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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.

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.

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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 HCl-pyridoxine HCl (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 and large hand and foot edema, obesity beginning at the age of three years without compulsive eating or aberrant choice of foods. Behavioural abnormalities included perseveration, patterned movements, easy distractability, and autistic tendencies, poor concentration, and an IQ of 50 which necessitated special educational facilities. Physical examination (aged seven) showed a height of 134 cm (above the 97th centile; at the 50th centile for nine and a half years), a weight of 59·5 kg (far above the 97th centile; 50th centile for age 15½ years), a head circumference of 56 cm (50th centile for 17 years), normal craniofacial morphology, bilateral myringotomy tubes in place, a normal phallus (6 cm, 60th centile) and testes, dermatoglyphic patterns of RUUUU on the left hand and UUUUU on the right, and mild hypotonia. The patient was withdrawn but responsive to commands and interacted with the examiner.

Prometaphase karyotyping with Giemsa-trypsin (fig 2) was performed on peripheral blood lymphocytes using standard methods. The absence of a dark band at q12.1 or q12.3 was noted in one of the 18 homologues of the proband. The normal distance from the centromere to the remaining dark band suggests that band q12.3 was deleted. Parental karyotypes were normal.

Patients with the 18q syndrome have characteristic facial dysmorphism and behavioural abnormalities such as hyperactivity, aggressiveness, and irritability with easy frustration. Wilson et al. 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, right hand and foot edema, and a developmental quotient of 30 to 40. While Wilson et al. also favoured the interpretation of del(18)(q12.2q21.1) in their patient, the different phenotype in our case argues against identical breakpoints. Two patients with a larger interstitial deletion of 18q (q11.2q21.3) had some characteristics of the 18q− syndrome, but shared obesity and deletion of band q12.3 with our proband. 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.

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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