Correspondence

we speculated that the karyotypic abnormality could be implicated in the infant’s disease.

Analyses of type I procollagen gene products synthesised by fibroblastic cells (#GM 09324, mother, and 09325, father; National Institute of General Medical Sciences Human Genetic Mutant Cell Repository, Coriell Institute for Medical Research, Camden, NJ, USA) cultured from biopsies of parental skin have now been completed. The techniques used are described elsewhere. The synthesis of proo1(1) and proo2(1) chains, the electrophoretic mobility of the chains, and the efficiency of secretion of the intact molecules were all normal. It seems unlikely that the karyotypic abnormality present in the mother and infant was responsible for the infant’s disease.

De novo mutations in type I collagen genes are the most frequent cause of lethal osteogenesis imperfecta. While it is possible that the rearrangement involving 7p13q22 could predispose to mutations which alter COL1A2, we have no direct evidence that supports this hypothesis. The product of this couple’s third pregnancy was normal by sonography at 18 weeks’ gestational age and the infant was normal at birth.

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References

Genetic heterogeneity in Waardenburg’s syndrome

Sir,

Since the original description, many associations and heterogeneity have been described in Waardenburg’s syndrome (WS). Here we report a study of three sibs of consanguineous parents (uncle–niece) with features suggestive of WS associated with obstructive ileal lesions, inherited as autosomal recessive trait, which we believe may be a variant of WS.

A female neonate was noted at birth to have a white forelock, bilateral blue irides, white eyelashes, a malformed right pinna, and multiple hypopigmented patches of varying sizes on the face (fig 1), both upper arms, and forearms. The inner canthal distance was 22 mm, interpupillary distance 44 mm, and outer canthal distance 70 mm, all within normal limits. Over the next 12 hours, the baby

FIG 1 White forelock, light coloured irides, and white eyelashes.

FIG 2 Atretic ileal segments.
Correspondence

Waardenburg’s syndrome has been described in many populations throughout the world.¹ However, the peculiar association of features suggestive of WS with total intestinal aganglionicism and ileal atresia and bands, manifesting in the neonatal period, with an autosomal recessive mode of inheritance, seems peculiar to the population of the Indian subcontinent.³ ⁴

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Current trends in the prevalence at birth of neural tube defects in Singapore

Sir,

A decline in the prevalence at birth of neural tube defects (NTD) has been reported in most western countries.¹ ² The reasons for the decline are unknown as the aetiology of NTD is still not well understood. Increased public health awareness, more demand for genetic counselling, improved maternal environment, better prenatal care, and early detection and termination of pregnancy have been suggested to be responsible for the reduction of the prevalence at birth of NTD.³ ⁴ On the other hand, some authors in the United Kingdom do not agree that prenatal screening and increase in the number of terminations of pregnancy are the main factors behind the recent decline.⁵ Furthermore, in the United States the decline started well before 1970 when screening was not widely available.⁶ ⁷

A retrospective study of NTD was conducted in our hospital to see if there was a change in the prevalence at birth of NTD in the last 12 years (1976 to 1987). In this period, as a general practice in this hospital, only mothers who had had an infant with

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