Lecture 7 (FW)
February 11, 2009
Phenotype and Genotype
Reading: pp. 51-62

Annoucement:
A review session for the first mid term will be held on Tuesday, 2/24, from 5-6:30 PM in 159 Mulford Hall.

The mid term exam is during regular class time (1-2 PM) on Wed., 2/25. Sections 102 (Monday 3-4) and 103 (Tuesday 8-9) should go to 101 Morgan Hall for the exam. Other sections please come to our customary location, 100 GPB.

Lecture 7. More Chromosomal Inheritance
Primary Goal: Homologous Chromosomes can exchange pieces. There are different kinds of alleles and different kinds of chromosomes.

I. Review
A. Segregation of characters., Independent assortment
1. Punnett squares are easy ways to predict the outcome of a cross. Each parent has two of each kind of gene; since there are two of each kind of chromosome, it follows there are two of each kind of gene, but only one of them will end up in a gamete. Which gene will end up in any given gamete is completely random. All possible combinations resulting from fertilization of the gametes from both parents can be easily set forth in a "square", e.g., consider the gene for smooth peas (S=smooth; s= rough). All F-1 will therefore have the genotype Ss. Now cross the F-1 generation to produce an F-2

	S	s
S	SS	sS
s	sS	ss

2. If there are two or more characters, they still behave independently of one another. You can draw another Punnett square to see all the possibilities and predict the frequency of different kinds of genotypes from this kind of table.

B. But the above only holds if different characters are unlinked, i.e., are on different chromsomes. If they are on the same chromosomes, the linked characters will be distributed together**, EXCEPT**:

C. Linked characters can be recombined during meiosis by a physical breaking and rejoining of chromosomes, called "**crossing over**".
1. Recall that during prophase of the first meiotic division the doubled chromosomes of the same type, i.e., **homologous** chromsomes, form tight packets of 4 daughter chromosomes (tetrads). The strands of homologous chromosomes are intimately

associated and are entwined around one another. Breakage and rejoining can occur, which means linkage of given genes can be changed in such a process.

2. Consider a chromsome carrying genes A and B. If the homolog of that chromosome carries the recessive "a" and "b" versions of those two genes, we can say that A and B are linked on one chromosome, and a and b are linked on the other. But, if breakage and rejoining occurs between the A position and B position, then we could obtain linkages like A to b and a to B.

3. This change in linkage of versions of a gene is important and can help to locate the position of a gene on a chromosome.

II. Kinds of genes and chromosomes

A. We have mentioned different versions of the same gene, like Hb-β, and Hb-S. They both encode the same single protein, but there is a mutation that changes one amino acid. The technical term for two forms of the same gene is "**allele**". The two genes above are alleles.

B. There are also different kinds of chromsomes. In most organisms there is one pair of chromosomes called " **sex chromosomes**" because these chromome pairs are different in males and females. For example, in humans, there are 22 pairs of chromosomes and a 23rd pair composed of 2 X chromosomes in females and and X and a Y in males. The X and Y are called sex chromosomes. The other 22 pairs are called **autosomes**.

III. Kinds of gene expression (genotype and phenotype)

A. In **haploids** there is only one gene, and it is much easier to see the connection between genotype and phenotype. Haploid organisms usually do not employ sexual reproduction, and the DNA is often a circle, rather than a linear polymer.

B In **diploids**, however, there can be two different alleles on the two homolgous chromosomes. It is not so easy to determine the phenotype because:

1. Some genes are **dominant**. Like the genes Mendel worked on in peas. An example in humans is Marfan's syndrome, a connective tissue disease that affects a protein of connective tissue called "fibrillin". Even if there is one normal gene, having some abnormal fibrillin is enough to weaken the connective tissue so that carriers of only one such gene have long, gangly limbs, distinctive facial expressions, and weakness of the walls of blood vessels.

2. Some genes are either incompletely dominant, or **co-dominant**. For examples, the blood group type genes (A,B, O) are co dominant. If A and B are both present on different homologs, both A and B antigens are expressed.

3. Many genes are **recessive**. One dose of the gene is not enough to produce a phenotype (e.g., cystic fibrosis, albinism).

4. Another way to characterizing different alleles is call them: "gain of function" or "loss of function" phenotypes.

C. Genes on **sex chromosomes** present a special case. Though human females have two homologous sex chromosomes (2 X's), males only have one X. The Y chromosome of males has very few genes.

D. **Polygenic characters**.

Many, perhaps most, traits (phenotypes) are the result of action of many genes. Crosses that involve polygenic traits may give results that look like "blending", and this

makes analysis in experimental animals difficult. It also makes analysis of human pedigrees difficult. Skin color, height, weight, some congenital malformations (cleft palate, type I diabetes) are polygenic.

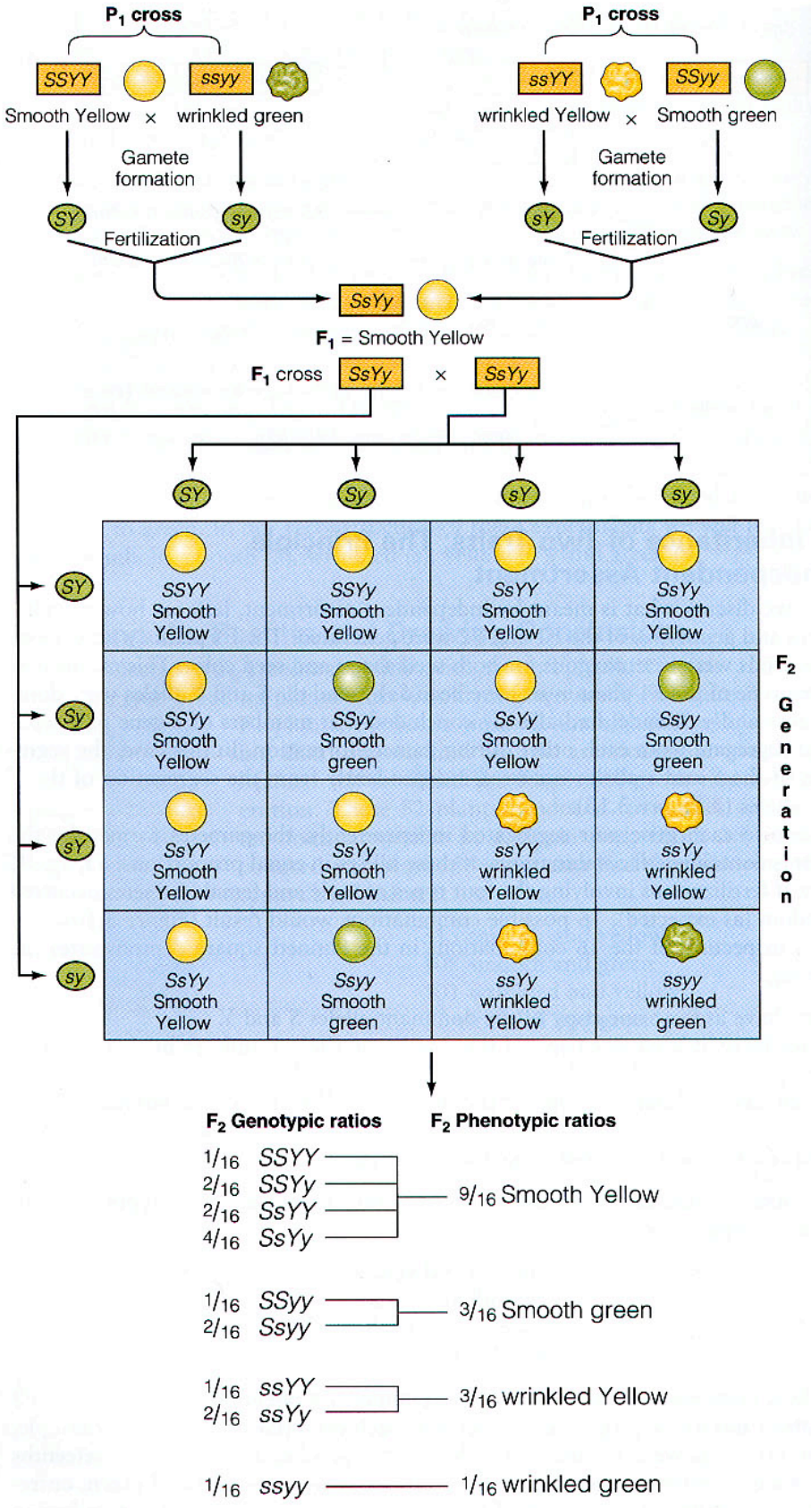
E. Pedigrees

Pedigress of phenotypes can often be useful in determining the kinds of alleles and chromosomal location of particular genes. The square (male) and circle (female), and lines connecting them are a useful shorthand for pedigree construction and analysis.

IV. Some terms to know: crossing over, homologous chromosomes, sex chromosomes, allele, haploid, diploid., co-dominant, phenotype, genotype

Take Home Message: The expression of genotype to create a phenotype shows a lot of variability, depending on ploidy, dominanace or lack thereof, location of sex chromosome, and how many different genes actually affect a phenotypic character, directly or indirectly.

Reading for next time: 51-62 again; 183-184; 143-153.


P1 cross
SSYY
ssyy
Smooth Yellow × wrinkled green
Gamete formation
SY
sy
Fertilization
P1 cross
ssYY
SSyy
wrinkled Yellow × Smooth green
Gamete formation
sY
Sy
Fertilization
SsYy
F1 = Smooth Yellow
F1 cross SsYy × SsYy
SY
Sy
sY
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SSYY Smooth Yellow
SSYy Smooth Yellow
SsYY Smooth Yellow
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SSYy Smooth Yellow
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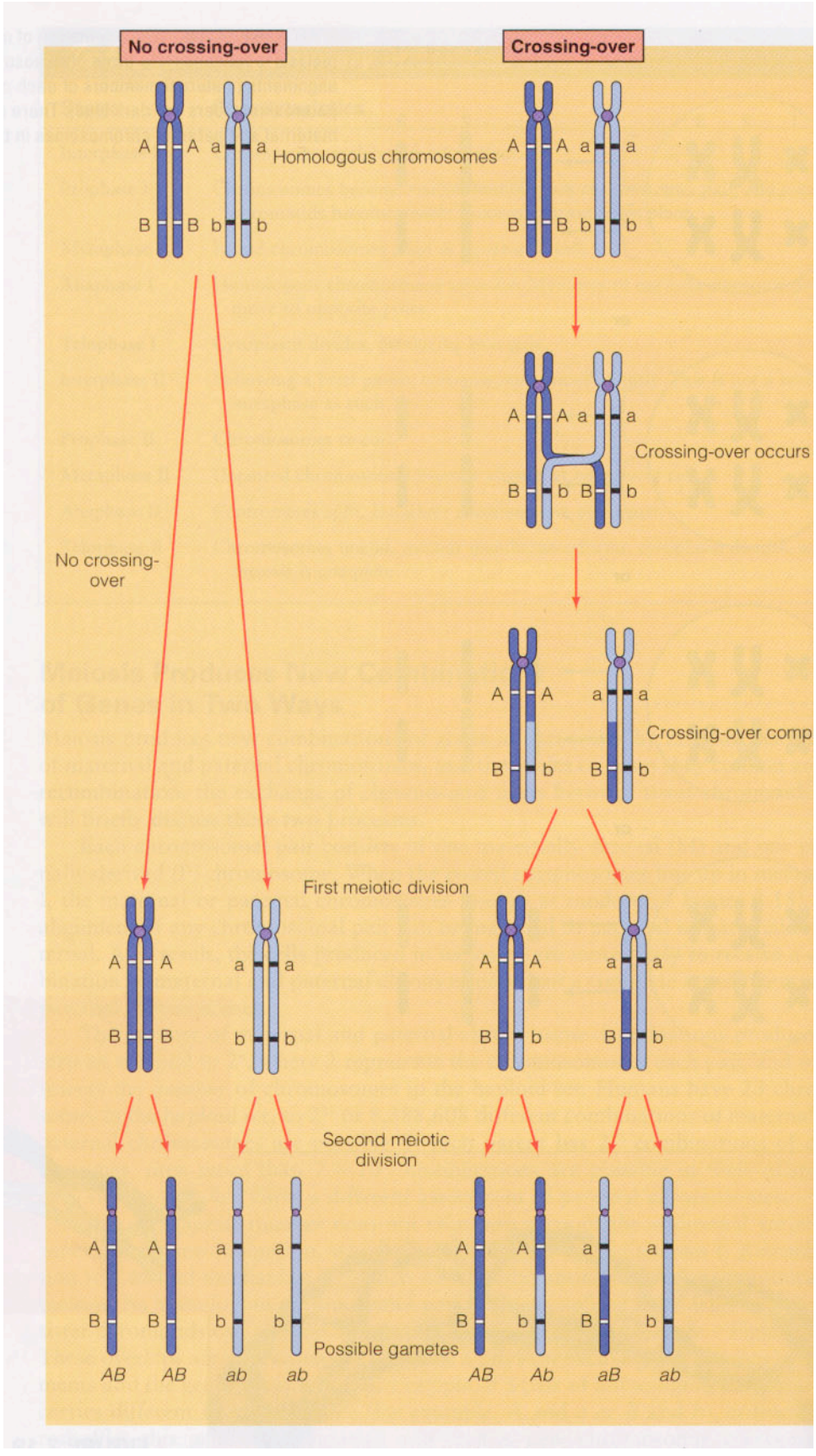
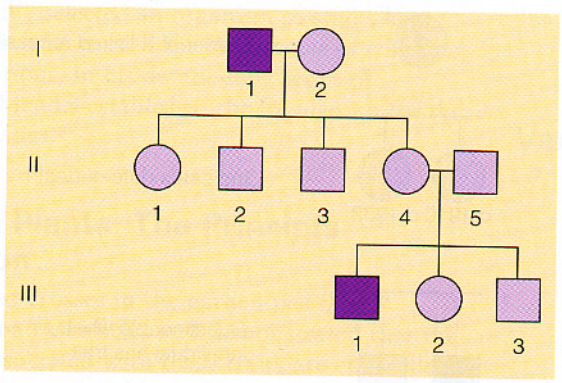
SsYY Smooth Yellow
SsYy Smooth Yellow
ssYY wrinkled Yellow
ssYy wrinkled Yellow
SsYy Smooth Yellow
Ssyy Smooth green
ssYy wrinkled Yellow
ssyy wrinkled green
F2 Generation
F2 Genotypic ratios
F2 Phenotypic ratios
1/16 SSYY
2/16 SSYy
2/16 SsYY
4/16 SsYy
9/16 Smooth Yellow
1/16 SSyy
2/16 Ssyy
3/16 Smooth green
1/16 ssYY
2/16 ssYy
3/16 wrinkled Yellow
1/16 ssyy
1/16 wrinkled green


No crossing-over
Crossing-over
Homologous chromosomes
Crossing-over occurs
No crossing-over
Crossing-over comp
First meiotic division
Second meiotic division
Possible gametes
AB
AB
ab
ab
AB
Ab
aB
ab


I
1
2
II
1
2
3
4
5
III
1
2
3