Short report

Leiomyosarcoma in a patient with trisomy 8 mosaicism

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A male newborn was found to have trisomy 8 mosaicism (46,XY/47,XY,+8) with 27% trisomic cells in lymphocytes. Tissue culture of a gastric leiomyosarcoma from the proband at the age of 6 showed two clones, one with 47,XY,+8 and one with 47,XY,2q+,+8,+20 (figure). No cells with normal chromosomes were found. Leiomyosarcoma in trisomy 8 mosaicism has not been previously reported.

The proband was delivered at term by caesarian section after an uncomplicated pregnancy in a 20 year old gravida 1, para 1 woman. Birth weight was 3450 g, length 52.5 cm, and head circumference 34 cm. Both

parents are unrelated Afro-American. The family history was unremarkable. Birth abnormalities included a 2.5×3.7 cm omphalocele, a small ventricular septal defect, broad hands and feet with deep palmar and plantar creases, raised triradius in "i" position, hypoplastic nails, clinodactyly of the fifth fingers, and undercurving of the second to fifth toes. Chest and abdominal x rays, skeletal survey, cystourethrogram, IVP, and barium enema were normal.

From 6 weeks of age he was treated for seizures. At 6 years of age, he presented with fever, weakness, abdominal pain, and tarry diarrhoea followed by constipation. Upper GI and CT scan of the abdomen showed a large, non-obstructing, soft tissue tumour in the lesser curvature of the stomach. By light and EM studies the tumour was a leiomyosarcoma composed of spindle cells, large, irregular nuclei, an abundant eosinophilic background stroma, areas of thrombosed vessels, and a high mitotic index. Liver biopsy and bone scan showed no evidence of metastasis. On follow up at the age of 14, the proband had mild

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G banded karyotype from a tumour cell showing 47,XY,2q+,+8,+20.

mental retardation, a long face, and stiff hands with limitation of joint movement.

Trisomy 8 is a relatively rare disorder with significant dysmorphic features, mental retardation, and extreme phenotypic variability. Of the approximately 110 cases reported, the majority are mosaics and support a lack of correlation between the phenotype and percentage of trisomic cells.¹ Our patient has many of the features which make this syndrome a recognised clinical entity.¹ The leiomyosarcoma in this patient may have been a coincidental finding. However, it is known that malignancies occur with increased frequency in certain constitutional chromosomal syndromes, such as trisomy 21, 13q-, and 11p-. The occurrence of trisomy 8 in certain leukaemic cells is also well known.¹ Further, there has been at least one case of leukaemia reported in a patient with constitutional mosaic trisomy 8.²

Leiomyosarcomas are rare neoplasms and only a few have been analysed cytogenetically. In a review of 13 leiomyosarcomas from various anatomical sites,³

neither numerical nor structural abnormalities involving chromosome 8 have been observed. Boghosian *et al*³ classified leiomyosarcomas into three cytogenetic subtypes: (a) a pseudodiploid chromosome number associated with simple reciprocal translocations; (b) a hypodiploid chromosome number ranging from 41 to 43 and monosomies of chromosomes 18, 22, and the short arm of 1; and (c) tumours with heterogeneous karyotypic findings. The finding of trisomy 8 cells in the leiomyosarcoma of our patient appears to be unique. Clinicians should be alert to the possibility of an increased risk of malignancy in trisomy 8 mosaicism.

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- 2 Gafter U, Shabtai F, Kahn Y, Halbrecht I, Djaldeiti M. Aplastic anemia followed by leukemia in congenital trisomy 8 mosaicism. *Clin Genet* 1976;9:134-42.
- 3 Boghosian L, Cin PD, Turc-Carel C, *et al*. Three possible cytogenetic subgroups of leiomyosarcoma. *Cancer Genet Cytogenet* 1989;43:39-49.