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COMPARATIVE STUDIES OF THE CHROMOSOMAL ARRANGEMENT IN THE C-METAPHASE BETWEEN NORMAL KARYOTYPE AND TRISOMY-21

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Abstract:

Human chromosomes in amnion cells and lymphocytes with normal karyotype and in lymphocytes with pathological karyotype (2n=47, +21) were compared as to their position in the metaphase. None of the collectives showed sex differences. Measurement of the radial distances revealed more peripheral position of the majority of large chromosomes. The satellite-carrying chromosomes of the D group always had a central position in the mitosis. The chromosomes of the groups D, E, F and G were closest to the centre; with the exception of chromosome 18 which was peripheral in all three collectives. For the male probands, the y-chromosome was shown in all three collectives to have a smaller radial distance than the x-chromosome. A typical distribution was found for the radial and homologue distances for the trisomic cells, two of them had a very large radial distance, the third a value corresponding to its size. For the homolarger measurements hereby the distribution is quite independent of parental source. Comparison of the groups showed no differences either between normal and trisomy cells or between the different cell types. Examination of chromosomes 6 and 15 proved conclusively that the chromosomes are not particularly orientated in the c-metaphase regarding the position of short and long arm. A preferential combination of particular satellite carrying chromosomes leads to the frequent fusions of chromosomes 13 and 14, or 14 and 21. Equally, no preferential association could be demonstrated of the chromosome 21 and the chromosomes with large heterochromatin blocks in the centromere region (chromosomes 1 and 9). The distances were of the same order of magnitude as those between 21 and chromosome 6, a submetacentric chromosome without a marked heterochromatin region. Both latter observations are of specific importance for genetic councelling of couples after birth of a child with a de Novo chromosome aberration asking for the recurrence risk.

Keywords:

"Chromosomal arrangements ، C-metaphase Radial & Homologue distances (+21) ، Satellite-association "

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