Refined mapping of the gene for otopalatodigital syndrome type I

T Kosho, T Uemura, M Tanimura, H Ohashi, K Muroya, T Ogata

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.

CASE REPORTS

The family pedigree is shown in fig 1. Case I.1 was dead and, allegedly, had had clinical features compatible with OPD-I. Case II.3 exhibited mild but definite supraorbital ridges and bilateral short first toes. Case III.2 showed overt supraorbital ridges, bilateral short first toes, and hearing loss. Cases IV.1 and IV.2 had typical OPD-I features such as supraorbital ridges, flat nasal bridge, hypertelorism, downward-slanting palpebral fissures, thick and arched eyebrows, microtia, spatulate distal digits, and hearing loss. Cases IV.1 and IV.2 had random X inactivation with the ratio of inactivation between the two X chromosomes being 60%:40% and 71%:29%, respectively; however, it was not informative in case III.2 because of lack of heterozygosity. 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.

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.


Electronic letter


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