Gonadal dysgenesis with 46,X,Xt(qter→p221 :: p223→qter) karyotype

SIR,

It was gratifying to read the case report of Ferraro *et al* in the Journal (1980;17:457–63) of a case of gonadal dysgenesis with 46,X,Xt(qter→p221 :: p223→qter) karyotype, as their findings confirmed those we found in a case we described with virtually identical karyotype and phenotype.¹ Our case was not referred to by Ferraro *et al* in their tables 1 and 2. Five other reports which I know of were also not included in these tables.^{2–6}

This illustrates a commonly encountered problem in correlating cytogenetic case reports. As not all laboratories see all journals, authors should state that their comparisons with similar cases are based on those reports “known to us” rather than to state that they are based on “all available . . . findings . . .”.

ARABELLA SMITH
Cytogenetics Unit,
Oliver Latham Laboratories,
Macquarie Hospital,
North Ryde,
New South Wales 2113,
Australia.

References

- ¹ Smith A, Donnelly PE, Elliott G, den Dulk G. An abnormal dicentric X chromosome in a patient with short stature and gonadal dysgenesis. *Ann Genet (Paris)* 1979;22:143–7.
- ² Pena J, Pombo M, Martinon JM, Ansedé A, Noya M. Fusion of the short arms of 2 X chromosomes in a patient with phenotype of achondroplasia. *J Genet Hum* 1977;25:215–20.
- ³ Becroft DMO, Costello JM, Shaw RL. 46,X,-XX terminal re-arrangement/45,X mosaicism in a child with short stature. *Clin Genet* 1977;11:122–7.

⁴ Valenta LJ, Higgins JV, Holzman GB. Ovarian dysgenesis due to 45X, 0/46 Dic(X) mosaicism. *J Clin Endocrinol Metab* 1977;45:702–6.

⁵ Schwanitz G, Tietze U, Pfeiffer RA, Grosse KP, Becker H, Egger H. Duplication deficiency of an X-chromosome with and without 45,X mosaicism in three girls. *Acta Genet Med Gemellol* 1977;26:277–81.

⁶ Stoll C, Lausacker C, Pennerath A. A girl with an end-to-end fusion of two X's. *Eur J Pediatr* 1979; 131:141–5.

This letter was shown to Dr Ferraro who replies as follows.

SIR,

We believe that the letter by Arabella Smith deserves a few comments.

(1) A clinical case is a single, peculiar, observation which cannot be confirmed or dismissed by any other paper.

(2) When we wrote our paper we did not know about her case report, which was the prime cause of the controversy. Our paper was sent to this Journal on 9 October 1979.

(3) Published papers we did not refer to were mostly either only clinically or cytogenetically oriented. In fact, we selected—let us complete Dr Smith's quotation—“all available clinical and chromosomal findings on *cytologically well documented* X;X translocations . . .” where “well documented” means, for instance, inclusive of replication studies.

M FERRARO
Istituto di Genetica,
Università degli Studi di Roma,
Città Universitaria,
00185 Roma,
Italy