
Two monographs summarizing independent Swedish data on the cytogenetic and clinical aspects of Down’s syndrome, have appeared recently. Both were published to conform with Swedish medical regulations and should be looked at as such. The first, “Down’s Syndrome: A clinical and cytogenetical investigation” by Karl-Henrik Gustavson, summarizes the results of investigations on 119 individuals with Down’s syndrome, or suspected Down’s syndrome. The basis for the clinical diagnosis of Down’s syndrome in this series, was taken to be mental retardation, together with four or more of Øster’s ten cardinal signs. The results showed 94 individuals to be trisomic-21 with 47 chromosomes; six to be chromosome mosaics; four to have G/G translocations and one a D/G translocation. Four cases had other chromosome anomalies, the most interesting of these being a patient with a presumptive 4-5/21 translocation inherited from the mother. Sixteen children in this series were found to have normal chromosome complements, none had Down’s syndrome according to the clinical criteria used by the author.

The second monograph, “Mongolism in Newborns: A clinical and cytogenetic study” by Bertil Hall, deals with the clinical and cytogenetic features of mongolism during the newborn period. The author has collected 63 cases of mongolism born in south Sweden, and diagnosed during the newborn period. These were all studied during the year July 1st, 1961, to June 30th, 1962, and represent a total population of 25,038. As a control series, 86 children were studied. These were born either in Lund or in one of the nine obstetric hospitals, from which the mongol population was drawn. All these children (mongols and controls) were examined soon after birth, and again if surviving at one year. The aims of this work are listed as a study of: 1. the different physical signs in mongoloid newborns; 2. the variability of physical signs in mongoloid newborns; 3. the possibilities of a clinical diagnosis of mongolism in newborns. The cytogenetic study adds little to our existing knowledge. 63 cases diagnosed as mongols were studied; 83 were trisomic for a 21-22 chromosome and one had an inherited D/G translocation. Five of these babies had normal chromosomes. All 86 control children had normal chromosomes.

A table summarizing the major aetiological and cytogenetic data would have been valuable to those wishing to abstract data rapidly. At present a careful check has to be made through several sections to obtain even the simplest summary of the more important data. It is a pity that the author finds it necessary to invoke hypotheses such as

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“activation” or “inactivation” of the trisomic chromosome to account for the variation of the mongol phenotype, as this is unnecessary. Even worse, is the use of the term “Lyonisation” to describe hypothetical genetic inactivation of an autosome. This unfortunate term was coined to describe the phenomenon of allocyly and random genetic inactivation of one of the two X chromosomes of normal human or mammalian females. If this term must be used, it should only be used in this sense. Its use to describe an autosomal phenomenon is totally incorrect.

The author argues from the finding of five “pseudomongoloids”, as he chooses to call them, that the clinical syndrome can occur without the chromosome aberration. It is a great pity that the clinical features of these five children were not described in more detail and summarised in tabular form, for comparison with the clinical features of the trisomics for, as has been previously stated (Hamerton and Polani, 1962) the establishment of an unequivocal clinical diagnosis of mongolism in a subject with normal chromosomes, would be of great interest and importance. This has not yet been done and on the author’s own admission, while these five children resemble mongols, they do not fit the syndrome; and in fact, only three out of these five children had even four of the ten cardinal signs of mongolism; there is therefore no justification for coining the term “pseudomongoloids”, implying, as this is said to, an individual with the clinical features, without the chromosome aberrations.

It seems a pity that the alternative name of “Down’s syndrome” has been rejected. This is rapidly replacing mongolism, which has unpleasant and misleading racial connotations.

In general, the standard of production of both monographs is high, and the photographic reproductions are good. Both are useful in that they collect together a body of data on Down’s syndrome in a readily accessible manner. Many of the conclusions drawn by Hall should, however, be carefully considered before acceptance. The price of both monographs is high considering their scope and context. This, however, is perhaps the
fault of the system which requires all M. D. and Ph. D. theses to be published, partly at the expense of the candidate, irrespective of their merits on the open market.

Reference


/ L. Hamerton, London