Correspondence

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A study of retinitis pigmentosa in the City of Birmingham

SIR,
We think it useful to add some follow up information to the above papers.¹

Firstly, since the paper was submitted for publication in May 1984, we have not ascertained, either in the Retina Clinic of the Birmingham and Midland Eye Hospital, or in the Diabetic Clinic of the General Hospital, any further patients with retinitis pigmentosa who had symptoms on prevalence day. We therefore believe that our observations on the prevalence of retinitis pigmentosa¹ are correct.

Secondly, we had attempted by examination of relatives to identify the type of retinitis pigmentosa occurring in 12 severely affected male index patients who had no symptomatic relative. Three of the 12 patients were recognised as having X linked retinitis pigmentosa, one did not have X linked disease because his adult daughter was not a carrier, and eight could not be classified. Two of those eight men are now discovered to have X linked disease, as the young sons of a sister and a daughter respectively have become affected. Their mothers were not available for earlier examination. This means that the likelihood of an isolated male with severe retinitis pigmentosa having the X linked form is about 1 in 2. In our whole series (regardless of severity or family history) we now have 21 male index patients out of 74 with X linked disease, an occurrence of about 1 in 3.5.

We have not yet encountered a male with X linked retinitis pigmentosa whose mother had healthy retinae.

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Reference

- ¹ Bunday S, Crews SJ. A study of retinitis pigmentosa in the City of Birmingham. I Prevalence. II Clinical and genetic heterogeneity. *J Med Genet* 1984;21:417–28.

Cornelia de Lange syndrome

SIR,

I was interested to read the article by Kumar *et al*¹ describing Cornelia de Lange syndrome in several members of the same family. My first impressions were that neither the facial features nor the hand abnormalities were really consistent with the diagnosis of de Lange syndrome. A diagnosis of Ruvalcaba syndrome² seems at least a possibility. The facial features and hand abnormalities are very similar, and the inheritance with variable dominant transmission would also fit. A relatively loose search of a computerised data base³ of 1250 syndromes from published reports showed 21 syndromes with the combination of eyebrow and hand anomalies. Of these, only the Ruvalcaba syndrome fitted the cases of Kumar *et al* (if one excludes the possibility of de Lange syndrome).

Radiographs of the hands, feet, and spine in the cases of Kumar *et al* would be most instructive, as these would most likely provide extra support for the diagnosis of Ruvalcaba syndrome.

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References

- ¹ Kumar D, Blank CE, Griffiths BL. Cornelia de Lange syndrome in several members of the same family. *J Med Genet* 1985;22:296–300.
² Ruvalcaba RHA, Reichert A, Smith DW. A new familial syndrome with osseous dysplasia and mental deficiency. *J Pediatr* 1971;79:450–5.
³ Winter RM, Baraitser M, Douglas JM. A computerised data base for the diagnosis of rare dysmorphic syndromes. *J Med Genet* 1984;21:121–3.

A pedigree study of perinatally lethal renal disease

SIR,

Bankier *et al* (*J Med Genet* 1985;22:104–11) are to be congratulated on their thorough study of perinatally lethal renal disease. Of interest, with regard to the group classified by them as multiple defects, is our case.