Proximal 15q monosomy

SIR,

We should like to comment upon the letter of Duckett¹ which criticises our case report of a retarded male with deletion of proximal chromosome 15 and terminal chromosome 10q.²

Firstly, the case is reported as a Short Report, the stipulations for which are that it takes only one page of the journal with no more than three references. It is not meant to be a review of the literature, but to state concisely and simply the salient features of the case. Therefore we did not list all the published papers we referred to which did not show monosomy 10q, but merely cited one reputable book to indicate that we had indeed looked. Also we could not include a lengthy discussion of the relationship of proximal 15q monosomy and the Prader-Willi syndrome (PWS), of which we are well aware,³ and as our patient clearly did not have the PWS we referred to “one similar case”. We did not profess to review the literature.

Secondly, we submitted our paper to the Editor in February 1981, 6 months before the paper of Duckett and Roberts⁴ appeared and 12 months before the paper of Ledbetter *et al.*⁵ My previous sentiments remain unchanged,⁶ namely that it is extremely difficult to review the literature ‘thoroughly’ in a given case. I make the point of referring to cases “known to us” or those “which we have found”.

Finally, we do believe that more cases of proximal 15q— are needed to enable an unbiased evaluation of this effect. While an association between PWS and a variety of chromosome 15 abnormalities is apparent, it is not proven that the PWS is ‘caused’ by proximal 15q deletion. Hawkey and Smithies⁸ report four cases of PWS owing to chromosome abnormality of chromosomes other than 15 and Guanti⁹ cites one further case. We found 24 cases of PWS in New South Wales and Victoria with a normal karyotype and two with an abnormal karyotype⁷ (one 47,XXY and one translocation³). Ledbetter *et al.*⁵ discuss several aspects of this problem and we would like to add our case as an example of proximal 15q— producing abnormalities not of the PWS type, of which one other case with similar abnormalities has also been reported. Duckett and Roberts⁴ themselves state “. . . no consistent phenotypic abnormality has

been delineated for partial monosomy 15 making identification of its effects difficult”.

ARABELLA SMITH AND GESINA DEN DULK
Cytogenetics Unit,
Oliver Latham Laboratory,
Macquarie Hospital,
North Ryde,
NSW 2113,
Australia.

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Serum gonadotrophins in Down's syndrome

SIR,

In the paper entitled ‘Serum gonadotrophins in Down's syndrome’ (*J Med Genet* 1982;19:98–9), the authors omitted to mention a study published in 1980¹ in which similar data and conclusions were presented. We feel that our data on 39 patients (both men and women), when added to those reported in the paper under discussion, are mutually supportive and constitute a good point of departure for further studies on this topic.