tracheo-oesophageal dysraphism, at least in populations with a low frequency of NTD.

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References
338–40.

This letter was shown to Dr David, who replies as follows.

Ilyina and Lurie’s letter is most welcome, though I would like to get my hands on whoever first used the awful term ‘tracheo-oesophageal dysraphism’. Oesophageal atresia is a most neglected defect, and it is sad that current interest mainly centres not on the defect itself, but either with the probably spurious increase of neural tube defects in sibs, or with the VATER or VACTERL association, which embodies the statistical and teratological misconception of a non-random association of defects.

Ilyina and Lurie’s hypothesis that there may be a relationship between the risk of neural tube defect for sibs of patients with oesophageal atresia and the population frequency of neural tube defects may be right. It is perhaps akin to the suggestion that the recurrence risk of neural tube defects is to some extent a function of the background population risk. However, the general notion of an increased frequency of neural tube defects in the sibs of children with other malformations is most likely to be attributable to sampling errors or bias.

The cases of Fraser and Nussbaum were a highly selected group and in no way representative of the general population of patients with oesophageal atresia, and the same applies to the data of Warren et al. It is not clear how the cases of Ilyina and Lurie were ascertained, but it is likely that they were selected in some way. Our own study was happily free from this defect, only to fall victim to the entirely fair criticism that we did not seek to identify all sibs but just used available medical records. Probably the only suitable data come from Sweden and Canada, and from these studies it does seem that there is no increase in neural tube defects in the sibs of patients with oesophageal atresia.

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References
338–40.

Routine diagnostic detection of the fragile X

SIR,

I enjoyed reading the recent report by Dr McDermott and his colleagues (J Med Genet 1983;
20:169–78) on the fragile X chromosome. I would, however, like to take issue with their assertion that busy diagnostic cytogenetics laboratories cannot routinely screen unselected cases for the fragile X chromosome. Furthermore, their request that referring practitioners alert the laboratory to the possibility of this finding on clinical grounds is one which simply cannot work. While some males have the full ‘fragile X syndrome’, so well described by McDermott et al., and a family history suggesting the presence of the fragile X many, particularly children, have neither. About one-third of females...