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LOD Score

By Levi Clancy for Student Reader on Monday 17th November, 2008 *updated 16th September, 2017*



The **LOD score** (aka *Z*) gives an estimation of how closely two loci are linked (for example, a marker locus and a disease locus) *and* quantitates sample size (data reliability).

A LOD score less than 2.0 means the two loci are not linked; a LOD score between 2.0 and 3.0 is inconclusive; and a LOD score greater than 3.0 strongly indicates linkage. Lod scores are always reported in association with the *recombination frequency* (θ , theta), measured in Morgans, which describes linkage without accounting for sample size. Please review linkage analysis if any of these concepts are unclear.

Step 1	Look at nothing more than affected/unaffected individuals and determine the mode of inheritance.
Step	Cross out individuals who cannot be identified as recombinants or

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nonrecombinants (uninformative individuals).

- 1. Persons whose parents are not shown.
- 2. Individuals without at least one heterozygous parent.
- 3. Children are uninformative when the child and both parents have the same haplotype.
- 4. If the allele of interest is dominant, then children with a homozygous recessive parent.

Step

3

Determine whether each informative individual is recombinant or nonrecombinant. Considering an informative individual's haplotype, is it consistent with their parent's haplotype and the mode of inheritance?

- 1. Yes, consistent with parental haplotype \rightarrow Not Recombinant.
- 2. No, inconsistent with parental haplotype \rightarrow Recombinant.
- 3. Remember that even unaffected individuals can be recombinant!

Step

4

Step

5

Count how many recombinants and non-recombinants there are.

- 1. If you are provided a *recombination frequency* $(\theta$, aka *theta*) then go straight to the equation below.
- 2. If you are provided the gene-marker distance (measured in *centimorgans* or *cM*) then divide that value by 100 to calculate θ . For example, a marker 10cM from a gene yields a Θ of 10/100 = .10.
- 3. If you are supposed to calculate the recombination frequency

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 (θ) , then use the equation below with θ values of .001, .10, .15, .20, .25, .30, .35, .40., .45 and .50. The correct θ value will yield the highest LOD Score.

LOD score =
$$Z = log_{10} \left(\frac{\theta^{(\# \text{ OF RECOMBINANTS})} \cdot (1-\theta)^{(\# \text{ OF NONRECOMBINANTS})}}{(1/2)^{(\# \text{ OF RECOMBINANTS})}(1/2)^{(\# \text{ OF NONRECOMBINANTS})}} \right)$$