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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