Short reports

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Deletion of chromosome 5q and familial adenomatous polyposis

An interstitial deletion of 5q was observed in two intellectually handicapped brothers with familial adenomatous polyposis (FAP). The site of this deletion was similar to that observed by Herrera *et al*[1] in a patient with Gardner's syndrome and, following the work of Bodmer *et al*,[2] can be localised to the area 5q15 to q22. Their retarded mother died of an inoperable carcinoma of the colon with extensive polyposis. Her surviving sons, who had different fathers, have a number of dysmorphic features.

The two sibs were unknown to each other and had been fostered or adopted from birth.[3] The patients' mother, who died in her early forties of carcinoma of the colon with extensive polyposis, was also intellectually handicapped and had a third child who died aged a few months.

The diagnosis of carcinoma of the colon and FAP was made at necropsy. Of the two sibs studied, the older was diagnosed as suffering from FAP and underwent colectomy in 1981. Diagnosis of FAP was later confirmed in the younger sib who had a colectomy and iliorectal anastomosis performed in 1987.

The two brothers, now aged 33 and 31, live semi-independently and function at a mild to moderately retarded level. They show similar dysmorphic features (fig 1) consisting of a mildly disproportionate facies, carp mouth, large nose, high forehead with early balding, macrognathia, and a generally large head. Both have a high arched palate and exhibit some of the dental anomalies (pointed canines and central incisors) and subcutaneous lesions associated with Gardner's syndrome.

Early IQ tests using a Binet LM scale were 50 and 51 respectively. An EEG showed evidence of bilateral temporal lobe epilepsy in the older brother and a non-specific abnormality in the younger brother. Cytogenetic examination of 72 hour methotrexate synchronised peripheral blood cultures showed evidence of an interstitial deletion involving bands 5q13→q15 or 5q15→q22 in each subject (fig 2).

This deletion is similar to that noted by Herrera *et al.*[1] Although bands 5q14 and 5q21 are cytogenetically indistinguishable, we consider that the presence of this deletion in these two sibs lends further weight to the proposal that the locus of the gene for FAP is probably close to the area 5q21 to 5q22. The dental anomalies and the subcutaneous lesions are characteristic of Gardner's syndrome as distinct from FAP, though the conditions may be allelic.

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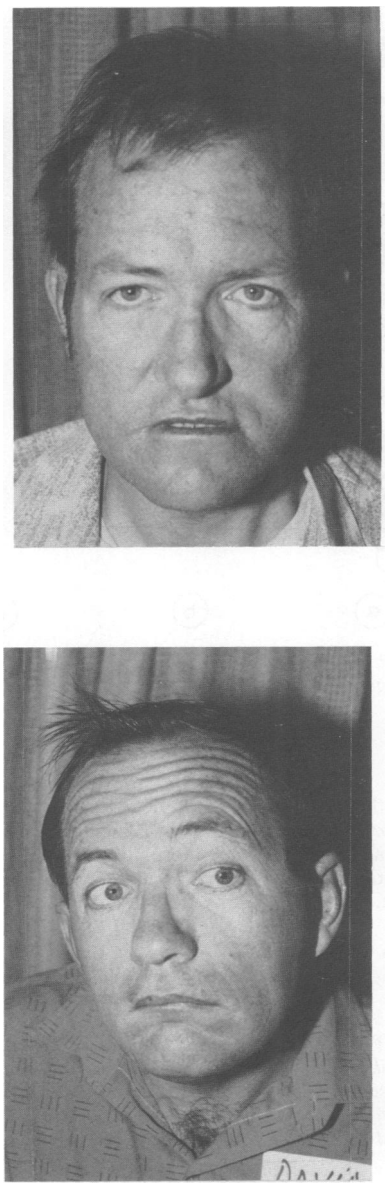

FIG 1 *Brothers with FAP and 5q deletion.*

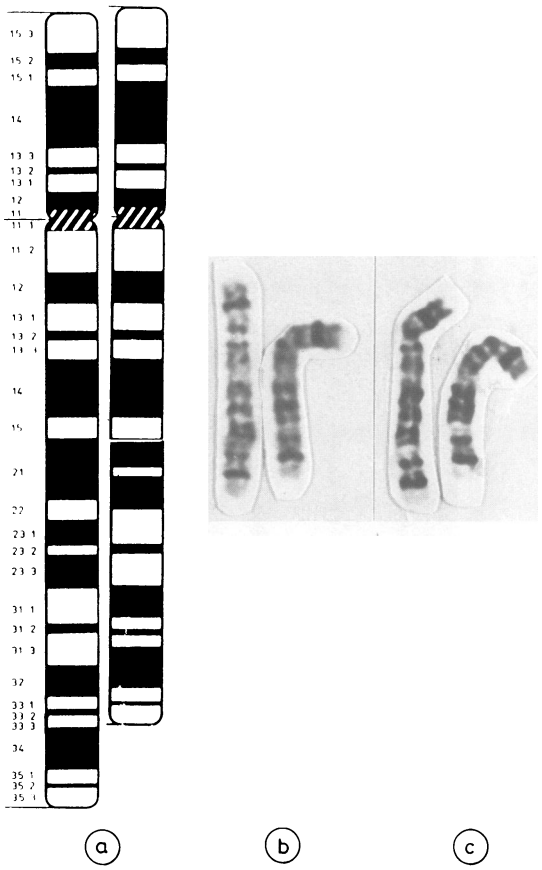

FIG 2 *Chromosomes 5: (a) ideogram, (b) older brother, (c) younger brother. The deleted chromosome is on the right.*

This is the first description of the deletion segregating in two, probably three if the mother is included, members of a family, all of whom are dysmorphic and mildly retarded. These findings are suggestive of another contiguous gene syndrome.

K A HOCKEY*, M T MULCAHY†, P MONTGOMERY‡, AND S LEVITT§
**Genetic Services, Health Department of WA, King Edward Memorial Hospital for Women, Subiaco, WA 6008; †Combined Clinical Cytogenetics Unit, State Health Laboratory Services, QE II Medical Centre, Nedlands, WA 6009; ‡Authority for Intellectually Handicapped Persons, PO Box 441, West Perth, WA 6005; and §Consultant Surgeon, QE II Medical Centre, Nedlands, WA 6009, Australia.*

References

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Correspondence and requests for reprints to Dr A Hockey, Genetic Services, King Edward Memorial Hospital, Bagot Road, Subiaco, Western Australia 6008.

Obesity and abnormal behaviour associated with interstitial deletion of chromosome 18 (q12.2q21.1)

A seven year old male with an interstitial deletion of band q12.3 of chromosome 18 is reported. Medical problems include developmental delay, obesity with onset at the age

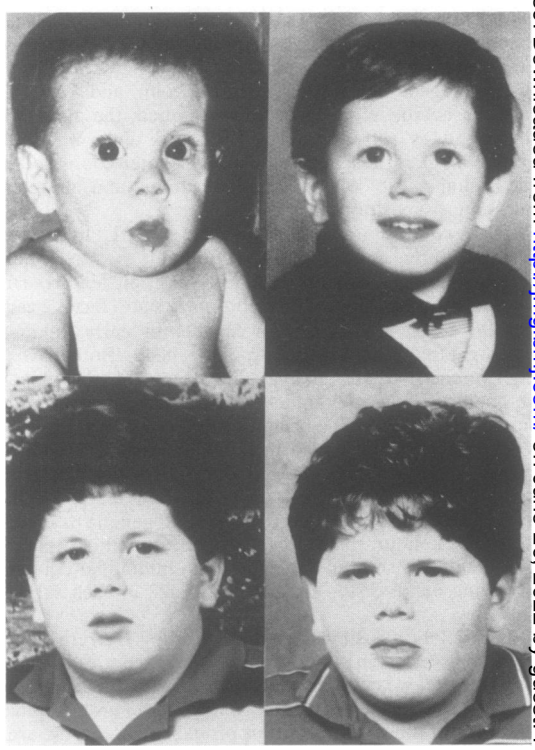

FIG 1 *The proband at ages eight months, three years, five years, and six and a half years.*

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