Journal of

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Some notes on nomenclature can be found in *J Med Genet* 1991; **28**:72.

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Nilbert *et al*[3] reported a uterine leiomyosarcoma with a t(8;13). In addition, several uterine leiomyomas with structural or numerical abnormalities of chromosome 8 were reported by Mark *et al*[4] and Teyssier and Ferre.[5] Especially interesting in relation to the case under discussion is the latter authors' report of trisomy 8 in another gastrointestinal smooth muscle tumour, an oesophageal leiomyoma.

Thus, the abnormalities in chromosome 8 in smooth muscle tumours described so far involve both numerical and structural abnormalities and concern both benign and malignant tumours. We think that these data give a different perspective to the discussion of the case reported by Lessick *et al*.[1]

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1 Lessick M, Israel J, Szego K, Wong P. Leiomyosarcoma in a patient with trisomy 8 mosaicism. *J Med Genet* 1990;27:643–4.
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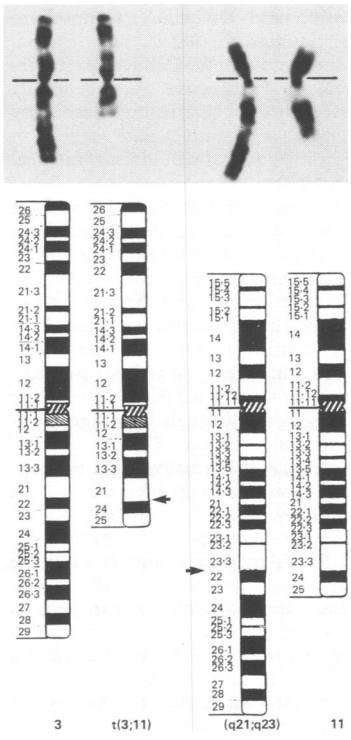
4 Mark J, Havel G, Grepp C, Dahlenfors R, Wedell B. Chromosomal patterns in human benign uterine leiomyomas. *Cancer Genet Cytogenet* 1990;44:1–13.
5 Teyssier JR, Ferre D. Frequent clonal chromosomal changes in human non-malignant tumors. *Int J Cancer* 1989;44:828–32.

Further evidence for the location of the BPES gene at 3q2

We read the paper of Smith *et al*[1] in this journal with interest. They suggested that blepharophimosis plus ovarian failure is a likely candidate for a contiguous gene syndrome, and recommended cytogenetic investigation of all cases of blepharophimosis, ptosis, epicanthus inversus syndrome (BPES). We would like to report a family with autosomal dominant BPES syndrome and a chromosomal abnormality. A father and his 6 month old son were referred for genetic counselling. Both showed the typical signs of BPES: blepharophimosis, ptosis, telecanthus, and epicanthus inversus. The father had no other dysmorphic features and was of normal intelligence. The son had a small nose with anteverted nostrils and cup shaped ears. His height, length, and head circumference were in the normal range and his mental development was normal. The father had two sons from a previous marriage who had the same eye anomalies. Unfortunately, they were not available for further investigations. Chromosomal examination on cultured lymphocytes of the father and son showed an apparently balanced translocation between the long arms of chromosomes 3 and 11, with respective breakpoints at 3q21 and 11q23. The karyotype was 46,XY,t(3;11)(q21;q23) (figure).

Recently, Fukishima *et al*[2] reported a newborn infant with BPES and a de novo balanced 3q23;4p15 reciprocal translocation. These findings strongly indicate that the gene for BPES is located in the 3q2 region. Furthermore, blepharophimosis, ptosis, and microphthalmia are consistent features in patients with an interstitial deletion of band 3q2,[3] reinforcing the location of the BPES gene at 3q2.



Karyotype of the proband.

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1 Smith A, Fraser IS, Shearman RP, Russell P. Blepharophimosis plus ovarian failure: a likely candidate for a contiguous gene syndrome. *J Med Genet* 1989;26:434–8.
2 Fukushima Y, Wakui K, Nishida T, Ueoka Y. Blepharophimosis syndrome and de novo balanced autosomal translocation [46,XY,t(3;4)(q23;p15.2)]: possible localization of blepharophimosis syndrome to 3q23. *Am J Hum Genet* 1990;47:29A.
3 Alvarado M, Bocian M, Walker AP. Interstitial deletion of the long arm of chromosome 3: case report, review, and definition of a phenotype. *Am J Med Genet* 1987;27:781–6.

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Pathology of the Human Embryo and Previable Fetus. An Atlas. D K Kalousek, N Fitch, B A Paradice. (Pp 230; DM 248.) Berlin: Springer-Verlag, 1990.

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