

## Instructions for the SexLink program

### The exercise

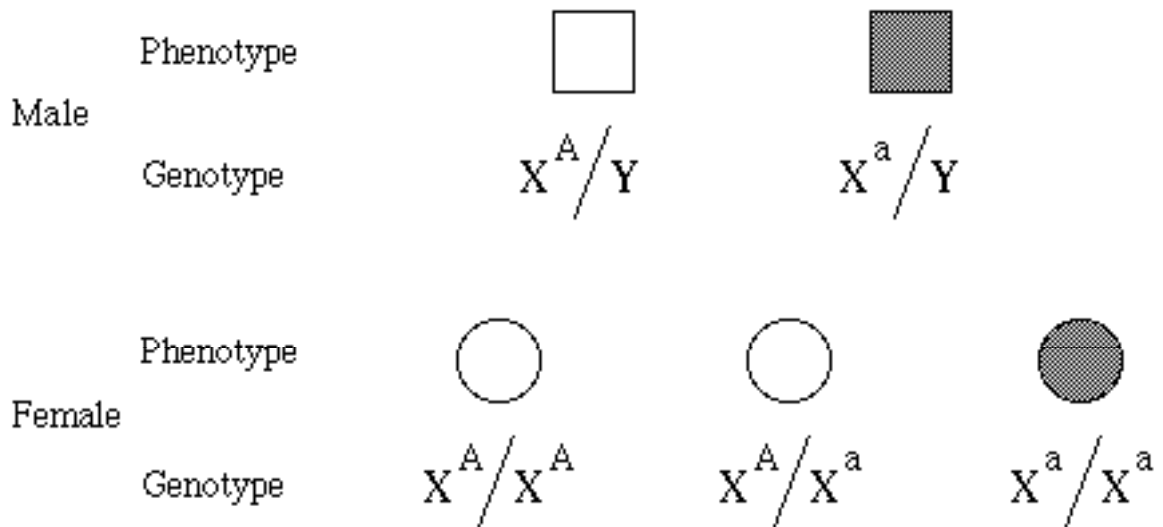
You are presented with a pedigree for a sex-linked recessive trait. The aim of the exercise is to fill in the genotypes of all individuals in the pedigree. When you have filled in all genotypes, click on the OK button, and your answers will be checked.

You will notice that there are four chromosomal genotypes provided in the box at the top of the screen. One of these is an X chromosome labelled with a question mark, and is intended to indicate that the genotype cannot be inferred with certainty. Your answer must take this into account. If you assign a genotype, say the A gene, to a genotype where either A or a is possible, then this answer will be marked wrong.

### The inheritance

The pedigree presents the inheritance of a sex-linked recessive trait. Filled symbols represent individuals with the trait.

The model of inheritance is therefore as follows:



### Rules for inheriting chromosomes

You will notice that the pedigree contains individuals whose parents are not shown. These 'founder' individual genotypes must be filled in with chromosomes dragged from the top box.

Individuals whose parents are shown must, on the other hand, inherit their chromosomes from their parents. Thus chromosomes dragged from the top box into these individuals will not be accepted. The one exception to this is the ? genotype, which can be placed anywhere in the pedigree.

## Frequency of trait

Two different situations are treated by this program. In the first, the affected character, or trait, is assumed to be rare in the population. This means that in any pedigree it is likely that the founder individuals will contain only a single occurrence of the deleterious recessive gene. The second type of character is assumed to be common. In this case you may need to invoke more than one occurrence of the recessive gene amongst founder individuals in order to explain the inheritance.

The program will tell you if you are dealing with a rare trait. In some pedigrees this may affect the answer. If, for example, you are told that the trait is rare and you have already assigned one recessive founder gene, then you can assume that all others are normal.

## Genotype placement

The position of a chromosome in an individual genotype will always be such that the chromosome inherited from the female parent is placed on the left and that from the male parent on the right. The genotype symbol is placed to indicate this. You do not need to worry about looking after this; the program will do this for you.

In founder individuals it is not clear which chromosome comes from a female or male parent. In these individuals the first chromosome assigned goes on the left, except that Y chromosomes will always be placed on the right side.

## To correct errors

Any chromosome wrongly placed in a pedigree may be replaced. The chromosome to be replaced must first be placed in the rubbish bin. Notice that this action will not replace any chromosomes that may already have been inherited from the replaced chromosome. It is up to you to ensure that the inheritance rules are not violated.

## Rare and common genes

This program has two possible settings. One assumes that the gene which causes the trait is rare, as is expected of a deleterious gene. The second setting of the program does not make this assumption.

If the program opens with "**Assume that the gene is rare in this exercise**", the first setting of the program has been chosen. This is taken to mean that **any pedigree will have only one occurrence of the gene in a founder individual**. If you try to put in a second occurrence, the program will object. Notice that females cannot show the trait in the pedigree under this assumption.

With the second program setting, there may be one or more initial occurrences of the recessive gene. In this case, the program will expect you to take into account **all** possible genotypes consistent with the pedigree.