



**FIGURE 26.2.** The rate of recombination between a marker and a disease locus can be estimated from pedigrees. (A) Two siblings are affected by a disease caused by a rare autosomal recessive allele (*shaded symbols*). The parents must both be heterozygous for the disease allele. The family is genotyped for a marker (A, a); the father and one daughter are heterozygous (*circles*: females; *squares*: males). (B) The probability that this pattern would be observed as a function of the recombination rate between the marker and the disease locus. This is known as the **likelihood** of the recombination rate. For this very small set of data, most likely there is complete linkage ( $r = 0$ , at *left*), with the marker A linked to the disease allele; but it is not much less likely that there is no linkage ( $r = 0.5$ , *right*). (C) With more data (here 200 families of the same size as in A), a better estimate can be made: The most likely estimate is that  $r = 0.13$  (*peak of graph*). The data were simulated with  $r = 0.1$ , so this is reasonably accurate. Likelihood is scaled relative to the maximum, at 1. See Chapter 14 (pp. 399–402) for further details of this method in the context of QTL mapping.