

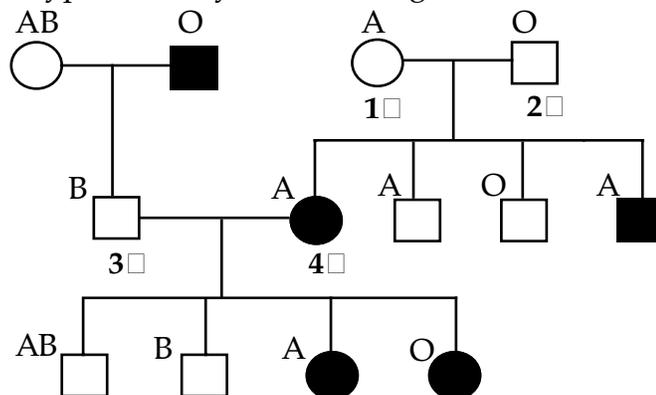
7.013 Section Problem Linkage & Pedigrees

These problems appeared on previous 7.01 exams.

I.

Alkaptonuria is an extremely rare disease. The gene for Alkaptonuria (ALK) has recently been shown to lie on human chromosome 9 and to be linked to the gene encoding the ABO blood group, with a recombination frequency of 11% between the loci.

A pedigree of a family with the disease is shown below, with affected individuals indicated in black. In addition, the blood type of family members is given.



The two alleles at the ALK locus will be denoted ALK⁺ and ALK⁻. The three alleles at the ABO blood group locus will be denoted A, B (which are co-dominant) and O (which is recessive to A and B).

You may also denote the genotypes as: chromosome from mom
 chromosome from dad

+ For homozygous positive ALK,
 $\frac{+}{+}$

$\frac{A}{A}$ for AA, $\frac{A}{B}$ for AB, $\frac{A}{O}$ for AO $\frac{O}{O}$ for OO, etc.
 $\frac{A}{B}$ $\frac{B}{B}$ $\frac{O}{O}$ $\frac{O}{O}$

So a person's blood and ALK genotype may look like this $\frac{B}{B} \frac{-}{-}$

a) What is the genotype of individual 1 at the ALK and ABO loci?

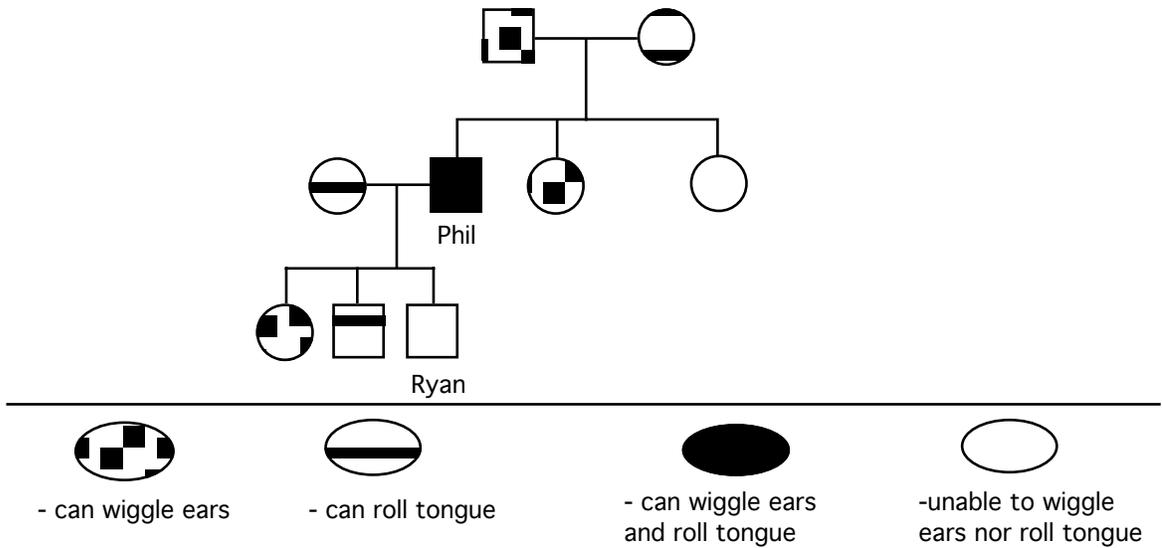
b) What is the genotype of individual 2 at the ALK and ABO loci?

c) What is the genotype of individual 3 at the ALK and ABO loci? Which alleles of each gene are carried on the chromosome he inherited from his father and which alleles are carried on the chromosome he inherited from his mother?

d) Individuals 3 and 4 are expecting their fifth child. A physician draws a prenatal blood sample and determines that the child has blood type B. What is the probability that the child will have alkaptonuria? Explain your answer.

II.

The ability to roll one's tongue in a U shape is an autosomal dominant trait maps to the same chromosome as another autosomal dominant trait, the ability to wiggle one's ears. Consider the following pedigree:



a) What are the genotypes of Phil and Ryan with respect to these two genes?

Use the letter **E** for ear wiggling and the letter **R** for tongue rolling.

Phil

Ryan

b) How do you account for the fact that Ryan is unable to roll his tongue or wiggle his ears?