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Giemsa Banding Patterns of 13/14, 21/21 Translocations, Philadelphia Chromosome And Trisomy 18*

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A special giemsa banding technique has offered the possibility of exact identification of individual chromosomes in the human karyotype.[1-4] Using this technique we have identified individual chromosomes involved in cases of Dq/Dq translocation carrier, Gq/Gq translocation mongol, chronic myeloid leukemia and trisomy 18.

Methods

Leukocytes were cultured 72 hours in McCoy's 5A medium with 15 percent fetal calf serum and harvested using potassium chloride (0.075 M) as an hypotonic solution, fixed in methanol, acetic acid mixture (3: 1), spread and air dried.[5] Slides were incubated 60-75 minutes at 60°C in 2 X SSC solution (0.3 M sodium chloride and 0.03 M sodium citrate), rinsed briefly in distilled water and air dried.[1] Only a portion of the metaphases revealed the characteristic banding pattern clearly enough for accurate identification.

Results

Case No. 1. (Dq/Dq Translocation Carrier): Her modal chromosome number was 45. There were only four D-group chromosomes: one

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with the characteristic banding pattern of no. 13, one with the banding pattern of no. 14 and two with band patterns of no. 15. In addition, an extra metacentric chromosome in the size-range of no. 3 was found. This chromosome had three bands on one arm which fit the characteristic banding pattern of no. 13. On the other arm of this metacentric translocation chromosome there were two dark bands: one was located distally and the other near the centromere. This latter pattern is homologous to that of no. 14 (Figure 1A).

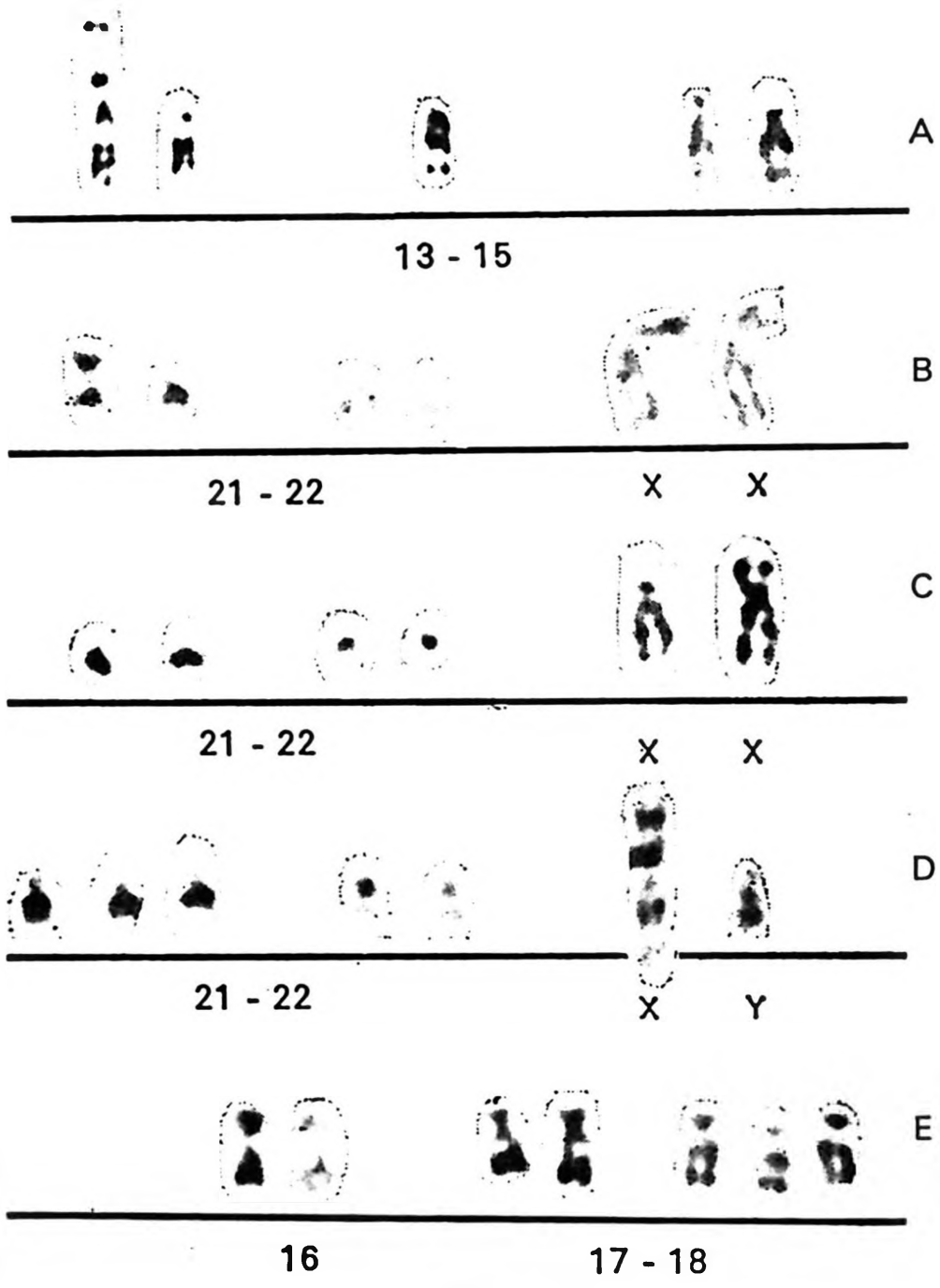

Figure 1

Partial karyotypes showing giemsa banding patterns of 13/14 translocation (A); 21/21 translocation (B); Philadelphia chromosome (C); extra chromosome in Down's syndrome (D); and in trisomy 18 (E);

Case No. 2 (Gq/Gq Translocation mongol): Her modal chromosome number was 46, but there were only three G-group chromosomes: one had a darkly stained long arm and was thus categorized as no. 21; the other two were categorized as no. 22 since they had no bands on their long arms. In addition, there was a small metacentric chromosome in the size range of the F-group chromosomes with dark bands on each arm. This was interpreted as a translocation chromosome between two no. 21's (Figure 1B).

Case No. 3 (Chronic Myeloid Leukemia): The modal chromosome number was 46. In the G group two of the chromosomes were stained densely on the long arm (no. 21) and the other two chromosomes showed darkly stained centromeres (no. 22); only one of these latter two had discernible long arms. It was clear the the Ph^1, which is a G-group chromosome with a deletion of the long arm, had a densely stained centromeric area and thus was categorized as no. 22 (Figure 1C). In the same figure, a partial karyotype of a regular Down's syndrome is shown for comparison (Figure. 1D).

Case No. 4 (Trisomy 18 Syndrome): The modal chromosome number was 47 and the extra chromosome was found to be no. 18, having two equally dark bands on the long arm; one proximally and the other distally (Figure 1E).

Discussion

Our results have provided further examples of the efficacy of the giemsa banding method. Especially in structural chromosome abnormalities, the need for more accurate identification than that obtained by conventional methods was obvious. The giemsa banding technique is simpler and less expensive when compared to autoradiography and quinacrine fluorescence and apparently serves as an accurate tool in chromosome identification.

This method has been used by Seabright to demonstrate a translocation between chromosomes no. 1 and 18, and by Abo, et al., to show a translocation between chromosomes no. 3 and 8.[6,7] To the best of our knowledge our cases are the first (13/14 and 21/21) translocations identified by this technique. However in the latter case, the isochromosome of the long arm of chromosome no. 21 could not be ruled out, theoretically.

Using quinacrine fluorescence, O'Riordan, et al., were able to show that the Ph^1 is one of the G group chromosomes not involved in Down's syndrome[8]. We confirmed their important and unexpected findings by means of the giemsa banding technique.

As was expected earlier, the value of this new method has been shown in identifying various other structural chromosomal rearrangements. Recently, Taysi et. al., using giemsa and quinacrine banding techniques were able to identify clearly the duplication-deficiency product of a pericentrically inverted chromosome no. 13 in a case of trisomy D.[9] Similarly, the giemsa banding method has been shown to be valuable in the precise identification of numerical chromosome aberrations as well. It has been used by Ridler and by Chernay, et al. in the identification of the chromosome involved in Down's syndrome;[10,11] by Halicioglu, et al., in trisomy 13 and by Caspersson et al. intrisomy 8. [12,13] In our study, we were able to identify the extra chromosome involved in trisomy 18.

The basis of the specific giemsa banding patterns in metaphase chromosomes has not been elucidated. DNA denaturation followed by renaturation at the elevated temperature has been thought to be responsible for the banding pattern. It has been suggested that repetitive sequences of DNA (constitutive heterochromatin) may account for the dark bands, since these would be expected to reanneal more rapidly (and thus stain more readily) than nonrepetitive sequences.[14]

Summary

With the aid of the giemsa banding technique, exact identification of four different chromosome abnormalities has been achieved. In the case of a Dq/Dq translocation carrier, the chromosomes involved were shown to be numbers 13 and 14 [45, XX, D-, D-t(13q 14q)+]. Using the same technique we identified the abnormal chromosomes in a case of translocation Down syndrome [46, XX, t (21q 21q)+-. We were also able to confirm the finding that the Philadelphia chromosome (Ph^1) is the one not involved in Down's syndrome and to show the banding pattern of the extra chromosome involved in trisomy 18 (47, XX, 18+).

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