Bio393: Genetic Analysis **Name:**_____
Quiz #4 - Friday May 22, 2015

Question 1 (2 pts each):
Circle the correct answer(s).

(a) Which type of variant is easiest to map using family-based analyses?

1 - Rare variants with variable penetrance
2 - Common variants with variable penetrance
3 - Rare variants with high penetrance
4 - Common variants with high penetrance

3 and 4 are both acceptable answers. High penetrance being the key factor in this case.

(b) Which type of variant is easiest to map using population-wide analyses?

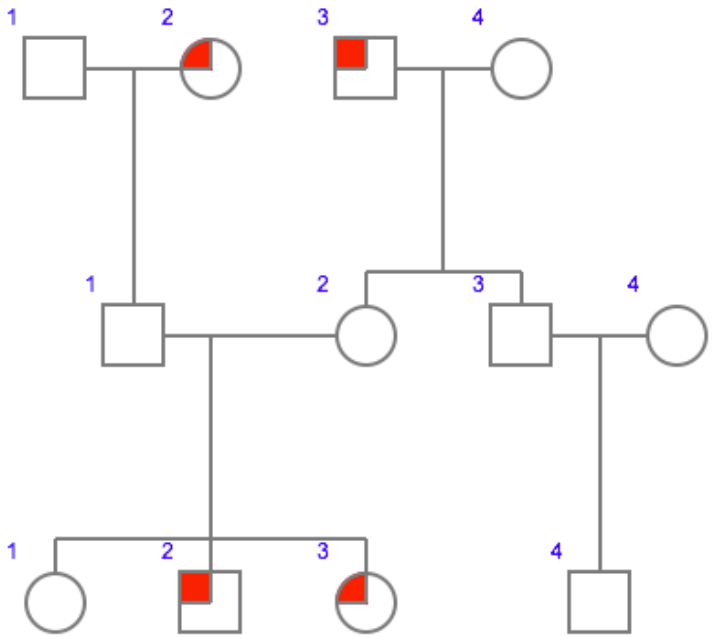
1 - Rare variants with variable penetrance
2 - Common variants with variable penetrance
3 - Rare variants with high penetrance
4 - Common variants with high penetrance

(c) How many individuals need to be sequenced to identify causal variants for Mendelian (single gene) diseases?

1 - Up to 10
2 - 1000
3 - More than 100,000

Of course, more sequenced individuals make the identification of genes even easier. Let's assume that money is a factor.

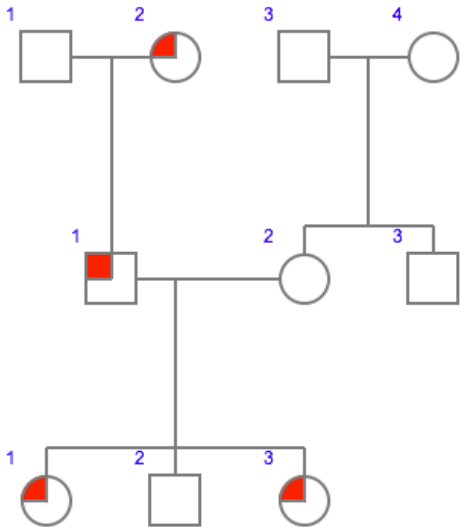
Question 2 (6 pts):
For the following two pedigrees, specify the mode of inheritance and explain your rationale. Indicate any ambiguities.



Autosomal recessive.

The mode of inheritance can't be x-linked unless individuals II-1 and II-2 are not penetrant.

Also, because affected individuals are not found in generation II, the disease is likely recessive.



X-linked recessive, x-linked dominant, or autosomal dominant

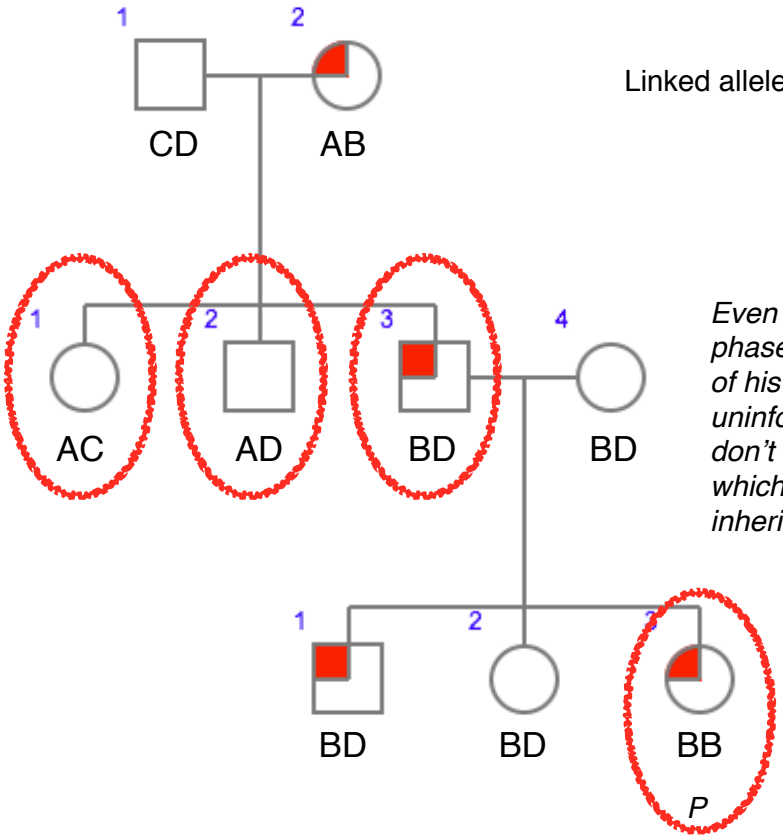
Because individuals in each generation are affected, the disease is likely dominant.

An affected son from an affected mother along with affected daughters from an affected father suggest x-linked inheritance.

Alternatively, small family sizes make the definitive answer about x-linked vs. autosomal difficult. It is possible that the disease is autosomal dominant because 25% of the time two affected children can come from a heterozygous affected parent.

It could also be x-linked recessive as long as individuals II-2 and I-4 are carriers

Question 3 (8 pts):
In the following three-generation pedigree, an autosomal dominant disease is thought to be linked to a neutral genetic marker. The marker has four forms in the population (A, B, C, and D). Please mark the following:
(1) The allele of the marker that could be linked to the disease-causing allele.
(2) Circle the individual(s) where we know the linkage phase.
(3) For every informative individual in generation III, please mark him or her as parental (P) or recombinant (R).



Linked allele: *B*

Even though we know the phase of individual II-3, two of his children are uninformative because we don't know which allele from which parent he or she inherited.

Please go to www.andersenlab.org/bio393 to fill out the post-quiz survey.
We are looking to improve the course! Your feedback is appreciated.