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. 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 . . .”.

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References

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.

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