Predisposition to spina bifida

SIR,
Last year, some of us (Stanway *et al.*, 1977) published a study of the gastric acid secretion in mothers of spina bifida offspring in southern England. There was no significant difference in mean serum level of group I pepsinogens (used as an indicator of gastric secretion) between the index mothers and matched control mothers. This result suggested that acid-labile teratogens are not a major factor in the causation of spina bifida in the UK.

An incidental and unexplained finding was that, among the index mothers, the observed variance of the concentrations was significantly larger than it was among the control mothers. There is no appealing biological explanation for this so the alternative interpretation, chance, might be appropriate.

Table Serum level of group I pepsinogens by age of Leeds mothers of a spina bifida offspring

Age (y)	Serum level of group I pepsinogens (ng/ml)
17-18	54
19-20	45, 65, 105
21-22	40, 45, 51, 69
23-24	33, 42, 43, 51, 58, 121
25-26	30, 31, 39, 50
27-28	45, 65
29-30	60
31-32	46, 71
Mean	54.7
SD	21.8

We now have some additional results which favour this interpretation. Twenty-three mothers who gave birth to offspring with neural tube malformations, and who were living in the Leeds area, had their group I pepsinogen concentrations recorded (Table). These concentrations do not show the high variance, 1227, noted in the corresponding data from the south. Indeed, the estimated variance, 475, is closely comparable with that for southern control mothers, 464. (These, of course, were not matched with the Leeds mothers.) Thus, even in the absence of suitable controls, the extra data do not seem to support any biological interpretation of the large variance among the index mothers from southern England.

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Reference
Stanway, P. A., Samloff, I. M., Brace, C. J., and Renwick, J. H. (1977). Predisposition to spina bifida: search for a relation to maternal gastric acid secretion. *Journal of Medical Genetics*, 14, 332-334.

A 'new' syndrome of mental retardation with characteristic facies and brachyphalangy

SIR,
Dr Hunter *et al.* described "A 'new' syndrome of mental retardation with characteristic facies and brachyphalangy" on page 430 of the December 1977 issue of this journal. I wonder if I might tentatively suggest that this is not a new syndrome but a variant of the trichorhinophalangeal syndrome. The photographs of the affected individuals, published in their report, show the characteristic pear shaped noses and early balding which are features of this syndrome. The x-ray changes in the hands show coning of the epiphyses and the affected individuals also had shortness of stature, mild mental retardation, and dominant inheritance with variable expression, all of which are features of the trichorhinophalangeal syndrome.
This in no way detracts from their report which broadens our understanding of this entity and includes exact details of the changes in measurement of the phalanges.
In the description of their family they do not consider the trichorhinophalangeal syndrome in their differential diagnosis and, therefore, I wonder if this entity may have escaped their notice.

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This letter was shown to Dr Hunter *et al.* who reply as follows:

SIR,

We are grateful for the opportunity to reply to Dr Turner's kind letter. Omission of a discussion of the trichorhinophalangeal syndrome from our paper was an oversight, as the diagnosis was considered. The trichorhinophalangeal syndrome (TRP) is subdivided into type I and type II (Langer-Giedion) syndromes (Stolzfus *et al.*, 1977). The latter is characterised by sporadic occurrence, a distinct facial appearance, skin and joint laxity early in life, and later onset of multiple exostoses. This condition is clearly distinct from that seen in our family. There were several findings in our family that led us to discard a diagnosis of type I TRP. The severe degree of retardation seen in V.1 and IV.5 is not a feature of type I TRP. Our patients lacked prominent ears, they had almond shaped eyes, a small mouth, and, though IV.5 does appear to have a rather 'pear shaped' nose, the children typically had small blunt noses rather than the bulbous, 'pear shaped' noses seen in TRP type I. The hair is described as sparse in patients with TRP type I and, though several of our patients had early pattern baldness, their hair was not sparse (V.1 in Fig. 2a is shaven). Perhaps the most interesting and least subjective difference is seen in the metacarpal-phalangeal profiles of the two conditions. Our patients showed a marked relative shortness of all distal phalanges and middle phalanges, whereas patients with TRP type I have relative shortness of the metacarpals, middle phalanges, and first distal phalanx, the second to fifth distal phalanges being of normal length (Say *et al.*, 1977).

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- Say, B. S., Barber, B., and Poznanski, A. K. (1977). Pattern profile analysis of the hand in trichorhinophalangeal syndrome. *Pediatrics*, **59**, 123-126.
Stolzfus, E., Ladda, R. L., and Lloyd-Still, J. (1977). Langer-Giedion syndrome: type II tricho-rhino-phalangeal dysplasia. *Journal of Pediatrics*, **91**, 277-280.

Supernumerary small ring chromosome

SIR,

We read with interest the case of 'Supernumerary small ring chromosome' reported by Kaffe *et al.* which was published in the December 1977 issue of *Journal of Medical Genetics*. We want to refer to a case previously published in the *Italian Journal of Pediatrics* (Calabro *et al.*, 1977) in the hope of contributing to a better delineation of the phenotype of subjects with this chromosomal aberration.

Our patient was born at term after a normal pregnancy to a 24-year-old mother and 26-year-old father. The delivery was performed with the help of a vacuum extractor. Apgar score at 5 minutes was 5. Birth weight was 3850 g. There was a history of generalised convulsions with electroencephalographic anomalies for which the patient had been on antiepileptic therapy since the neonatal period.

She came to our notice at 6 years of age. The anthropometric measurements were all between the 25th and 50th centile. The following dysmorphic features were observed: large prominent nasal root and broad nose tip, and downward slanted palpebral fissures. A mild epicanthus was present bilaterally and there was macrostomia with malaligned teeth, enamel hypoplasia, prognathism, and low-set ears with large lobes. Dyspraxia and mild generalised hypotonia were also present and the mental age was 3 to 4 years.

Dermatoglyphs showed a prevalence of ulnar loops and the palmar axial triradius was in the 't' position bilaterally. Routine laboratory tests were normal.

Chromosome analysis, performed on peripheral leucocytes, showed a mosaicism 46,XX/47,XX+ring, with a predominance of the aneuploid line of 91%. The supernumerary chromosome was smaller than a G chromosome, monocentric, and relatively stable. The different banding techniques, GAG, RBA and CBG, showed that the structure of the ring was very similar to the one present in the case reported by Kaffe *et al.*, but in our case, as in the case of Kaffe more precise definition of the nature of the ring chromosome was not possible.

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