Translocation t(1;20)(q21;q13) in an azoospermic man

Translocations involving chromosomes 1 and 20 are uncommon. Seabright (1981, personal communication) has reported a woman carrier of a t(1;20) (q32;q13) and Norman and Boucher[1] have described a woman with chronic myelogenous leukaemia whose karyotype was 46,XX,t(1;20)(q21;q13) in bone marrow cells and 46,XX in lymphocytes. In the present report we describe a balanced translocation t(1;20) (q21;q13) detected in the lymphocytes of a phenotypically normal man presenting with azoospermia.

The analysis of the proband's metaphases, stained by the Q banding technique, shows the presence of a reciprocal translocation between the long arms of chromosomes 1 and 20 in all the 50 cells examined (figure). Differential staining with the C banding technique shows that the breakpoint on chromosome 1 is below the secondary constriction (figure).

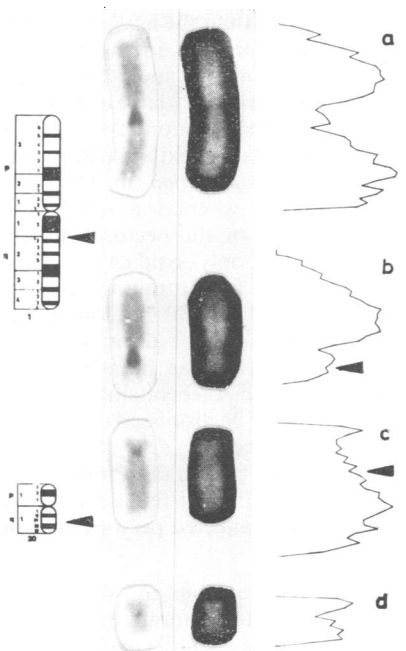

FIGURE *Chromosomes involved in the translocation (stained with C and Q banding) and their densitometric profiles: (a) structurally normal chromosome 1; (b) chromosome 1pter→1q21::20q13→20qter; (c) chromosome 20pter→20q13::1q21→1qter; (d) structurally normal chromosome 20. Arrows indicate the breakpoints.*

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The mean densitometric profiles[2] of the two translocated chromosomes compared with those of the structurally normal chromosomes 1 and 20 show that the band sequences of the two rearranged chromosomes are, respectively, 1pter→1q21::20q13→20qter and 20pter→20q13::1q21→1qter. The breakpoint on chromosome 1 is at the distal end of the q21 band, while the breakpoint on chromosome 20 is localised distally on the q13 band. Therefore, the proband's karyotype is: 46,XY,t(1;20) (q21;q13).

It was impossible to obtain blood samples of the proband's parents and sister, but his only brother had a normal karyotype.

Reports of male sterility associated with autosomal translocations are rare,[3] and meiotic studies in the carriers of this type of rearrangement are even rarer. A positive correlation, however, seems to exist between spermatogenic failure and chain configurations of the quadrivalents at meiosis, when one breakpoint is proximal and the other more distal to the centromere.

The translocation reported here, with breakpoints at 1q21 and 20q13, could give chain configurations at meiosis.

Unfortunately it was impossible to obtain a testicular biopsy from the patient. He is the only male balanced carrier of t(1;20) (q21;q13) detected to date. Therefore we can only speculate about the possibility that azoospermia is directly attributable to the translocation detected in the patient's lymphocytes.

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