Genetics Challenges & Pedigrees

In this lab, you will finish applying what you have learned in lecture about genetics and get ready for the Take Home Exam.

(1) Notebook Review

To be sure that you have all the information you need for this lab, with your lab partners, go over the notes you took while doing the SPOC and from lecture and be sure you have the information listed below. You should fill in any gaps in your notes so everyone in your group has all they need. You can check these items off as you go.

- For each of these modes of inheritance:
  - What are the proper allele symbols for it?
  - What patterns of inheritance are consistent/inconsistent with it?
- The modes of inheritance are:
  - Autosomal Recessive (AR)
  - Autosomal Dominant (AD)
  - Sex-linked Recessive (SLR)
  - Sex-linked Dominant (SLD)
- How to count “unrelated carriers” in a pedigree

You will then discuss these with your TA as a class to clarify any issues that remain.

(2) Review
First, we'll go over any topics left over from the past two sessions that need further exploration.

(3) Pedigrees
In analyzing pedigrees, the objective is to produce a genetic model that fits the data given. Assume the trait is associated with one gene with two alleles, one disease allele and one normal allele. A complete model includes the following two components:

1) The mode of inheritance: autosomal recessive, sex-linked recessive, autosomal dominant, or sex-linked dominant.
* Note that there are two ways that questions regarding the mode of inheritance can be phrased:
Which modes of inheritance are consistent with this pedigree? Of the three modes of inheritance, which can explain this pedigree? There can be more than one answer to this question.

What is the most likely mode of inheritance? If more than one mode is consistent with the pedigree, then it is sometimes possible to decide which of the consistent modes is more likely. To evaluate which mode of inheritance is more likely, determine the number of unrelated carriers that are required for each mode. If you can assume that unrelated carriers are rare, the mode that requires the fewest unrelated carriers is the more likely.

2) The genotypes of all individuals in the pedigree.
If the trait is rare, individuals who marry into the family are unlikely to be carriers. We also assume that rare events like nondisjunction and mutation do not occur.

For each of the following pedigrees:
- What is the most likely mode of inheritance for this trait?
- Give the genotypes of all individuals in this pedigree.

Key to Symbols:

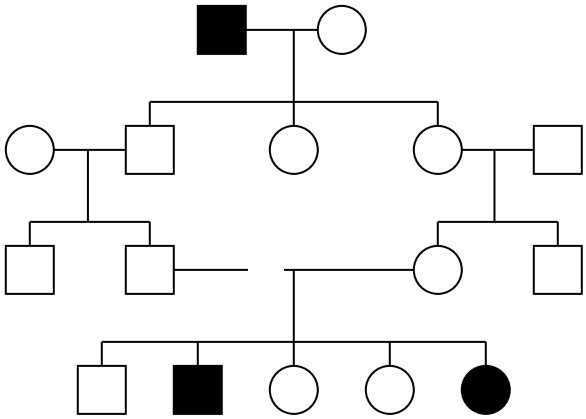
= normal male

= normal female

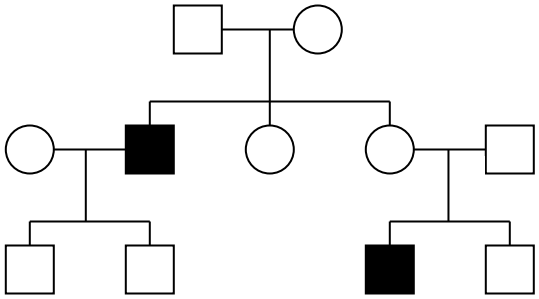
= affected male

= affected female

a)



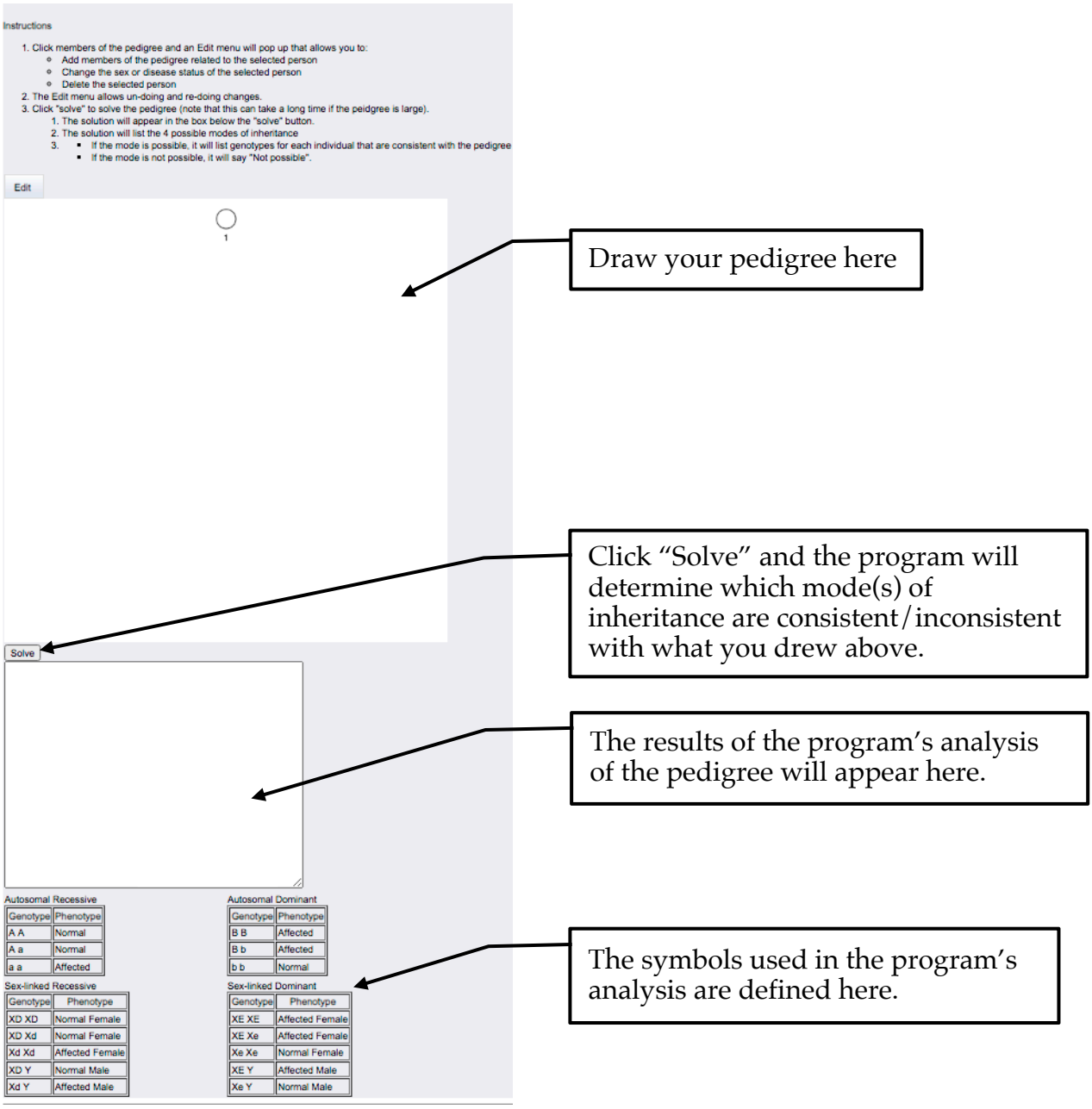
b)



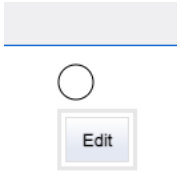
(4) jsPedigrees

jsPedigrees is a program that allows you to draw pedigrees and have the software determine which mode(s) of inheritance are consistent or inconsistent with the pedigree. It is a very useful tool for learning to solve pedigrees.

Launch jsPedigrees by clicking the link in the OLLM. You should see something like this (you will need to scroll down to see it all):



You draw a pedigree like this:
Click on the circle in the pedigree drawing window – she is the original founding mother of the family – and you'll see a little "Edit" button pop up. Note that, sometimes, this button appears in odd places on the screen and you need to look for it (sorry; I haven't figured out how to fix that bug).



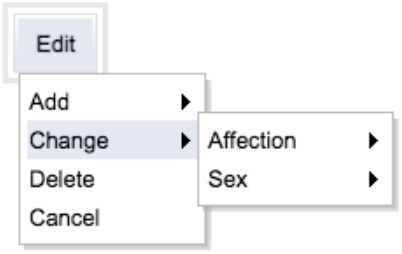
If you then click on the "Edit" button, you get this menu of choices with sub-menus:

Add

Choosing from the sub-menu adds various members to the family as described.

You can edit the sex of the individuals with the **Change** menu.

Change



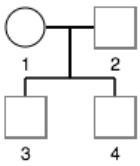
Changing **Affection** lets you make an individual affected or unaffected.

Changing **Sex** lets you set an individual to male or female.

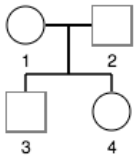
Delete deletes the selected individual and their children.

Cancel returns you to the pedigree drawing window.

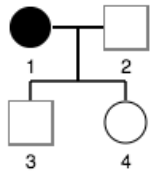
To make a simple family of a mother, father, son, and daughter, choose "Add -> 2 sons" and you should see:



Click on 4 and change his sex to female ("Change -> Sex -> Female") and you should see:



Just to see what happens, make the mother (1) affected ("Change -> Affection -> Affected") and you should see:



Take a few minutes to work out which model(s) this pedigree is consistent with. Remember that there are four:

- Autosomal Recessive (AR)
- Autosomal Dominant (AD)
- Sex-Linked Recessive (SLR)
- Sex-Linked Dominant (SLD)

You can then check your work by clicking the “Solve” button. You should see the results below the “Solve” button. See if you were correct.

You should now practice solving pedigrees and checking your work. One way to do this is to ask a neighboring group to draw a pedigree and then you solve it and check your work. You can then make a pedigree for them to solve. See what it takes to make more or less challenging pedigrees until you are confident in your understanding.

Preparing for the Take Home Exam

Take Home Exam 5 will be, in part, based on this lab. You should look at the exam on Blackboard before you leave lab today. You may want to use some of the remaining time in lab to prepare for the exam.