Partial trisomy 13q resulting from a paternal reciprocal Yq;13q translocation

J Nikoliš, K Ivanović, V Diklić

We report on a newborn male with congenital anomalies consistent with Patau's syndrome resulting from a partial trisomy 13q inherited from the father with a reciprocal t(Y;13)(q12;q14). This adds to the evidence for the possibility of differences in behaviour during meiosis of the Y;autosome reciprocal translocation.

The proband, a male infant, was born in 1989. Pregnancy and labour were normal. His mother was 28 years old at the time of his birth and his father was 34. Birth weight was 4620 g, length 55 cm, and head and thoracic circumferences were both 36 cm. Physical examination showed a picture typical of Patau's syndrome, including frontal bossing, narrow temporal area, beaked nose, low set ears with prominent antihelix, hypotelorism, microphthalmia and enophthalmia, high arched palate, and micrognathia. There were haemangiomas in the occipital and neck regions, bilateral hexadactyly of the hands, hyperextensible thumbs, partial fusion of the pelvis and scrotum, bilateral inguinal hernia, and heart defect. Re-examination at 3 months showed the infant severely retarded in his development (weight 4400 g, length 60 cm, head circumference 38 cm). Microcephaly and muscular hypotonia were noted. At the time of this report the child was 10 months old.

The mother reported that two sperm analyses done at an interval of eight months in 1984 and in two different clinical laboratories had shown azoospermia in her husband, the proband's father. Artificial insemination with donor sperm in 1986 had resulted in the birth of a healthy and chromosomally normal boy. In 1987, the mother, to everybody's surprise, became pregnant by her husband, but abortion was induced in the first trimester of the pregnancy

because of radiation received for diagnostic purposes. In 1989 she became pregnant again by her husband and our proband was born.

Analysis of 20 Ag-NOR-G banded metaphases from a lymphocyte culture of the proband's peripheral blood showed 46 chromosomes with a Y chromosome very similar in size and band pattern to chromosome 13. Karyotypic examination of the parents showed the mother to be normal, but the father, in 30 Ag-NOR-G banded metaphases, to be a balanced carrier of a Yq;13q translocation. C banding showed the breakpoint in the father's Y chromosome to be within the heterochromatic segment (q12) and a small portion to have been translocated to 13q14, indicating a reciprocal

Partial karyotypes showing chromosomes 13 and Y using Ag-NOR-G banding and C banding (proband, top two rows, father, bottom two rows).

Institute of Biology and Human Genetics, Belgrade University Medical School, Visegradska 26, 11000 Belgrade, Yugoslavia.

J Nikoliš, V Diklić

Department of Pediatrics, General Hospital, Pancevo, Yugoslavia.
K Ivanović

Correspondence to Dr Nikoliš.

Received for publication 3 July 1990.

Revised version accepted for publication 4 October 1990.

t(Y;13)(q12;q14) in the father and confirming a 46,X,der(Y),t(Yq;13q)pat karyotype in the proband (figure). The father has only one brother and his karyotype is normal, indicating a de novo translocation in the father.

Partial trisomy for distal 13q, because of the additional 13q material on the Yq, occurred together with the clinical characteristics of Patau's syndrome in our proband. It appears, therefore, that partial trisomy for a distal three-quarters, approximately, of 13q (q14→qter) has the same consequences as complete trisomy of chromosome 13. The finding of the Y;13 translocation also in the father was unexpected. The father, however, has refused to come in for additional study, thereby precluding further sperm and meiotic studies.

Y;autosome translocations are rare occurrences. They most frequently involve an exchange of heterochromatin between the long arm of the Y chromosome and the short arm of an acrocentric and do not affect fertility.¹ A reciprocal translocation between the long arms of the Y and acrocentric chromosome has been reported by Petit *et al.*² Their t(Y;14)(q12.2;q11.1) patient was azoospermic, as is commonly the case with males with a de novo Y;non-acrocentric translocation.³ There is one published report of a patient with t(Y;16)(q11;q13) and severe oligospermia⁴ and even one of a patient with t(Y;10)(q12;q24)⁵ with a good sperm count. The latter came to be studied

because of the birth of a child with congenital malformations. Our case, then, is the second case report of a Y;autosome reciprocal translocation ascertained through the birth of a malformed child, and the first of a Yq;13q reciprocal translocation.

The reports cited above show that characteristic secondary spermatocyte or spermatid arrest⁵ can be less than complete⁴ or even absent.⁵ In the case of the father of our proband, since there is no record of any illness at the time of either spermatogram, we must assume that he was actually severely oligospermic and not azoospermic, despite the clinical laboratory reports. Otherwise it would be very difficult to explain the inconsistency in the behaviour during meiosis of the Y;autosome translocation.

- 1 Smith A, Fraser IS, Eliot G. An infertile male with balanced Y;19 translocation. Review of Y;autosome translocations. *Ann Genet (Paris)* 1979;22:189-94.
- 2 Petit P, Unglik A, Fryns JP. Translocation 46,X,t(Y;14)(q122;q111) in a case of sterility in the male. *Ann Genet (Paris)* 1982;25:63-4.
- 3 Laurent C, Chandley AC, Dutrillaux B, Speed RM. The use of surface spreading in the pachytene analysis of a human t(Y;17) reciprocal translocation. *Cytogenet Cell Genet* 1982;33:312-8.
- 4 Faed MJW, Lamont MA, Baxby K. Cytogenetic and histological studies of testicular biopsies from subfertile men with chromosome anomaly. *J Med Genet* 1982;19:49-56.
- 5 Laurie DA, Palmer RW, Hultén MA. Studies on chiasma frequency and distribution in two fertile men carrying reciprocal translocations; one with a t(9;10) karyotype and one with a t(Y;10) karyotype. *Hum Genet* 1984;68:235-47.