

## CORRESPONDENCE

### Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1: exclusion of aggrecan as a candidate gene

We have conducted further investigation of the locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1.

On the basis of linkage data, we previously reported a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1 that excluded the aggrecan gene. The map position for aggrecan that we reported differed from available sequence data at that time and hence we concluded that "the inconsistencies between the linkage and sequence data clearly need to be resolved before the targeted analysis of other genes within the linked region can be systematically performed".<sup>1</sup>

We have continued our studies of this family and have now identified a genotype error in one of the members of the above

family: individual III-1 in the pedigree in fig 2 for SNP AGC1.2 which was given as 1/1 and should be 1/2. This error was found following an alteration of the primers used to sequence the exon that contained the SNP and using a new DNA sample for that individual. The consequence of this misgenotyping is that the map position for aggrecan now falls within the linked region in this family and so remains a candidate gene for this disorder.

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## Reference

- 1 Eyre S, Roby P, Wolstencroft K, Spreckley K, Aspinwall R, Bayoumi R, Al-Gazali L, Ramesar R, Beighton P, Wallis G. Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26. 1: exclusion of aggrecan as a candidate gene. *J Med Genet* 2002;39:634-638.