Correspondence

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Sex chromatin and the biological effects of triploidy

Sir,

The causes underlying the pathological defects in triploid fetuses and newborn infants continue to baffle cytogeneticists. Gosden *et al.* (1976) cite sex chromatin findings as evidence for a disturbed relation between regulatory and structural genes in the presence of three sets of chromosomes. I would like to suggest, however, that the lack of constancy of sex chromatin data reported in triploidy is a result of the terminology used rather than of a lack of stability inherent in the triploid condition.

Theoretically, the incidence of sex chromatin in cells can vary between 100 and 0%. However, much of the data are not reported in terms of percentage incidence but as 'sex chromatin positive' and 'sex chromatin negative'. If we assume that the incidence of sex chromatin bodies per cell found in different tissues of diploid individuals with 46,XX chromosomes varies between 90 and 20%, all such individuals will clearly be 'positive'. However, additional sets of autosomes have the effect of suppressing sex chromatin formation (Harnden, 1961). Thus, though the maximum number of sex chromatin bodies is the same in triploid and diploid cells with the same number of X chromosomes (Mittwoch *et al.*, 1963), the tendency to form bodies is lower in the triploid cells, thus reducing the incidence of sex chromatin bodies (Edwards *et al.*, 1967). If we assume that the incidence of sex chromatin bodies in triploid cells with 69,XXY chromosomes varies between 40 and 0%, the findings will be reported as falling into two classes, 'sex chromatin positive' and 'sex chromatin negative', even though the range of sex chromatin incidence is no greater in triploid than in diploid cells.

The same reasoning applies to cells containing three X chromosomes. In diploid 47,XXX cells there will be a sizeable proportion of cells containing two sex chromatin bodies, so that all individuals and tissues can be reported as 'double positive'. By contrast, the proportion of cells with two sex chromatin bodies in triploid 69,XXX cells will be small, thus giving rise to 'double positive' and 'single positive' reports.

It seems unlikely that considerations of chromosome imbalance will materially advance our understanding of the pathogenesis of triploidy. On the other hand, it has been shown that the amount of nuclear DNA will affect mitotic cycle times in plants (Evans *et al.*, 1969) and in vertebrates (Grosset and Odartchenko, 1975). While these findings cannot be expected to provide a simple solution to the problem of how to explain the abnormal development of triploid zygotes, they are surely relevant to attempts at doing so (Mittwoch, 1973).

Yours, etc,
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Extra chromosome in Kallmann's syndrome

Sir,

I read with great interest the publication by Ventruto and colleagues (*Journal of Medical Genetics*, **13**, 71, 1976) which described a male with Kallmann's syndrome and a chromosomal constellation of 47,XY,mat?+. So far the genetic aspects of olfacto-genital dysplasia (Kallmann's syndrome) have been intriguing and puzzling. In most patients, male and female, a normal karyotype has been described. Yet in 1971, Agulhon and coworkers pointed out