

This letter was shown to Dr Hunter *et al.* who reply as follows:

SIR,

We are grateful for the opportunity to reply to Dr Turner's kind letter. Omission of a discussion of the trichorhinophalangeal syndrome from our paper was an oversight, as the diagnosis was considered. The trichorhinophalangeal syndrome (TRP) is subdivided into type I and type II (Langer-Giedion) syndromes (Stolzfus *et al.*, 1977). The latter is characterised by sporadic occurrence, a distinct facial appearance, skin and joint laxity early in life, and later onset of multiple exostoses. This condition is clearly distinct from that seen in our family. There were several findings in our family that led us to discard a diagnosis of type I TRP. The severe degree of retardation seen in V.1 and IV.5 is not a feature of type I TRP. Our patients lacked prominent ears, they had almond shaped eyes, a small mouth, and, though IV.5 does appear to have a rather 'pear shaped' nose, the children typically had small blunt noses rather than the bulbous, 'pear shaped' noses seen in TRP type I. The hair is described as sparse in patients with TRP type I and, though several of our patients had early pattern baldness, their hair was not sparse (V.1 in Fig. 2a is shaven). Perhaps the most interesting and least subjective difference is seen in the metacarpal-phalangeal profiles of the two conditions. Our patients showed a marked relative shortness of all distal phalanges and middle phalanges, whereas patients with TRP type I have relative shortness of the metacarpals, middle phalanges, and first distal phalanx, the second to fifth distal phalanges being of normal length (Say *et al.*, 1977).

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#### References

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- Stolzfus, E., Ladda, R. L., and Lloyd-Still, J. (1977). Langer-Giedion syndrome: type II tricho-rhino-phalangeal dysplasia. *Journal of Pediatrics*, **91**, 277-280.

#### Supernumerary small ring chromosome

SIR,

We read with interest the case of 'Supernumerary small ring chromosome' reported by Kaffe *et al.* which was published in the December 1977 issue of *Journal of Medical Genetics*. We want to refer to a case previously published in the *Italian Journal of Pediatrics* (Calabro *et al.*, 1977) in the hope of contributing to a better delineation of the phenotype of subjects with this chromosomal aberration.

Our patient was born at term after a normal pregnancy to a 24-year-old mother and 26-year-old father. The delivery was performed with the help of a vacuum extractor. Apgar score at 5 minutes was 5. Birth weight was 3850 g. There was a history of generalised convulsions with electroencephalographic anomalies for which the patient had been on antiepileptic therapy since the neonatal period.

She came to our notice at 6 years of age. The anthropometric measurements were all between the 25th and 50th centile. The following dysmorphic features were observed: large prominent nasal root and broad nose tip, and downward slanted palpebral fissures. A mild epicanthus was present bilaterally and there was macrostomia with malaligned teeth, enamel hypoplasia, prognathism, and low-set ears with large lobes. Dyspraxia and mild generalised hypotonia were also present and the mental age was 3 to 4 years.

Dermatoglyphs showed a prevalence of ulnar loop and the palmar axial triradius was in the 't' position bilaterally. Routine laboratory tests were normal.

Chromosome analysis, performed on peripheral leucocytes, showed a mosaicism 46,XX/47,XX ring, with a predominance of the aneuploid line of 91%. The supernumerary chromosome was smaller than a G chromosome, monocentric, and relatively stable. The different banding techniques, GAG, RBA and CBG, showed that the structure of the ring was very similar to the one present in the case reported by Kaffe *et al.*, but in our case, as in the case of Kaffe more precise definition of the nature of the ring chromosome was not possible.

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#### Reference

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