Correspondence

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Computerised chromosome data bank

SIR,

I was preparing the material contained in the excellent report by Martin *et al.* (1977) of mosaic trisomy 14 in a liveborn infant for entry into my computerised data bank of *Catalog of Chromosomal Variations and Anomalies in Man* (Borgaonkar, 1975, 1977a), and came across this sentence: 'We report here a liveborn infant whose complement was 46,XX/47,XX,+14, the first such case reported to our knowledge'. The claim may indeed be justified, but if it implies that this is the first report of such a case in the literature then it is misleading. Rethoré *et al.* (1975) have also reported a mosaic trisomy 14 newborn female infant.

I have pointed out elsewhere similar instances of misleading claims (Yu *et al.*, 1976; Borgaonkar, 1977b) and have desisted from pointing out many others.

What is needed is a central data bank wherein the reports of chromosomal variants and anomalies are filed appropriately. Such a facility could then be called upon to provide service to investigators in bibliographic citations. This facility would be somewhat like the National Library of Medicine (USA) projects of MEDLINE, MEDLAR, and TOXLINE. Our data banks have been used by many cytogeneticists around the world precisely for that purpose. Additionally, such a facility, if properly directed, could be used in collating data which may be helpful in providing the descriptions of new chromosomal syndromes, as has been done by Martin *et al.* (1977), by us (Borgaonkar *et al.*, 1976), and by several authors (Yunis, 1977).

Yours, etc,

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References

- Borgaonkar, D. S. (1975). *Chromosomal Variation in Man. A Catalog of Chromosomal Variants and Anomalies*. The Johns Hopkins University Press, Baltimore and London.
Borgaonkar, D. S. (1977a). *Chromosomal Variation in Man. A Catalog of Chromosomal Variants and Anomalies*, 2nd ed. Alan R. Liss, New York.

Borgaonkar, D. S. (1977b). Y to X chromosome translocations. *Human Genetics*, **40**, 113–114.

Borgaonkar, D. S., Lacassie, Y. E., and Stoll, C. (1976). Usefulness of chromosome catalog in delineating new syndromes. *Birth Defects: Original Article Series*, **12** (No. 5), 87–95. The National Foundation—March of Dimes, New York.

Martin, A. O., Ford, M. M., Khalil, N. T., Turk, K. B., and MacIntyre, M. N. (1977). 46, XX/47, XX, +14 mosaicism in a liveborn infant. *Journal of Medical Genetics*, **14**, 214–218.

Rethoré, M. O., Couturier, J., Carpentier, S., Ferrand, J., and Lejeune, J. (1975). Trisomie 14 en mosaïque chez une enfant multimalformée. *Annales de Génétique*, **18**, 71–74.

Yu, C. W., Borgaonkar, D. S., and Bolling, D. R. (1976). Paracentric inversion in man. *Human Genetics*, **34**, 117.

Yunis, J. J. (Editor) (1977). *New Chromosomal Syndromes*. Academic Press, New York.

Heterozygote advantage for the phenylketonuria allele

SIR,

Further to the correspondence on heterozygote advantage for the phenylketonuria gene, though individual women tend to have pregnancies of similar gestational length, birthweight usually increases with parity. The increase is determined by the sex of the previous, as well as the later, child, and is usually greatest in the second pregnancy (Karn *et al.*, 1951; Trotnow *et al.*, 1976). When pregnancy is allowed to proceed naturally, mean increase between first and second child equals the discrepancy in birthweight between the sexes (approximately 140 g). This is the case in present day Norway and Denmark, less so in Scotland and Germany, and in USA mean increase is negligible (Table). In England, no information is available after 1958 concerning mean increase between first and second child. The 138 g mean increase between the first and later-born in England in 1970 is of uncertain importance, since we have no information concerning the magnitude of the increase in birthweight between consecutive later pregnancies. According to Baird (1976), the policy of induction, aimed at forestalling death, was determined by the epidemiological findings in Aberdeen in the late 1940s when more than 20% of all first births were to women 30 years of age or more. In addition 40% of all perinatal deaths in first pregnancy were the result either of birth trauma or of placental insufficiency, and the pregnancy had lasted 42 weeks or more in more than