Refined mapping of the gene for otopalatodigital syndrome type I

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Electronic Letter

The gene for OPD-I has been mapped to the Xq27-28 region by linkage analyses in two families. Hoar et al.7 localised the OPD-I gene to a region distal to DXS100 on Xq25, with a maximum lod score of 1.99 at θ=0 for DXS100 and DXS1108 was shared in common by two boys with the typical OPD-I phenotype (cases IV.1 and IV.2) and two obligate carrier females with a mild or overt OPD-I phenotype (cases II.3 and III.2). This particular haplotype was absent in four normal males (cases II.1, II.2, II.4, and III.1) and in one normal female who should be free from a mutant OPD-I gene (case I.2). Two point linkage analysis was performed with the program MLINK of the LINKAGE package version 5.10 under the assumption of 100% penetrance in affected males and 80% penetrance in carrier females and showed a maximum lod score of 0.90 at θ=0 for DXS1177, DXS15, BGN, DXS1073, and DXS8087.

The X inactivation pattern was examined in the female family members by previously described methods.8,9 Analysis of the methylation pattern of the PGK1 gene indicated skewed X inactivation in case III.2. The results of the X inactivation pattern, though examined for leucocytes, were consistent with random expression of the mutant OPD-I allele in case II.3 with a mild OPD-I phenotype and preferential expression of the mutant OPD-I allele in case III.2 with an overt OPD-I phenotype.

Summary

In summary, the present study suggests that the OPD-I critical region is further narrowed down from the −12 Mb region distal to DXS539 to the −6 Mb region between DXS8011 and DXS1108, with a combined maximum lod score of 4.09. Further studies will permit a better localisation of the gene for OPD-I.

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References


Abbreviations: OPD-I, otopalatodigital syndrome type I

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Figure 1  Family with otopalatodigital syndrome type I (OPD-I). Black squares indicate males with the typical OPD-I phenotype, circles with a dot depict obligate carrier females with a mild or overt OPD-I phenotype, and white squares and circles represent clinically normal subjects. The loci examined at Xq26-28 are shown at the bottom right. DXYS154 and DXYS225 lie in the long arm pseudoautosomal region, and the remaining 16 loci reside in the X differential region. The alleles are arbitrary, indicated by Arabic numbers according to their sizes. The region between DXS8011 and DXS1108 is shared by affected males and females and is absent in clinically normal subjects examined.

