Acrocentric Associations in Mongol Populations*

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Summary. Association patterns of acrocentric chromosomes in 2100 metaphases from sample populations of 30 chromosomally normal individuals, 20 mongols and 20 parents of mongol children have been studied. In this study the data are concerned with the overall numbers of acrocentric chromosomes present in associations and not with the numbers of associations or with the ways in which they may be arranged. The individual pairs of acrocentric chromosomes were identified by Leishman staining following trypsin digestion.

Acrocentric chromosomes showed a random participation in associations both in normal and in mongol populations, where the additional 21 chromosome significantly alters expected frequencies. In the parents of mongol populations, however, significant deviations from the model of random participation in associations were documented.


Introduction

The abundant literature on acrocentric associations gives contradictory evidence of variation due to differences between individuals, culture techniques and slide preparation; and of correlation with age, sex and chromosome abnormality. The interested worker might well conclude that association patterns are not a reliable measure of chromosome behaviour.

Acrocentric chromosomes, however, are found in trisomic condition in two clinically definable syndromes (Down’s and Patau’s syndrome) and are involved in a variety of cytogenetic abnormalities including translocations (Hecht et al., 1968), deletions (Kolch et al., 1971) and rearrangements (Hauksdottir et al., 1972). The implication of these abnormalities is that acrocentrics are specially vulnerable to non-disjunction and DNA exchange and rearrangement.

The location of nucleolar organising regions on the acrocentrics (Ferguson-Smith, 1964) intimately associates these chromosomes with the events of the

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nucleolar cycle. Consequently any events which affect the nucleolar cycle will have related effects on chromosome behaviour. The nucleolar events which may affect association patterns are represented in Fig. 1. The persistance of the nucleolus through prophase into anaphase seems to be responsible for anaphase lag and the formation of chromosome bridges (Heneen and Nichols, 1966). These events may lead directly to non-disjunction but can not be studied using colcemid-arrested metaphases. Secondly, fusion of a number of nucleoli to form the nucleolus of interphase (Gonzalez and Nardone, 1968; Gani, 1973) is generally seen as the mechanism which brings the acrocentrics into association. At this time an opportunity is provided for the exchange of DNA (Ohno et al., 1961). It is the results of nucleolar fusion which are scored in association work and quite clearly association patterns are simply relics of the \textit{in vivo} situation.

\textbf{Methods and Materials}

Unstained chromosome preparations were made from standard short term blood culture using both the separated leucocyte technique and whole blood micro-technique. The prepared slides were left for 3 weeks, subsequently the material was differentially banded using a modification of Seabright’s (1971) method for producing bands in human chromosomes. This method has been fully described in a previous paper (Curtis, 1972). Banded metaphase chromosomes were analysed visually with reference to the published karyotypes for Leishman stained material (Seabright, 1971) and for Giemsa stained material (Sumner et al., 1971). The two