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LETTERS TO THE EDITOR

Another example favouring the location of BPES at 3q2

Recently in this Journal, de Die-Smulders *et al*[1] reported a father and son both affected with BPES (blepharophimosis, ptosis, epicanthus inversus syndrome) and carrying a balanced 3;11(q21;q23) translocation in the absence of mental retardation. These findings provided new evidence for the location of the BPES gene at 3q2, as previously suggested by Fukushima *et al*,[2] who reported an infant carrying a de novo translocation involving the 3q23 region.

We report on a young girl with mild mental retardation, blepharophimosis, ptosis, telecanthus, epicanthus inversus, and a de novo apparently balanced 3;8(q23;p21.1) translocation (fig 1). The patient is microcephalic and mildly mentally handicapped with a few other clinical features such as an arched palate, ears of normal size but low set and abnormally shaped with incompletely folded helices, short neck, bilateral limited elbow pronation and limited thumb adduction, and mild hypotonia. Cytogenetic analysis of the parents' karyotypes indicated a paternal origin of the abnormal chromosome 3. No cell line is available from this patient at present.

Blepharophimosis, microcephaly, and psychomotor developmental delay are common findings among the rare published cases of 3q2 interstitial deletions.[3] In patients with BPES, intellectual impairment is only occasional and usually mild.[4] It is possible that in our mildly retarded patient with an apparent balanced translocation a submicroscopic deletion may be present. Cytogenetic studies are recommended in BPES, especially if mental retardation or other dysmorphic features are associated. It seems that BPES, as Smith *et al*[5] have suggested, is emerging as another example of a contiguous gene syndrome. Future endocrinological follow up of our patient regarding ovarian function is indicated.

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A new approach to prenatal cystic fibrosis carrier screening

Two approaches to prenatal cystic fibrosis screening have been discussed: stepwise and couple screening.[1,2] Mennie *et al*[1] recently found that carriers detected by stepwise screening experienced significant levels of stress (detected by a general health questionnaire, GHQ[3]). Stress disappeared after the male parner's test was reported as 'normal'. Couple screening has been said to be superior to stepwise screening because carriers whose partner's test result is negative are not made anxious while awaiting their partner's result, nor by the knowledge that their risk is higher than it was before they were tested (pretest risk of having affected child 1/2500, post partner test risk 1/600). However, it does not allow relatives of identified carriers to be informed of their increased carrier risk, nor does it alert detected carriers of their risk with new partners.[4]

We propose an alternative form of couple screening (disclosure couple screening) that circumvents these disadvantages: carrier testing is only performed if a DNA sample is provided by each partner but carrier test results are fully disclosed to the couple. If one partner is found to be a carrier the other's sample is immediately available for analysis, therefore both partners' results can be imparted simultaneously, allowing couples at low risk of having an affected child to be counselled about their carrier status. Either or both samples could be tested as long as patients were fully informed which samples were tested.

We believe that disclosure couple prenatal screening may decrease test associated anxiety without altering the uptake rate of screening. It may be that an informed carrier woman will be more likely to assign paternity correctly than one who is unaware of her carrier status. As with any genetic test, good pretest counselling is essential. Full evaluation is necessary before any method of screening becomes routine practice.

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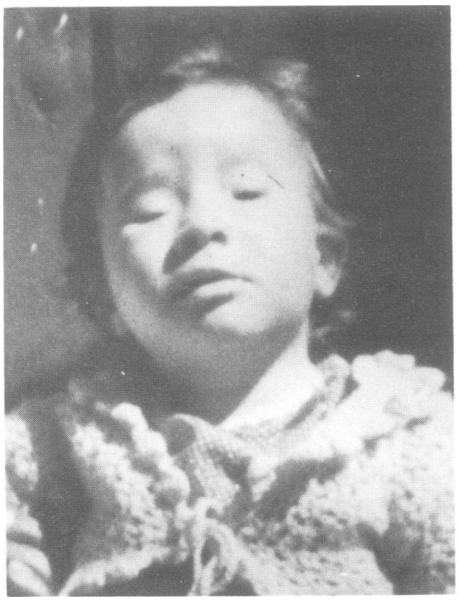

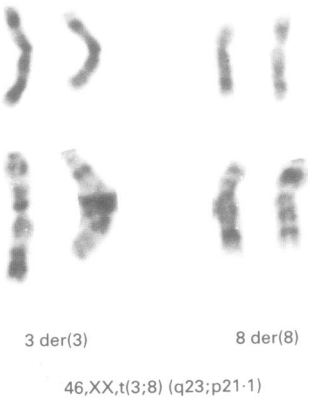

The proband at 1 year 10 months and two partial GTG karyotypes showing her balanced translocation involving chromosomes 3 and 8.