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Craniosynostosis

SIR,

In their interesting paper on families with craniosynostosis, Professor Carter and co-workers¹ suggest that our higher incidence of apparent autosomal dominant coronal synostosis may result from our inclusion of patients with Saethre-Chotzen and other syndromes among our study group.² Certainly, if the minor hand anomalies to which we referred have the same aetiology as the craniosynostosis, then some families had 'private' syndromes. We agree that family number 1 has what the authors call the 'split face syndrome', and that it is like the family reported by Slover and Sujansky.³ However, in the interest of clarity, we would like to point out that we did not make a diagnosis of Saethre-Chotzen syndrome unless at least one family

member had the typical syndactyly. Therefore, as the title implied, our families with Saethre-Chotzen syndrome were not included in the paper and we do not believe that families 4, 8, or 10 had this condition.

The degree to which the reported heterogeneity of the craniosynostosis syndromes represent true genetic heterogeneity must await biochemical or linkage markers for the genes or both. In the meantime it remains true that patients who have craniosynostosis in association with other dysmorphic features, notably of the hands, are more at risk to represent a single gene mutation than are those with isolated craniosynostosis.

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Pericentric inversion of chromosome 13

SIR,

In 1972 a paper from our laboratory described a large family with a pericentric inversion of chromosome 13, leading to a duplication deficiency

FIG 1 Pedigree of family.