Interstitial deletion of chromosome band 13q14 associated with squamous cell carcinoma

The proband is a 58 year old male Negro residing in Zambia. He presented at his local hospital with oedema of the right leg and an ulcerated mass overlying the tibial crest, which had been present for about 4 months. Physical examination showed bilateral tender enlarged inguinal lymph nodes, but no generalised lymphadenopathy. The presence of haemorrhagic nodules on the right thigh and lower leg, together with local oedema, suggested a diagnosis of Kaposi's sarcoma (which is endemic in Zambia). However, histological examination of biopsy material from skin nodules and an inguinal lymph node showed moderately differentiated squamous cell carcinoma.

Peripheral blood and lymph node samples were sent to England for investigation of cytomegalovirus (CMV) and human T cell leukaemia virus (HTLV) involvement when the patient was suspected of having Kaposi's sarcoma. Antibodies to CMV were assayed by an enzyme linked immunosorbent assay. Serum was found to contain CMV specific IgG, but no CMV specific IgM or IgA was detected. Antibodies to HTLV membrane antigen were assayed by a two step fluorescence test using two HTLV producer cell lines. The results showed no evidence of infection with HTLV.

Monolayer cell cultures were established from small explants of the affected lymph node tissue. The cultures showed fibroblastoid cell growth. Cytogenetic analysis revealed a small interstitial deletion from the long arm of a chromosome 13 in all cells examined, karyotype 46,XY,del(13)(pter→q14.11::q14.3→qter) (figure). Lym-

phocyte culture from peripheral blood showed an apparently normal constitutional karyotype.

Previous reports of interstitial deletion of the chromosome band 13q14 associated with malignancy have, to our knowledge, been confined to cases of retinoblastoma. Ophthalmological examination of the patient did not reveal any evidence of retinal tumours. Recent reports¹ suggest that retinoblastoma tumorigenesis results from the development of homozygosity for the mutant allele at the retinoblastoma (Rb-1) locus. However, cytogenetic analysis at the level of resolution available failed to reveal a deletion from the 'normal' chromosome 13 homologue, although it is possible that this may be present submicroscopically.

The esterase D (EsD) locus has also been assigned to the chromosome band 13q14 and has been shown to be closely linked to the retinoblastoma locus.² Investigation of EsD expression in the cultured cells by use of starch gel electrophoresis showed a normal but weak EsD-1 pattern compared with control cells. This result is consistent with deletion of the EsD locus in the aberrant cell line, resulting in hemizygous expression.

It is of interest to note that in our patient the deletion of band 13q14 has been detected in association with a malignant disorder other than retinoblastoma. These findings suggest that altered expression of a putative oncogene linked to the Rb-1 and EsD loci could be responsible for neoplastic transformation in tissues other than the retina.

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FIGURE GTG banded chromosome 13 homologues, demonstrating interstitial deletion of band 13q14 in cells from lymph node tissue.