Hereditary brachydactyly with nail dysplasia

SIR,

In the April 1978 issue of the *Journal of Medical Genetics*, a family is reported with hereditary brachydactyly and nail dysplasia (Schott, 1978). The author suggests that this is a new syndrome. However, both the external appearance of the hands and the x-rays are very similar to type B brachydactyly (Battle *et al.*, 1973). I have recently reviewed the inherited brachydactylies (Fitch, 1979) and in 24 of 41 x-rays of type B the thumbs were normal. The simultaneous occurrence of nail and digit abnormalities has been discussed in a most interesting way by MacArthur and McCullough (1932).

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References

- Battle, H., Walker, N., and Thompson, M. (1973.) MacKinder's hereditary brachydactyly. *Annals of Human Genetics*, **36**, 415-424.
Fitch, N. (1979). Classification and identification of inherited brachydactylies. *Journal of Medical Genetics*, **16**, 36-44.
MacArthur, J., and McCullough, E. (1932). Apical dystrophy. *Human Biology*, **4**, 179-207.
Schott, G. D. (1978). Hereditary brachydactyly with nail dysplasia. *Journal of Medical Genetics*, **15**, 119-122.

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

SIR,

I was most interested in Dr Fitch's comments on my paper on hereditary brachydactyly and nail dysplasia. The main difference between the family I reported and those families Dr Fitch refers to is the complete absence of any abnormality, clinical or radiological, involving the feet of affected patients. Another minor point is that neither among those members of the family personally seen nor, as far as could be ascertained, among other members of the family, were other abnormalities such as syndactyly, double nails, and bifid and double phalanges encountered.

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The Rieger syndrome

SIR,

The Rieger syndrome, as reported by Fitch and Kaback (1978), is a heterogeneous malformation syndrome. We have recently prepared a paper on our experiences with several families with the Rieger syndrome and with a consequent review of published reports (Jorgenson *et al.*, 1979). On the basis of the cases studied, we defined the Rieger syndrome as the autosomal dominant association of goniodysgenesis, hypodontia, and failure of involution of the periumbilical skin. We also mentioned the possible association of hypospadias. On the basis of the review of published reports, we concluded that at least six syndromes have been described as the Rieger syndrome: anal atresia and goniodysgenesis, deafness and goniodysgenesis, myopathy and goniodysgenesis, arachnodactyly and goniodysgenesis, the SHORT syndrome, and the Rieger syndrome. Fitch and Kaback list another association that may be more than coincidental and that we had overlooked: the association of ocular albinism and goniodysgenesis.

The paper by Fitch and Kaback also supports our statement that inappropriate terminology has led to extensive confusion about the nature of the Rieger syndrome. Our preference for the term goniodysgenesis is, in part, an effort to circumvent preconceived notions about such terms as Rieger anomaly and Axenfeld anomaly, and to stress that these latter malformations constitute a continuum.

Our only criticism of the paper under discussion pertains to the family study. The proband is described in the text as having only goniodysgenesis, but is indicated to have the Rieger syndrome in the pedigree. Furthermore, if one considers posterior embryotoxon, the Axenfeld malformation, and the Rieger malformation as a continuum (goniodysgenesis), three generations of goniodysgenesis are shown. Such an observation is compatible with earlier reports that any one of these malformations may be inherited as an isolated autosomal dominant trait.

Goniodysgenesis, then, may be an isolated malformation or one component of several malformation syndromes. The best way to differentiate the isolated from the associated malformation, and one of the malformation syndromes from another, is a careful family study. Attention must be paid to the presence of each minor clinical variation, regardless

of how innocuous it seems, and a pedigree of the type shown by Drs Fitch and Kaback must be constructed and properly interpreted.

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References

Fitch, N., and Kaback, M. (1978). The Axenfeld syndrome and the Rieger syndrome. *Journal of Medical Genetics*, **15**, 30–34.

Jorgenson, R. J., Levin, L. S., Cross, H. E., Yoder, F., and Kelly, T. E. (1978). The Rieger syndrome. *American Journal of Medical Genetics*, **2**, 307–318.

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

SIR,

Dr Jorgenson *et al.* correctly point out that Rieger eye malformations should replace Rieger syndrome in the pedigree. No-one in our family had failure of involution of the periumbilical skin. It is very exciting to be able to report that ophthalmological examination of the propositus at 9 months of age (Dr Saheb) showed a notable improvement in both eyes.

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Errata

In the October 1978 issue of *Journal of Medical Genetics*, an error appeared on page 348. The note under the Table should read 'C, cysts' not 'C, carcinoma of colon'.

In the December 1978 issue, a line was omitted on page 464. The first sentence of the second column should read: 'The figures also imply gene frequencies between 0·0012 and 0·0044, and heterozygote carrier frequencies ranging from 0·0025 to 0·008, that is, 1 in 400 to 1 in 125'.