USE OF BANDING TECHNIQUES FOR ZYGOSITY DIAGNOSIS IN TWINS

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Detailed comparison of human variant chromosomes found by Q- and C-banding technique is performed in a pair of twins to show that these variants can be used for zygosity diagnosis.

Q- and C-banding techniques in human cytogenetics have revealed a polymorphism of chromosomes which is very specific for each individual. Already five years ago it was suggested that, except for an identical twin, each person's karyotype might prove to differ from everyone else's, perhaps being the cytological equivalent of the fingerprint (Hecht 1971). In a recent study of a human newborn population no two individuals with identical Q- or C-band patterns were found (Müller et al. 1975).

During the last three years we have used these chromosomal variants to determine the parental origin of chromosomes, e.g., in trisomic children and in prenatal diagnosis of female fetuses to exclude contamination with maternal cells.

Detailed comparison of chromosomal variants has also turned out to be a valuable addition to existing techniques for the determination of twin zygosity (Van Dyke et al. 1975).

We want to demonstrate the technical procedure for diagnosis of twin zygosity in a single pair of newborn male twins who were referred to chromosome analysis from the Department of Pediatrics because twin B was suspected for Down's syndrome.

Chromosome examination in the twins was performed on preparations from 72 hours lymphocyte cultures. The preparations were stained with Quinacrine Mustard and 15 metaphases from each twin were photographed in a Zeiss fluorescence microscope. The films were analyzed by projection at a magnification of about × 6000.

The same preparations were later destained in 95% ethyl alcohol and restained for C-banding (Holbek et al. 1974).

The same metaphases which had been photographed in the QM stained preparations were rephotographed in the C-band-stained preparation. Analysis of the C-bands was done by projecting the Q- and C-band films simultaneously beside each other. Q-band variants from the twins were compared in projection and C-band variants were compared in 5 prints made from each twin.

Chromosome examination in the parents was performed only in QM stained preparations from lymphocyte cultures.

Classification of Q-band variants was done following the suggestion of the Paris Conference (1971).

Classification of C-band variants was done on morphological criteria, especially the location of centromeric hetorochromatin either in the middle of the centromere, towards the short arm, or towards the long arm of the chromosome. But also differences in length of centromeric heterochromatin were taken into account when comparing homologous chromosomes of the twins and their parents.

In twin A we found a normal male karyotype 46,XY and in twin B we found the karyotype 47,XY,+21. Identical patterns in the Q-band variants of chromosomes 3, 4, 13, 14, 15, 21, 22, and Y, were found in both twins (Fig. 1), and it had to be investigated if it was a pair of MZ twins where nondisjunction in twin B had occurred in one of the early mitotic divisions.

In that case one should most probably expect to find a mosaic of type 46,XY/47,XY,+21 in twin B, but screening of 100 metaphases from twin B in the microscope showed no cells with the normal karyotype 46,XY. A

Fig. 3. Chromosomes No. 21 in the father, mother, twin A, and twin B.

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Table. *Phenotypes of the probands and their parents*

Marker system	Twin A	Twin B	Father	Mother
HL-A	2, 8, W10, W19	2, 2, 12, W10	2, 8, 12, W19	2, 9, W10, W15
AB0	0	0	B	B
Rhesus	CDe	CDe	cde	CDe
MNS	MNSs	NSs	NSs	MNSs
Le	Le(a—b+)	Le(a—b+)	Le(a—b+)	Le(a—b—)
Fy	Fy(a+b+)	Fy(a+b+)	Fy(a+b+)	Fy(a+b+)
Lu	Lu(a—)	Lu(a—)	Lu(a—)	Lu(a—)
K	K—	K—	K—	K—
Jk	Jk(a+b+)	Jk(a+b+)	Jk(a—b+)	Jk(a+b—)
P	P_2	P_2	P_2	P_1

comparison of the C-bands in both twins was therefore performed and showed a clear difference in the location and the length of C-heterochromatin within pairs Nos. 1, 2, 3, 9, and 18 (Fig. 2). Examination of blood types and tissue types in both twins showed that also in these genetic marker systems there was a difference in HL-A types and in MNS blood types (Table).

Comparison of chromosomes No. 21 in the parents and the twins showed a centromere marker in one chromosome No. 21 in the mother, and this marker was found in two of the No. 21 chromosomes in twin B. Nondisjunction had thus occurred in the second meiotic division of the mother (Fig. 3).

In our case it was possible to show a difference of chromosomal variant pattern by C-banding, but not by Q-banding, but both marker systems are valuable in twins to show if they are MZ or DZ. If a difference is found in one of these systems or in both, it can be regarded as an indication of dizygosity.

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