Short report

Gardner's syndrome in a man with an interstitial deletion of 5q15.1


3 Hockey A. Genetic studies in adoption. MD thesis, University of Western Australia, 1977.

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

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 Levi§

*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

1 Herrera L, Kakati S, Gibas L, Pietrzak E, Sandberg AA.

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

FIG 2 Chromosomes 5: (a) ideogram, (b) older brother, (c) younger brother. The deleted chromosome is on the right.
of three, and behavioural abnormalities such as perseveration, patterned movements, easy distractability, and autistic tendencies.

The proband (fig 1), born at term, was the 4600 g, 51 cm product of a first pregnancy of a 32 year old primigravida. The father was 34 years old. The pregnancy was complicated by gestational diabetes and, during the first trimester, exposure to doxylamine-dicyclomine HCI-pyridoxine HCI (Bendectin®). The family history was unremarkable. The patient had hypotonia and delayed developmental milestones and speech. A borderline abnormal EEG was documented after spells of staring or left arm posturing with abnormal vocalisation. He had frequent hyperactivity, aggressiveness, and irritability with easy frustration. Wilson et al² reported seven patients with the typical 18q— phenotype who had common deletion of band 18q21. A male patient with deletion of band 18q12.1 or q12.3 had a different phenotype with epicanthic folds, prominent upper lip, micrognathia, cryptorchidism, left simian crease, weight below the 5th centile, seizures, and a developmental quotient of 30 to 40. While Wilson et al² also favoured the interpretation of del(18)(q12.2;q21.1) in their patient, the different phenotype in our case argues against identical breakpoints. Two patients with a larger interstitial deletion of 18q (q11.2;q21.3) had some characteristics of the 18q— syndrome, but shared obesity and deletion of band q12.3 with our proband.³ Further patient studies are needed to refine the phenotypic/cytogenetic correlations on 18q, since this is a region where psychological testing and high resolution cytogenetics may identify several loci relevant to cognitive function.

Partial trisomy 16q secondary to a maternal 9;16 translocation

A six month old dysmorphic female was found to have partial trisomy for the long arm of chromosome 16 owing to a maternal translocation: 46,XX,t(9;16)(p24;q21). The

Correspondence and requests for reprints to Dr G N Wilson, Department of Pediatrics, University of Texas Southwestern, Medical Center of Dallas, 5323 Harry Hines Boulevard, Dallas, Texas 75235, USA.

References


Received for publication 1 March 1988.
Revised version accepted for publication 28 March 1988.