would lead to failure of suppression of HOXB-8 expression in the anterior limb field. Ectopic expression of HOXB-8 would then specify cells in the anterior limb field as ZPA, leading to partial or complete duplication of gnathopod, autopod, and stylopod.

There are no reports of human (humers/fermus) duplication in Laurin-Sandrow syndrome, which would argue against the above hypothesis. To my knowledge, however, no author has to date thoroughly examined the humeri and femora of Laurin-Sandrow patients for any evidence of abnormal patterning. Further delineation of the Laurin-Sandrow phenotype, especially with regard to more proximal skeletal elements, would be most helpful in the search for candidate genes for this intriguing syndrome.

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12 Skovbjerg F, Schröder H, Skov P, et al. The older boy was diagnosed as having Klinefelter syndrome at the age of 5 years. He had reduced left elbow supination and radioulnar synostosis. Diagnosis was made in the younger sib at the age 3 years because of undescended testes and a "willowy appearance" similar to his older affected brother. Both had heights approximately on the 50th centile, weights on the 3rd centile, and reduced upper to lower body segment ratios of 0.87 and 0.85. The older sib is at Tanner stage 1 of sexual development at 11 years. Both are otherwise normal on examination, of normal intellectural ability, and neither has any behavioural problems.

Both families were healthy, normal on examination, and aged 24 years at the time of birth of their first child with Klinefelter syndrome. The couple had no difficulty in conceiving and no recognised pregnancy losses. They are first cousins from the Punjab and are themselves products of first cousin marriages (fig 1). There are no other family members known to have chromosomal anomalies, infertility, or frequent miscarriages.

Chromosome analysis of peripheral blood lymphocytes was performed on the nuclear family members, 50 cells being examined in each case. The brothers with Klinefelter syndrome were non-mosaic 47,XXY, other family members having normal results. X chromosome haplotype analysis was performed using an Applied Biosystems 373 DNA Sequencer and Genescan 672 software. The microsatellite present in intron 13 of the factor VIII gene at Xq28 was informative, but nine microsatellites from the ydroxypherin gene were uninformative. The results are shown in fig 1; both boys with Klinefelter syndrome apparently inherited an X chromosome from their father.

These sibs with Klinefelter syndrome had typical clinical features, non-mosaic 47,XXX, and their additional X chromosome was the result of a paternal meiosis I error. Possible explanations for this situation are that the father is a hypogonadal male mosaic which seems unlikely, or that the father has an autosomal recessive disorder affecting meiosis. We have been unable to investigate the family further by single sperm analysis and because no other relative is known to have had a child with aneuploidy.

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A study of brothers with Klinefelter syndrome.

C G Woods, J Noble and A R Falconer

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