A Comparative Study of Cognitive Traits in Human Sex Chromosome Aneuploids and Sterile and Fertile Euploids

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Three groups of male and female subjects (aneuploid, sterile, and fertile) were administered tests of field dependence (RFT, EFT, BDT, ABCT), spatial ability (S1, VZ1, IBT), right–left discrimination, and verbal ability. Statistical analyses indicated that (1) aneuploid males, with karyotype 47,XXY, do not disclose significant differences in field dependence or spatial abilities from karyotypically normal males or females, (2) aneuploid Turner and Turner-like females do exhibit a significantly higher field dependence and lower spatial orientation and visualization ability than other females, and (3) psychological distress caused by sterility does not seem to influence the cognitive pattern. The importance of socializing environment and hormonal factors in determining the observed differences is briefly discussed in the light of these results, and it is concluded that sex chromosome heterochromatin must play a role in the development of specific cognitive traits.

KEY WORDS: spatial visualization; field dependence; genetics of cognitive traits; sex chromosome aneuploidy; Klinefelter's syndrome; Turner's syndrome.

INTRODUCTION

The study of psychological traits in carriers of chromosome aberrations may be a promising approach to the genetic–environmental analysis of

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behavior in man. Although there are inherent difficulties in establishing unequivocal relationships between phenotype variation and genome changes brought about by chromosome aberration, this kind of research can offer new facts and open new perspectives for the understanding of the genetic background of behavior.

To date, psychological traits have been studied mainly in XYY individuals and in subjects with Turner's or Klinefelter's syndrome, whose karyotype is 45,X or 47,XXY, respectively. Using Cohen's method (1957, 1959) for the analysis of WAIS scores, Shaffer (1962) found a low score for the two factors of "perceptual organization" and "freedom from distractability" in females with Turner's syndrome. In the same type of patients a specific cognitive deficit pertaining to space-form relations has been reported, which shows up, in the Wechsler intelligence test, in the non-verbal, mainly pictorial, constructional and numerical items, while verbal ability is not affected, which can produce a spurious indication of moderate retardation when the verbal and performance scores are combined (Money, 1963, 1964, 1965; Alexander et al., 1964; Alexander and Money, 1965; Money and Alexander, 1966; Garron and Van der Stoep, 1969; Money and Mittenthal, 1970; Garron et al., 1973). The results of cognitive tests given to males with Klinefelter's syndrome compared with those obtained in karyotypically normal patients affected with severe hypogonadism seem to indicate in the former significantly lower scores for "verbal comprehension" and "freedom from distractability" but normal scores for those cognitive functions in which perceptual organization or analytical ability plays a dominant role (Nielsen et al., 1969; Theilgaard et al., 1971).

A number of hypotheses have been formulated to relate these deficiencies in cognitive capacities to specific causes. It may be taken for granted that genetic, hormonal, psychological, educational, and sociocultural factors must interact in the expression of every subject's cognitive pattern. However, very little has been done so far to analyze the specific role played by each of these factors. In regard to the genetic component, one line of opinion, which was initiated by Vandenberg's research on twins (1962, 1969), is that some spatial abilities, such as "spatial visualization," and some cognitive styles, such as "field dependence," are determined by genes assignable to the X chromosome (Stafford, 1961; Garron, 1970; Hartlage, 1970; Bock and Kolakowski, 1973; Guttman, 1974; Goodenough et al., 1977). But, when analyzing from karyotype to phenotype, it should be remembered that no difference should be expected between sex chromosome aneuploids and karyotypically normal subjects of both sexes if only X-linked genes are involved in the determination of a particular trait. In fact, in man as in other mammals, the X chromosome undergoes dosage compensation through which, by an