Another example favouring the location of BPES at 3q2

Recently in this Journal, de Die-Smulders et al. 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., 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 hyponia. 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. In patients with BPES, intellectual impairment is only occasional and usually mild. It is possible that in our mildly retarded patient with an apparently 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. 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. Recently, Smith et al. found that carriers detected by stepwise screening experienced significant levels of stress (detected by a general health questionnaire, GHQ). Stress disappeared after the male partner’s test was reported as ‘normal’.

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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.