ANOTHER PHENOCOPY FOR CHONDRODYSPLASIA PUNCTATA IN ADDITION TO WARFARIN EMBRYOPATHY?

To the Editor: In 1966, Di Saia[1] reported the association of maternal warfarin anticoagulant therapy with a disorder characterized by a hypoplastic nasal cartilage and stippled epiphyses. This was regarded as a phenocopy of Conradi-Hünermann syndrome (chondrodysplasia punctata) because of identical stippled mineralization of the epiphyses found in both conditions (Fig. 1).

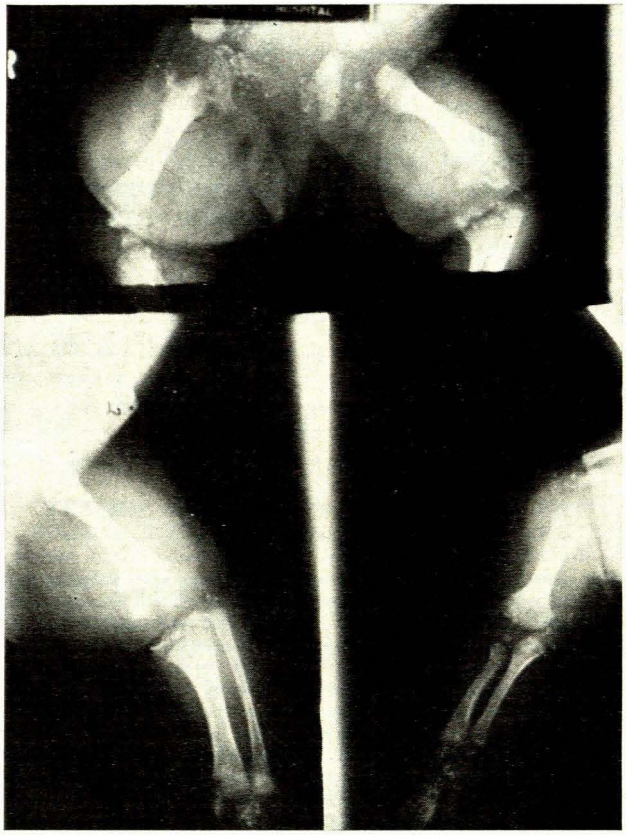

Fig. 1. Autosomal recessive rhizomelic chondrodysplasia punctata.

We have recently seen a Coloured female child with features suggesting warfarin embryopathy.[2] She had severely deficient nasal cartilage causing obstructed breathing (Fig. 2) in association with radiological findings of chondrodysplasia punctata, although less severe and not associated with rhizomelic dwarfism, as shown in Fig. 1. In addition, the distribution of stippling displayed by the radiographs of this patient corresponded to previous descriptions,[3] by being present mainly along the vertebral column, the sacral area and the proximal femurs. No other stigmata of chondrodysplasia punctata were present. No evidence of warfarin prescription or ingestion could be found and neither had there been any indication for such therapy for the mother. She had attended the antenatal clinics regularly and had remained in perfect health throughout the pregnancy, which was carried to term. She took only iron and folic acid supplements. The delivery was uneventful and no neonatal problems were encountered. On examination at the age of 2 months the baby was completely normal in all respects other than those mentioned above. Consanguinity or increased parental age did not play a role.

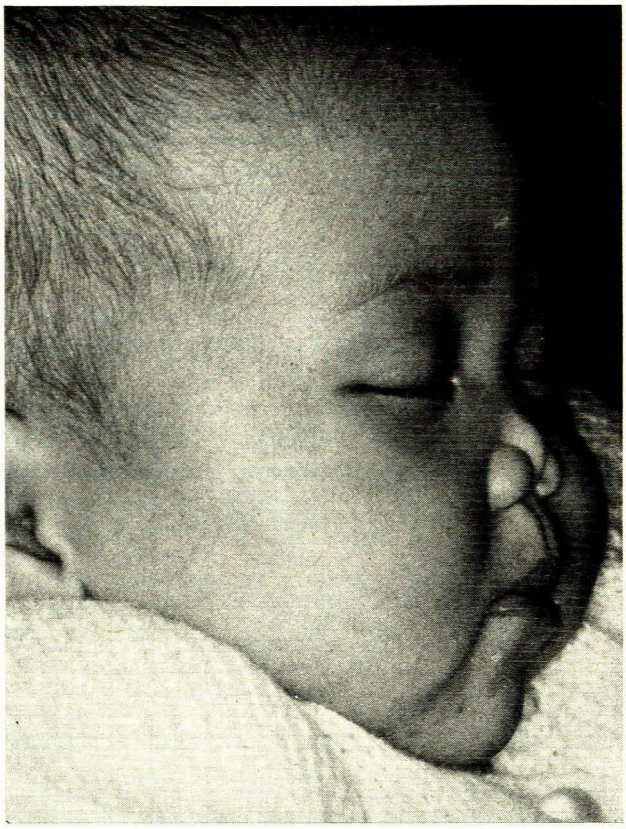

Fig. 2. Typical facies associated with warfarin embryopathy.

We therefore postulate that there may be factors other than warfarin which cause this phenotype. The apparent ease with which this mutant gene for epiphyseal development is switched on, as indicated by the existence of two syndromes (autosomal dominant and rhizomelic recessive types of chondrodysplasia punctata)[4] and two phenotypes, may stimulate further research into this specific chondrodysplasia.

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2. Smith, D. W. (1976): *Recognizable Patterns of Human Malformation*, 2nd ed., p. 342. Philadelphia: W. B. Saunders.
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4. Smith, D. W. (1976): *Op. cit.*[2], pp. 211 - 213.

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