Two whole arm reciprocal translocations

Whole arm reciprocal translocations (rcpt) are rare. We report here two such cases.

Case 1 was a 24 year old woman who had had seven consecutive early pregnancy losses. Cytogenetic studies of peripheral blood with GTG banding showed a large reciprocal translocation between the long arms of chromosomes 1 and 5 with breaks in both chromosomes at their centromeres (figure a). C banding showed that the break in chromosome 1 had occurred on the p side of the centromere. The break in the chromosome 5 was within the centromere. The translocation was de novo. The husband had normal chromosomes. The next two pregnancies, monitored by amniocentesis, produced a normal male in the first and a female balanced carrier in the second.

Case 2 was a 35 year old woman who had had four early spontaneous abortions and no living children. Karyotype of her husband was normal. She had a reciprocal translocation between the long arm of chromosomes 10 and 14 with the breakpoints within the centromere of both chromosomes (figure b). At the age of 36 she had another pregnancy; amniocentesis at the 16th week showed the fetal karyotype to be 46,XX,-14,+der14,t(10;14) (10p14q;14p10q)mat. This pregnancy subsequently aborted spontaneously. The products of conception were not available for study. The proband’s father, brother, and two maternal aunts were normal but her mother was dead, so the inheritance of this translocation remains unknown.

In both of these cases the exchanges of the long arms which were virtually identical in length would have remained undetected without banding.

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