Lecture 4 Gene Interactions

1. Multiple Alleles

2. Codominance and incomplete dominance

3. Lethal alleles

4. Epistasis

5. Complementation

6. Penetrance and expressivity

7. Chi-square (χ^2) analysis

1. Multiple alleles. Blood groups are distinguished by the presence or absence of a certain antigene on the surface of erythrocytes.

O	i/i
A	I^A/i or I^A/I^A
B	I^B/i or I^B/I^B
AB	I^A/I^B

When both alleles are phenotypically revealed in a heterozygote, a **codominance** phenotype is observed.

P I^A/I^A (A) x I^B/I^B (B)

F_1 I^A/I^B (AB)

F_2 ¼ I^A/I^A 2/4 I^A/I^B ¼ I^B/I^B

In case of codominance, the phenotypic ratio coincides with the genotypic ratio (1:2:1).

Multiple alleles are not necessarily associated with codominance: C (full color) > c^{ch} (chinchilla) > c^h (himalayan > c (albino). There is a simple dominance, and a cross between any two will give a 3:1 ratio.

2. Codominance and incomplete dominance. In snap dragon plants:

P: RR (red) x rr (white)

F1: Rr (pink)

F2 1 RR 2 Rr 1 rr

This is an example of **incomplete dominance**

Codominance: heterozygote displays phenotype of *both* homozygotes (a sum of two phenotypes).

Incomplete dominance: it displays phenotype *intermediate* between the two homozygotes (an average of two phenotypes).

3. Lethal alleles. A cross of two yellow mice:

P	A^Y/A	X	A^Y/A	color: A^Y is dominant to A
F1	1 A/A	2 A^Y/A	1 A^Y/A^Y	viability: A^Y is recessive to A
	normal	yellow	(dead)	

A^Y is a dominant allele for color, but it is a recessive lethal allele for viability - a ***pleiotropic*** gene (a gene with multiple phenotypes).

4. Epistasis. In rodents the fur color is determined by several genes:

A series - distribution of the pigment: A^Y – yellow; A - agouti (normal), a - black, (A^Y>A>a)

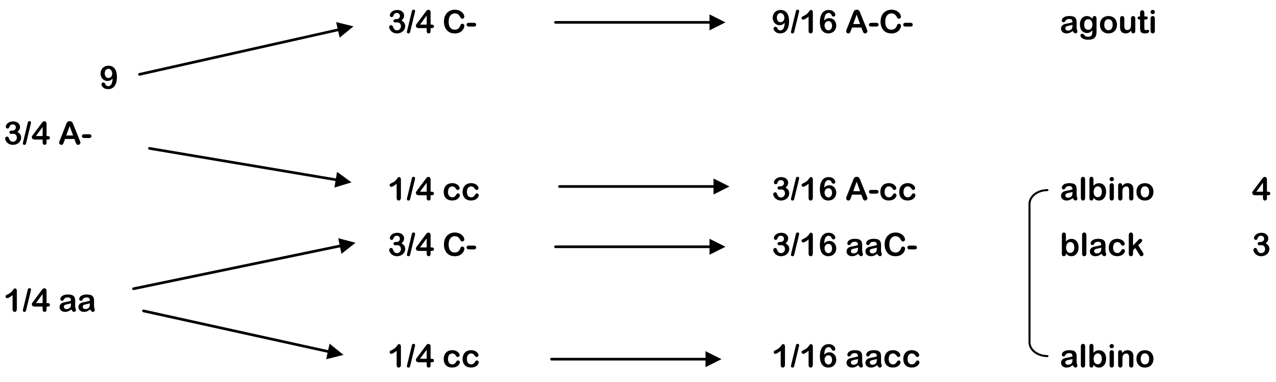
C series - ability to synthesize the pigment: C - full color, c – albino (C > c)

A cross of two true breeding mice:

P: AA CC (agouti) x aa cc (albino)

F_1: AaCc - all agouti

F_2



The phenotypic ratio is a modified Mendelian 9:3:4 ratio. In this case ***recessive alleles of one gene, when present in a homozygous form (cc), prevent expression of alleles of another gene (A or a).*** This condition is called ***recessive epistasis*** (from the Greek word "standing on"). A gene, whose alleles mask the other gene's expression, is called ***epistatic*** (*c*), and a gene, whose alleles are masked, is called ***hypostatic*** (*A, a*).

In summer squash:

Y/-; w/w are yellow, but *Y/-; W/-* are white; *y/y; w/w* are green, but *y/y; W/-* are white.

P Y/Y; W/W (white) x y/y; w/w (green)

F1 Y/y; W/w (white)

F2 9/16 Y/-; W/- 3/16 y/y/\;W/- 3/16 Y/-; w/w 1/16 y/y; w/w

white yellow green

12 3 1

This is called ***dominant epistasis***. *W* is epistatic, *Y* and *y* are hypostatic.

Biochemical explanation:

In w/w: X/- Y/- In W/-: X/- Y/-

White --↓---> Green --↓----> Yellow White ----|---> Green -------> Yellow

The W allele encodes **an inhibitor,** a compound that prevents conversion of the white substance into the green substance, and therefore also the subsequent conversion of the green into the yellow.

5. Complementation. In sweet peas:

P: A/A;b/b x a/a;B/B both are white

F1: A/a;B/b all are purple

F2: 9/16 A/-;B/- 3/16 a/a;B/- 3/16 A/-;b/b 1/16 a/a;b/b

purple white

9 7

Biochemical basis of ***complementation***:

A/- B/-

White (1) ↓------> White (2) ↓---------> Purple

aa B/-

White (1) -|----> White (2) ↓---------> White

The first white substance is converted into a purple pigment by two steps. An interruption of the pathway at any step in a recessive homozygote (by defective enzymes encoded by recessive alleles *a* or *b*) results in a white color. Only when at least one dominant allele of each gene is present (both *A/-* and *B/-*), is the purple substance produced.

6. Penetrance and expressivity. ***Penetrance is a percentage of individuals displaying a certain phenotype within the range of individuals carrying the corresponding genotype.*** Some genes have a 100 % penetrance, such as blood group genes. Others, such as brachydactyly (short fingers, autosomal dominant character) have a 50-80% penetrance. In addition, among individuals, which express a certain phenotype, the extent to which this phenotype is expressed can vary (e.g. very short fingers or slightly short fingers). This phenomenon is called variable ***expressivity.***

7. Chi-square analysis. Are the results of the following testcross consistent with independent assortment and a 1:1:1:1 ratio?

A/a;B/b	140
a/a;b/b	135
A/a;b/b	110
a/a;B/b	115

1. Choose a testable **null hypothesis**. In our case - independent assortment that predicts a 1:1:1:1 ratio.

2. Determine the **expected** number of progeny organisms of each phenotypic class according to the predicted ratio (in our case, 125).

3. Calculate $\chi^2 = \Sigma(O\text{-}E)^2/E = (140\text{-}125)^2\text{+}(135\text{-}125)^2\text{+}(110\text{-}125)^2\text{+}(115\text{-}125)^2 /125 = 5.2$

4. Determine the number of ‘degrees of freedom’: df = (number of classes - 1) = 3

5. Use these values and a table on page 30 of the textbook to find probability that observed differences between the observed progeny numbers and expected numbers are due to chance.

$0.5 > P > 0.1$ or $50\% > P > 10\%$

6. Make the final conclusion.

If $P > 5\%$ (0.05) ➔ accept the null hypothesis (cannot reject)

If $P < 5\%$ (0.05) ➔ reject the null hypothesis

In our case, the hypothesis of 1:1:1:1 ratio has withstood the test.