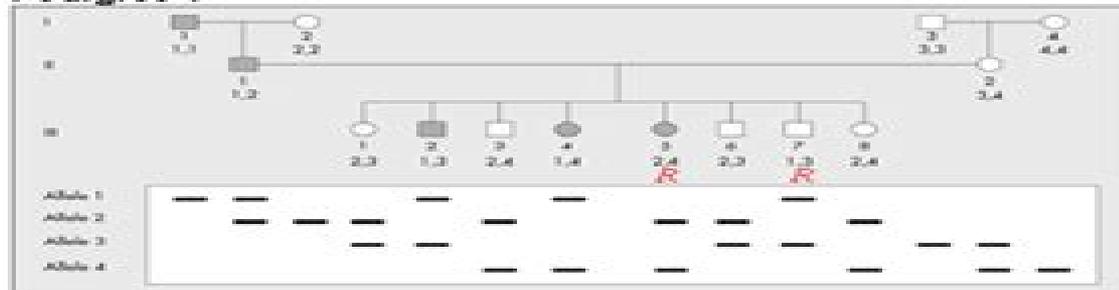


Gene Mapping 1 Tutorial

1) All family members of Pedigree 1 have been genotyped for a four-allele microsatellite marker, as shown in the autoradiogram below the pedigree.

Pedigree 1



R = Recombinant

Determine linkage phase for the disease and marker locus in the affected male in generation II and estimate the recombination frequency based on the data you have from the offspring in generation III.

In general, linkage phase refers to the knowledge of how alleles at two or more loci are arranged on the same chromosome (homologue) and whether they are inherited together. It helps determine how alleles are passed on together from each parent. In this informative family, linkage phase is straightforward - the disease allele D must be on the homologue that contains marker allele 1 in this male II-1 (D1/d2 haplotypes). In other words, allele D from the affected father (I-1) was passed on with marker allele 1 to his son, II-1.

Two recombinants so the recombination frequency is $2/8 = 0.25$

Based on the information you have (and without calculating the LOD score) is there any initial evidence for linkage? Explain your reasoning by assuming full penetrance.

Assuming 100% penetrance, we can say that yes, there is some evidence of linkage. We can observe that allele D is inherited from the affected father (II-1) to his affected offspring (III-2 and III-4), along with marker allele 1. Except for the two recombinants (III-5 and III-7), the remaining unaffected offspring (III-1, III-3, III-6, and III-8) have the expected allele d paired with marker allele 2, as would be predicted under the assumption of linkage.

This provides initial evidence for linkage, further supported by a recombination frequency smaller than 0.5. However, it's crucial to remember that this is preliminary evidence from a single family. To confirm linkage, additional families should be studied, and a comprehensive linkage analysis using a larger number of marker loci should be conducted under different hypotheses of θ . This will help determine whether there is strong evidence of linkage ($LOD > 3$) or not.

Gene Linkage And Mapping Power Notes Answers

Philip R. Muskin M.D.



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