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”.

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.

s eyre
arthritis research campaign epidemiology research unit, the university of manchester, manchester, uk

p rob
renovo ltd, manchester incubator building, manchester, uk

k wolstencroft
bioinformatics unit, the university of manchester, manchester, uk

k spreckley, r aspinwall
arthritis research campaign epidemiology research unit, the university of manchester, manchester, uk

r bayoumi
department of biochemistry, sultan qaboos university, muscat, sultanate of oman

l al-gazali
department of paediatrics, united arab emirates university, al ain, united arab emirates

r ramesar, p beighton
department of human genetics, university of cape town, cape town, south africa

l gleghorn, g wallis
the wellcome trust centre for cell-matrix research, the university of manchester, manchester, uk

correspondence to: g wallis, the wellcome trust centre for cell-matrix research, the university of manchester, manchester, uk; gillian.a.wallis@manchester.ac.uk

current investigators: l gleghorn, g wallis.

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reference
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