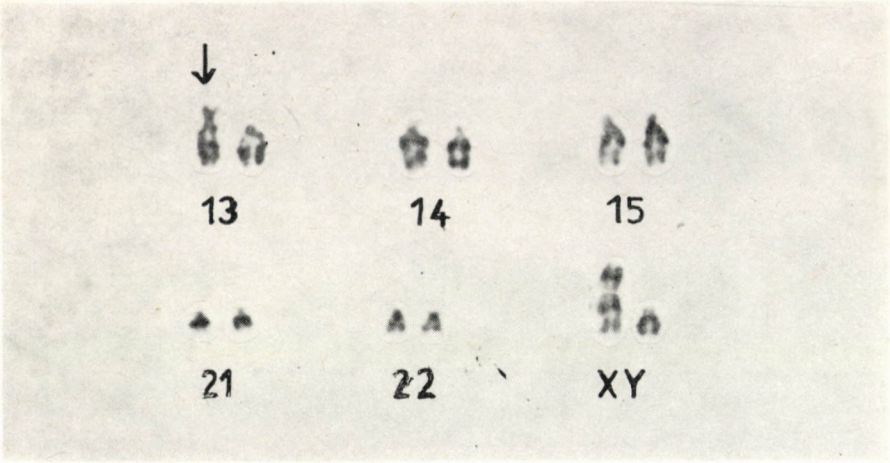


FIG. 3. Partial karyotype of the patient showing and extra material on 13p

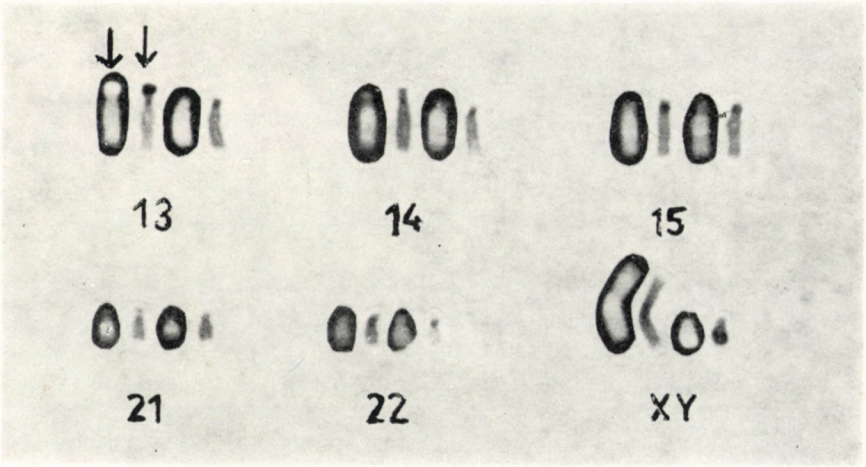


FIG. 4. Sequential Q and C-banding of the same chromosome

(Fig. 4). Since in 23% of interphase lymphocytes two fluorescent Y-bodies were found (Fig. 5), the anomaly was interpreted as t(Y;13) (q?p1) translocation. In NOR preparations a further peculiarity was noted: one of the chromosomes 13 had three darkly stained dots in each of the mitoses.

*Family investigations*

The pedigree is demonstrated in Fig. 6. As shown by the symbols, the lymphocyte cultures of 15 family members in three generations were analysed by means of G, C and Q bandings and NOR techniques. Ex-

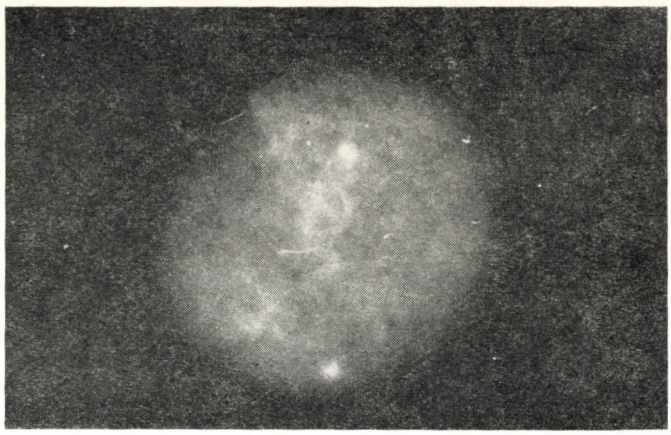

FIG. 5. Two fluorescent Y-bodies in the interphase lymphocytes

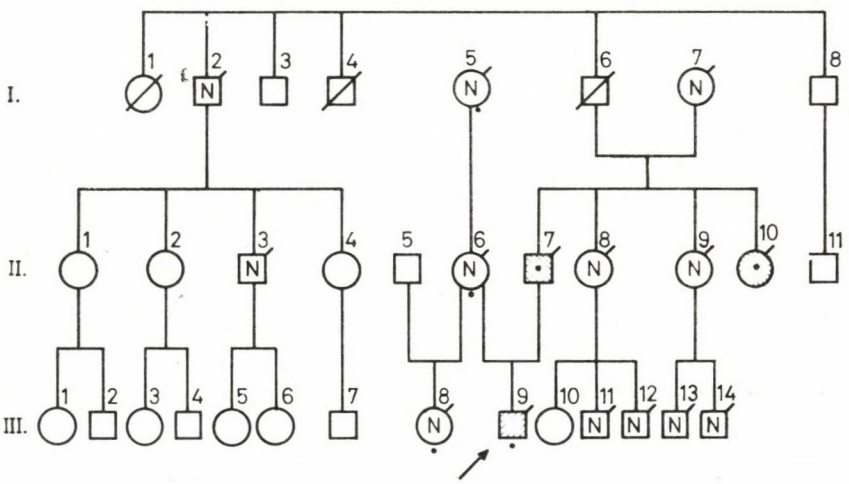


FIG. 6. Pedigree of the family. Symbols: personally examined, cytogenetically tested; ♂ □ subjects with 13pYq translocation; ⊙ family members with three NOR positive dots on 13p; N normal karyotype

cept for the proband all the subjects examined were phenotypically normal. The father (II/7) had the same t(Y;13) (q?p1) translocation as his son.

The father's youngest sister (II/10), a healthy 18-year-old girl, had the abnormal 13p+ with one fluorescent Y-body in her interphase lymphocytes. The other paternal relatives had normal karyotypes.

The mother (II/6), her 5-year-old daughter from her first marriage (III/8) and the maternal grandmother (I/5) had a normal karyotype, but in each of the mitoses an increased number of satellite association was noticed (Fig. 7). In the NOR preparations of

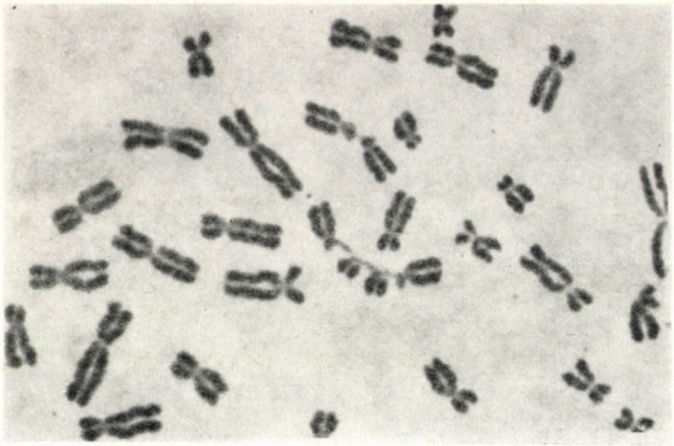

FIG. 7. Increased tendency to satellite-associations in the mother (II/6)

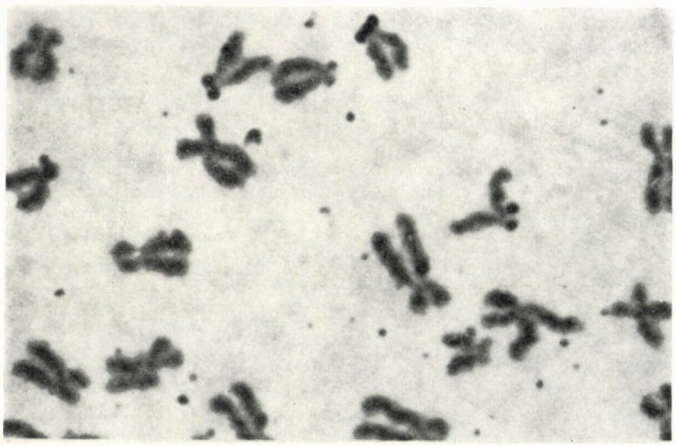

FIG. 8. Three NOR positive dots on one of chromosomes 13

these subjects the conspicuous three dots on one of the chromosomes 13 was observed (Fig. 8).

## DISCUSSION

Investigation of the family could have been more reliable with the application of distamycin A-DAPI banding for distinction between Yq and possible extra large satellites [3], with determination of H-Y antigen for estimation of genetic activity of the presumed extra Y material, and with in situ r-RNA-DNA hibridization in order to clarify the nature of the peculiar three NOR dots on chromosome 13. Since, however, in the majority of the presumptive Y/autosome translocations called in question retrospectively [1, 4] the

autosomes involved were Nos 15 and 22, it was unequivocally No. 13 in our family. Since the carrier father and aunt were somatically and psychologically normal, we believe that they had a duplication of a genetically inactive part of the Y chromosome translocated to chromosome 13. This would mean that the symptoms of the proband had nothing to do with the 13/Y translocation.

We have no explanation for the familial NOR variant on the maternal side but, considering the observations of Schwarzacher et al [2], one may reckon with an unusually increased NOR-activity in this region. The phenomenon may be interpreted as a probably rare population variant, which has certainly no phenotypic consequences. However, the coincidence of two presumably harmless inherited anomalies of chromosome 13 in a severely malformed child seems at least curious.

## References

1. Schinzel A: Catalogue of Unbalanced Chromosome Aberrations in Man. de Gruyter, Berlin 1984, pp 806—807
2. Schwarzacher HG, Mikelsaar AV, Schnedl W: The nature of the Ag-staining of nucleolus organizer regions. Cytogenet Cell Genet 20:24—39, 1978
3. Schweizer D, Ambros P, Anderle M: Modification of DAPI banding oligopeptide anti-biotic: distamicin A. Exp Cell Res 111:327—332, 1978
4. Spowart G: Reassessment of presumed Y/22 and Y/15 translocations in man using a new technique. Cytogenet Cell Genet 23:90—94, 1979

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