

detected 95% heterozygosity in females (*Nucleic Acids Res* 1987;15:9616). We have investigated the potential use of this probe to differentiate between active and inactive X chromosomes through the methylation patterns revealed by the restriction enzyme *MspI* (recognition site CCGG) and its methylation sensitive isoschizomer *HpaII*. Experiments were carried out using DNA prepared from blood and from cell lines with defined patterns of X inactivation. Preliminary results suggest that the CCGG sites flanking the *DXS255* locus are extensively methylated on active X chromosomes and unmethylated on inactive X chromosomes. The high degree of heterozygosity detected, combined with this differential methylation pattern, indicate that this simple system should be applicable to many situations where assessment of X inactivation status is required (for example, imprinting, females with X linked disorders, analysis of clonal development in tumours).

Reduced activity of enzymes bound to the microvillar membrane fraction of amniotic fluid from pregnancies with cystic fibrosis

D A AITKEN, J A H MAATOUK, G W GRAHAM, E GRACEY, AND J M CONNOR

Duncan Guthrie Institute of Medical Genetics, Yorkhill, Glasgow.

Amniotic fluid (AF) microvillar enzyme activity is known to be reduced in second trimester pregnancies where the fetus has cystic fibrosis (CF). An alternative approach to assaying the soluble component of these enzymes is to isolate the fragments of microvilli present in the AF supernatant for direct analysis (Potier *et al*, *Prenat Diagn* 1986;6:429-36). We report here the results of a preliminary investigation of the enzyme activity associated with the microvillar membrane fraction of AF supernatant from three pregnancies in which the fetus was terminated with

CF, and in a series of normal controls. The yield of microvilli from the CF samples was found to be in the same range as that from the control group, but the specific activities of the microvillar enzymes maltase, gammaglutamyltranspeptidase, and intestinal alkaline phosphatase were significantly lower ($p < 0.001$) in the microvilli preparations from the CF cases. These results suggest that enzyme analysis of purified microvillar membrane fragments may be a more discriminating test for the prenatal diagnosis of CF than assay of these enzymes in AF supernatant.

Wide spectrum of symptoms in Stickler's syndrome

M TOLAROVÁ J ŠPINDRICH, L BAŘINKA, AND J SAMOHÝL
Institute of Experimental Medicine, Lid milicí 61,12000 Prague 2, Czechoslovakia.

Stickler's syndrome (hereditary progressive arthropathopathy) is a serious autosomal dominant condition, which is increasingly diagnosed in genetic counselling clinics. Owing to variation in severity and expressivity, this condition often presents diagnostic problems. The main features are progressive myopia and choroidoretinal abnormalities in childhood, sometimes leading to retinal detachment or glaucoma, and Robin anomalad and enlargement of large joints, particularly of the wrist, knees and ankles, which may be present at birth. In middle age repeated episodes of acute arthritis may occur and precede degenerative arthropathy, considerably impairing the physical activity of the patient. Inconsistent features include conductive deafness, cleft palate, structural abnormalities of the vertebrae, and mental retardation. Ten families with 16 cases have been examined and clinical symptoms, dysmorphological features, and radiology, characteristic for different ages, are pointed out.

Correction

In the abstracts of the November 1987 meeting of the Clinical Genetics Society (*J Med Genet* 1988;25:274-83), an error occurred in the abstract by Redha *et al* on page 278. The penultimate sentence should have read "In the 45,X cases loss of the paternal homologue was observed in all cases".