Correspondence

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Angelman’s syndrome and 15q11–13 deletions

Sir,

In the February issue of the journal we read with interest the finding of the paper by Pembrey et al. After their initial finding of this type of deletion in two patients they were able to re-examine 10 patients who have “a secure diagnosis of Angelman’s syndrome”. In fourteen of these an interstitial del(15) (q11q13) was found. In one other case a pericentric inversion inv(15)(p11q13) was found, which was also present in his normal mother. Two of the patients with an undetectable chromosomal deletion had an affected, or possibly affected, sib.

During the past few months we have had the opportunity to perform high resolution G banded chromosome studies in 10 patients (nine girls and one boy) who had been diagnosed as having Angelman’s syndrome in the past. Eight were isolated cases and two were sisters. In six of the 10 patients the presence of a del(15)(q11q13) was detected. As in the experience of Pembrey et al, a deletion was not seen in the two sibs with this MCA/MR syndrome.

The present data confirm that with the available cytogenetic techniques a 15q11–13 deletion can be visualised in at least half of the patients with Angelman’s syndrome. The data also support the conclusion of Pembrey et al that de novo visible deletions and autosomal recessive cases combined give an overall sib recurrence risk of less than 25%.

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Reference


Anencephaly: a vanishing problem in Bedouins?

Sir,

In Kuwait, preliminary data from the congenital malformations register showed a dramatic decline in the birth prevalence of anencephaly during the last two decades among Arabs with Bedouin ancestors. In a classification based on epidemiological data, Kuwait was put in the high prevalence group on the basis of a 1968 study, which showed a prevalence of 3/2/1000 total births among the 4625 deliveries in the Maternity Hospital. During 1983, 48 cases of anencephaly were ascertained among the 36138 consecutive births in this hospital and the three new regional general hospitals (Jahra, Farwania, and Adan). This study showed (1) a marked decrease in the prevalence of anencephaly to 1.33/1000 (less than half the previous figure); (2) a marked geographical variation among the three regional hospitals; and (3) the highest prevalence figures (2.05/1000) among deliveries in Jahra hospital. (This hospital serves an Arab population of 300 000 with 80% belonging to the Bedouin community, who maintain their own traditions including dietary habits.) (4) The lowest prevalence (0.85/1000) was among deliveries in Farwania hospital. This hospital serves a mixed Arab and non-Arab population of 400 000 with only 15% belonging to the Bedouin community.

We have established the Jahra Regional Liaison-Community Genetics Programme during the last decade and in 1984 introduced a mass educational dietary programme to the Bedouin women. The main objective of this is the prevention of neural tube defects (NTDs). The programme trained 42 primary health care doctors, community nurses, and social workers who were the active ‘genetic associates’. We explained to the Bedouins the importance of adding fresh vegetables and fruit to their traditional food as a means of decreasing the prevalence of NTDs. In simple language, we showed them the definite relationship between maternal diet and prevalence of NTDs and how either dietetic counselling or pre/conceptional supplementation with physiological doses of folic acid or multivitamins led to a marked diminution of the recurrence risk of NTDs. The response of the Bedouin to this