Correspondence

Journal of Medical Genetics, 1985, **22**, 319–323

Triple aneuploidy

SIR,

Webb *et al* (*J Med Genet* 1984;**21**:232) state “This is the first reported case of triple aneuploidy in a male, that is, additional chromosomes X, Y, and 18” while publishing their very interesting case report on 49,XXYY,+18 in a liveborn male.

However, there is a report of triple aneuploidy, that is, additional chromosomes X, Y, and 8 by Sutherland *et al*¹ (cited by me²) in a mosaic state, mos48,XXYY/49,XXYY,+8, found at paediatric necropsy.

DIGAMBER S BORGAONKAR
Cytogenetics Laboratory,
Wilmington Medical Center,
PO Box 2850,
Wilmington, Delaware 19805, USA.

References

- ¹ Sutherland GR, Carter RF, Bauld R, Smith I, Bain AD. Chromosome studies at the paediatric necropsy. *Ann Hum Genet* 1978;**43**:173–81.
- ² Borgaonkar DS. *Chromosomal variation in man. A catalog of chromosomal variants and anomalies*. 3rd ed. New York: Alan R Liss, 1980:558. (A fourth edition, 1984, is now available.)

would seem that the mean time of conception and of fetal developmental stages relative to ovulation, and to the whole sequence of maternal endocrinological events following it, is earlier in the case of males than females. In other words, the sequence of maternal events occurs at slightly different developmental stages, on average, for fetuses of the two sexes (at least early in pregnancy). If this is so, then the effect of therapy may be to alter the rate at which one of these sequences occurs so as better to synchronise the mother with the fetus. From conception on, male fetuses—or rather those fetuses which will later turn out to be males—are perhaps better synchronised on average with the sequence of maternal events than are female fetuses, from the point of view of NTD.

Many congenital malformations are sex selective,⁵ and one might wonder whether this is related to the proposed difference between male and female fetuses in regard to developmental timing vis-a-vis maternal timing.

WILLIAM H JAMES
MRC Mammalian Development Unit,
Wolfson House,
4 Stephenson Way,
London NW1 2HE.

References

- ¹ Seller MJ, Nevin NC. Periconceptional vitamin supplementation and the prevention of neural tube defects in south-east England and Northern Ireland. *J Med Genet* 1984;**21**:325–30.
- ² James WH. The sex ratio in anencephaly. *J Med Genet* 1979;**16**:129–33.
- ³ James WH. Gonadotrophin and the human secondary sex ratio. *Br Med J* 1980;**281**:711–2.
- ⁴ James WH. Time of fertilisation and sex of infants. *Lancet* 1980;**i**:1124–6.
- ⁵ Arena JFP, Smith DW. Sex liability to single structural defects. *Am J Dis Child* 1978;**132**:970–2.

Clinical features of homozygous $\alpha 2(I)$ collagen deficient osteogenesis imperfecta

SIR,

I have read with interest the above article by Nicholls *et al* (*J Med Genet* 1984;**21**:257–62). I would, however, like to take issue on the comments made about the teeth. On page 260 it is stated in the text “. . . relatively normal teeth without dentinogenesis” and the caption for fig 5(e) mentions

A possible mechanism underlying the sex selectivity of neural tube defect

SIR,

Dr Seller and Professor Nevin¹ find that the recurrences of neural tube defect (NTD) in the presence of vitamin supplementation are predominantly in male fetuses. It has been suggested that female anencephalics are more frequently associated with environmental determinants than are male anencephalics.² Seller and Nevin remark that their finding supports the related supposition that the less commonly affected sex would respond less well to environmental therapy. They add “This fact is difficult to explain since the neural tube closes before sex differentiation has occurred”.

I should like to offer a form of explanation. I have suggested that the sex of the human zygote is partially controlled by the level of maternal gonadotrophin at the time of conception, high levels being associated with female offspring.^{3,4} If this is so, it