

A THEORY OF THE ASSOCIATION OF CHROMOSOMES IN KARYOTYPES, ILLUSTRATED BY DR. PATRICIA JACOBS' DATA

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1. Introduction

Geneticists are interested in studying karyotypes. A karyotype is a micro-photograph of a human cell just prior to mitotic division, taken under certain standard conditions. They are able to distinguish between the main groups of chromosomes in the Denver classification [2], and in particular they are interested in a phenomenon called "association" (sometimes it is called "overlap") when several chromosomes approach particularly close together. Figure 1 shows two instances of association. These associations are concerned with members of groups 13-15 and groups 21-22 of the Denver scale. All the data used in this paper refer to these two groups. In what follows, groups 13-15 are called group D and groups 21-22 are called group A. Dr. Patricia Jacobs examined for association a number of karyotypes (each for a different person) from (i) normal individuals (ii) mongols and (iii) abnormals excluding mongols. Nearly all the members of group (iii) had Klinefelter's syndrome, but three were mosaics and one was a case of Turner's syndrome. These people are abnormal in sexual development, Klinefelter's are all of male types; cases of Turner's syndrome are of female type; one mosaic was of male type and two of female types. Groups (i) and (ii) both have four A's and six D's; but the mongols have five A's and six D's that is, an extra chromosome. This is supposed to be the cause of their characteristic abnormalities.

2. Basic model for associations of chromosomes in karyotypes

Now let us consider, say, five chromosomes in the field of observation. They may all five be separate; two may be associated and three separate; two may be associated and the other three associated, and so on. Clearly, each possible case corresponds to a partition of the number 5. The question examined here is whether the associations may be accounted for by the hypothesis that the

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