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,X/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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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 foetal mortality, 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

References


Conceiving a continuation of the secular downward trend in birthweight observed in England between 1958 and 1970, it ought to be mentioned that owing to the adverse effect on mean increase with parity of widespread use of elective delivery, the reducing effect on birthweight of declining parity may be less important than expected. This is reported from the USA where between 1960 and 1973 mean parity declined from 2.92 to 2.13, but the effect on birthweight was negligible (10 g or so). It would have approached 50 g with similar increase in birthweight with parity in Norway and Denmark (Saugstad, 1978b).

As regards the later pregnancies of heterozygotes for PKU, who have already given birth to a PKU homozygote, labour is likely to be induced more frequently than usual, if not terminated by an abortion.

Yours, etc,

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References


Correspondence


Complete trisomy 5p

Sir,

In their recent report on ‘Complete trisomy 5p’, Brimblecombe et al. (1977) imply that the phenotype in partial trisomy 5p is nonspecific, and that asymmetry of the jaw is a common finding. In our recent review on partial trisomy 5p, we suggested that there was a recognisable and consistent phenotype based on a review of published cases and 3 additional patients of our own (DiLiberti et al., 1977). The consistent findings include a full face with the appearance of jowls, apparent ocular hypertelorism, bulbous nose, long philtrum, full lips with down-turned corners of the mouth, and mild malar hypoplasia. Asymmetry of the jaw was infrequent. It is of interest that the photograph of the patient of Brimblecombe et al. suggests most of these features and, in fact, with the exception of the ear and eye anomalies, looks remarkably like several of the young patients with partial trisomy 5p.

Additional features present in most patients were an excess of ulnar loops, arch tibial patterns in the hallucal area, and seizures. None of these is commented upon in the current report.

Since the majority of patients with partial trisomy 5p lack significant malformations, it is tempting to speculate that the extra chromosomal material distal to band p13 produces the characteristic facial appearance and retardation, and that the more proximal portion is responsible for the structural abnormalities of the eye and ear.

Yours, etc,

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References


This letter was shown to Drs Brimblecombe and Vowles who reply as follows:

Sir,

Thank you for the opportunity to reply to the letter of Dr DiLiberti. Despite the fact that DiLiberti et al. (1977) suggest that there is a clinically indentifiable syndrome for partial trisomy 5p, we still maintain that there appears to be some overlap in the clinical findings described in trisomy of the short arms of chromosomes 4 and 5.

Owen et al. (1974) compared the clinical features of 1 definite and 1 probable previously described cases of trisomy of the short arm of chromosome 4 with those of their own case. Darmady and Seabright (1975) described a further case involving most of the short arm of this same chromosome. Each of these 4 cases had between 1 and 4 of the following features described by DiLiberti et al. (1977): ocular hypertelorism, bulbous nose, long philtrum, impaired mobility of elbow joints, high dermal ridge count, and mental retardation. Furthermore, they each had between 3 and 6 different features in common with our case of total trisomy 5, such as scaphocephaly, macroglossia, malformed ears, microphthalmia, narrow palpebral fissures, long fingers and toes, scoliosis, and talipes.

Tempting though it is to speculate that certain phenotypic characteristics can be assigned to different portions of the chromosome, we do not feel that this is justified as yet in the light of the above observations.

Yours, etc,

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Dominance in human genetics

The following letter from Professor E. B. Ford, FRS, arises from a definition of dominance which appears in the glossary of ‘Human Genetics and Medicine’ by C. A. Clarke (1977). This reads: ‘Dominance: A character is said to be dominant if the gene controlling it produces the same effect in the heterozygous as in the homozygous state. Loosely, however, for clinical purposes, a condition is said to be dominant if it appears in heterozygotes. The homozygote has often never been observed’.

References


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L F Saugstad

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