AP* BIOLOGY

MENDELIAN GENETICS AND χ^2

Teacher Packet

Mendelian Genetics and χ^2

Objective
To review the student on the concepts and processes necessary to successfully answer questions over Mendelian genetics and chi square analysis problems.

Standards
Mendelian genetics and chi square analysis are addressed in the topic outline of the College Board AP Biology Course Description Guide as described below.

II. Heredity & Evolution
- A. Heredity
  - Meiosis and gametogenesis
  - Eukaryotic chromosomes
  - **Inheritance patterns**
- B. Molecular Genetics
  - RNA and DNA structure and function
  - Gene regulation
  - Mutation
  - Viral structure and replication
  - Nucleic acid technology and applications

The principles of are tested every year on the multiple choice and occasionally make up portions of the free response section. There does seem to be an emphasis on sex linked inheritance and linked genes in general based on released exam material. Mendelian crosses are asked on all released multiple choice material that is currently available. The list below identifies free response questions that have been previously asked over this topic. These questions are available from the College Board and can be downloaded free of charge from AP Central http://apcentral.collegeboard.com.

Free Response Questions	
2003 Question #1	

Mendelian Genetics and χ^2

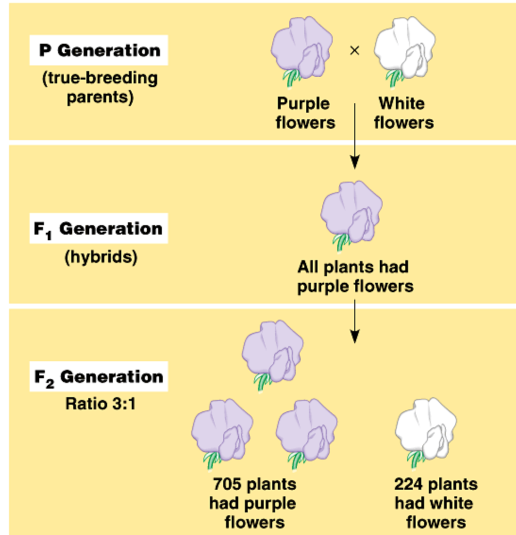
Mendelian or Classical Genetics

Gregor Mendel is credited as the 1st to actually quantify genetic crossing experiments.

Mendel's Experimental Design
→Use pure strains (self fertilization)
→Cross fertilize
→Track data quantitatively (ratio is key)

Results
→ The F_1 generation displayed no blending of traits.
→ F_2 noticed some recessive traits re-emerged showing a phenotypic ratio of 3:1. Note that the actual genotypic ratio is 1:2:1 since recessive traits are present but masked.

Explanation?→ According to F_1 results one allele can mask another allele. According to F_2 results the masked allele is not destroyed, but simply "dominated" by the other.



Mendel's Laws of Heredity

Law of Segregation
→Alleles don't blend. One of two alleles is passed on in sex cells; probability or chance alone determines which allele will be passed to the gamete. Punnett squares work because of this law.

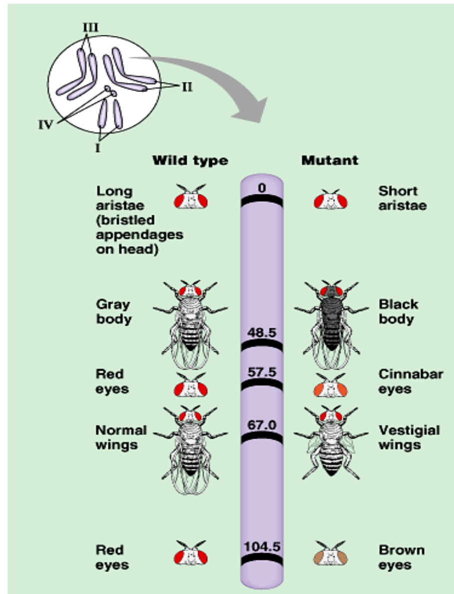
Law of Independent Assortment
→Each pair of alleles segregates independently of other alleles.

Mendel meets the modern genetic language…

The stand alone meanings of many words used frequently in genetics often seem similar. The following is a “relative” summary of the language of genetics.

→Parents transmit **genes**; genes combine to form a **trait** (like purple or white). A chromosome is full of genes found at **loci** (location of the gene) that can be seen on a **gene map**. Each version of this gene is called an **allele** (like “W” or “w”). A sexually reproducing individual will receive one allele from each parent. Based on the relationship of the alleles inherited, a pair of alleles may be described as a **homozygous genotype** or a **heterozygous genotype**. Each allele does not destroy the other. The **dominant allele** is the one that is the outwardly expressed trait (**phenotype**) in a heterozygote and is said to “mask” the **recessive allele**.

Gene Map

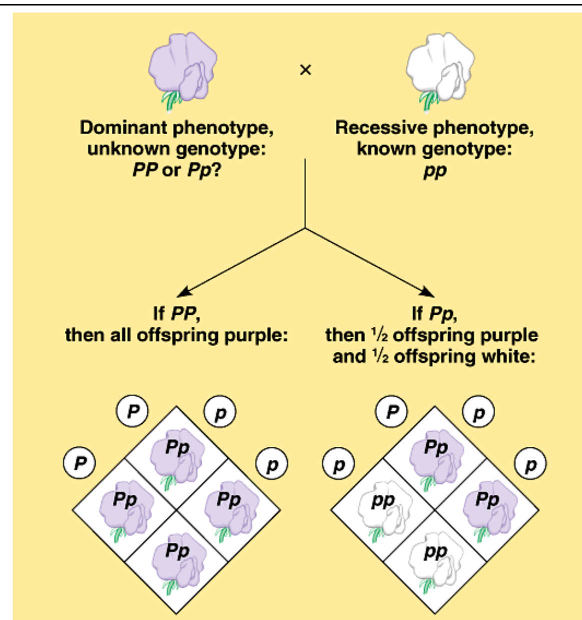


The significance of the **test cross**.

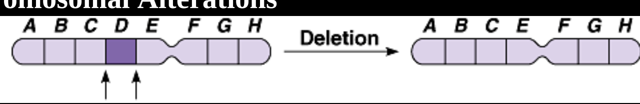
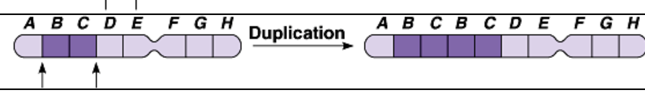
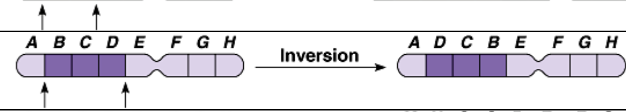
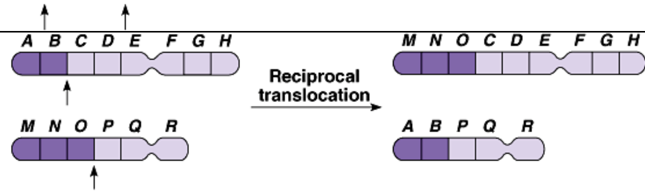
→Suppose that an individual shows a dominant phenotype. How might one determine the genotype of this individual as is could be homozygous dominant or heterozygous? The individual could be crossed with a homozygous recessive individual. If an adequate sample size is present, and all individuals in the F_1 generation continue to display the dominant phenotype it is highly likely that the individual in question is homozygous dominant for the trait of interest.

Note: The dominant allele is not always the most common in a population (polydactylism).

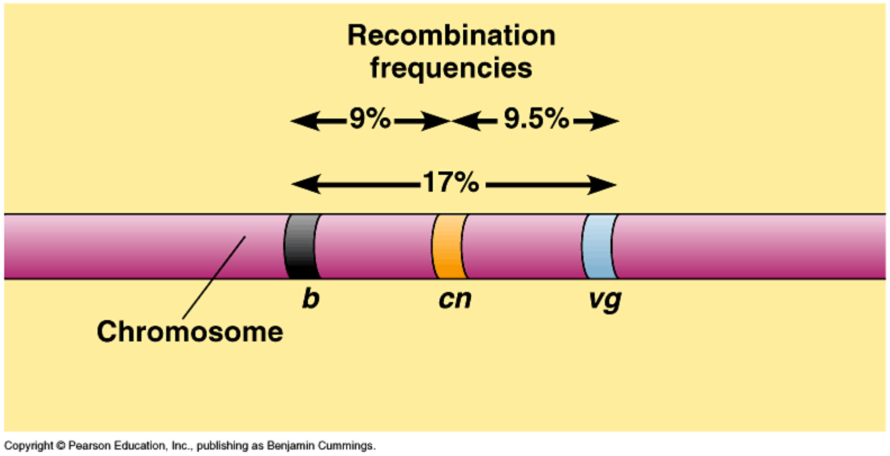
Note: There are several genetic crosses that are “non-Mendelian” and will be discussed shortly



Exceptions to Mendelian Genetics	
Incomplete Dominance →A Blending of traits: A homozygous white flower crossed with a homozygous red flower for example will create a pink heterozygote.	Codominance →Both traits are expressed simultaneously: A homozygous white flower crossed with a homozygous red flower would create a red and white flower.
Epistasis →Many genes controlling one phenotype or the gene at one locus controlling the expression of a gene at another locus: Consider the biochemical cascade A→B→C→D. If a single mutation occurs in A,B, or C, the product D will be effected.	Pleiotropy →One gene controlling many phenotypes (essentially the opposite of epistasis): The agouti gene in mice is linked to coat color, obesity, and certain tumors.
Polygenic Inheritance →Many genes controlling one phenotype with an additive effect: Laborador Retreiver colors are polygenic AABBCC = Black Lab AaBbCc = Lighter Aabbcc= Lightest *Comparatively, epistasis is more like a moving assembly line where each gene is dependent on the next.	Multiple Alleles →More than 2 alleles controlling one trait and therefore more than a simple dominant/recessive relationship between alleles: Blood types are the most common example of multiple alleles. Both A and B are dominant while O is treated as recessive. Type A = I^AI^A or I^Ai Type B = $I^B I^B$ or I^Bi Type AB = $I^A I^B$ Type O = ii
Sex linked traits →The genes of interest are located on the sex chromosomes. Sex linked traits are carried on the X chromosome as no genes shared by both male and female can be carried on the Y chromosome. Hemophilia is common example on the AP exam.	Environmental Effects →Environmental cues give to rise to modifications in phenotype: The coat of the arctic fox changes from white to earthy brown as the temperature rises due to a color related enzyme. The fox benefits from this well timed camouflage.

Chromosomal Alterations	
Deletion →removal of a chromosomal segment	
Duplication → repetition of a chromosomal segment	
Inversion → reversal of a chromosomal segment	
Translocation →Movement of a chromosomal segment from one chromosome to another nonhomologous chromosome. Crossing over (meiosis) by contrast occurs between homologous chromosomes. Reciprocal translocation means that chromosomal material is traded (give and receive).	

Genetic Maps, Linked Genes, & Recombination



Recombination

→Recombination through crossing over occurs during prophase of meiosis I. Since crossover is random, the likelihood that 2 genes crossover increases as the distance between those 2 genes increases since there are more points between them where crossover may occur. The unit of measure of this distance is the map unit.

Gene Linkage Maps

→Using crossover frequencies one can construct a linkage map to visually represent the relative distances between genes.

Linked Genes

→Genes that are close together are said to be linked as they often “travel together” during crossing over.

Gene Linkage Example Problem:

Reconstruct the order of the gene map based on the recombination frequencies listed below.

GENE	CROSSOVER FREQUENCY
A-C	30%
B-C	45%
B-D	40%
A-D	25%

1. It is usually most efficient to begin with the genes that are farthest apart.

2. Next, begin overlapping genes. It is sometimes more productive to attempt to pick internal genes first.

3. Remember that crossover frequencies cannot exceed 50%

B------------------45------------------C

insert the possibilities for D

D-------------40--------------B-----------------40-------------D---C *(could go either way)*

blindly choose B-D-C over D-B-C

B-----------------40-------------D----C

insert A and deduce map distances to match the table

B-----15----A--------25--------D--5-C

Correct Answer: BADC

Chi Square Analysis

What's the point?

→If a person were to flip a coin 100 times. How many heads and how many tails should he get?...50 heads and 50 tails. Suppose that he got 51 heads and 49 tails….did he just disprove probability? What about 55:45? What about 60:40? At what point does one conclude that the coin is likely abnormal or that some other aspect of the experiment is likely askew? Chi Square to the rescue! Chi square determines whether or not results seem to confirm or call into question a hypothesis.

Null Hypothesis vs. Alternative Hypothesis

→Null: no statistically significant difference between expected and observed results

→Alternative: explanation (hypothesis) of why the results are so different from what's expected

$$\chi^2 = \sum \frac{(F_o - F_e)^2}{F_e}$$

F_e = *Frequency expected*
F_o = *Frequency observed*

How to use the information.

→Compare χ^2 results to the χ^2 table. From this information, one can determine with a specified amount of certainty that the data is legitimate.

→ **Probability values**: P values measure from 0 - 1. P = 1 is perfectly expected data (like 50 heads and 50 tails) meaning that we accept our null hypothesis. P = 0 is completely unexpected data (like 100 heads and 0 tails OR 0 heads and 100 tails) and we should create an alternative hypothesis. Most data will fall somewhere in between. In "life science circles", it is accepted that a P value of 0.05 or higher is considered acceptable.

→**Degrees of Freedom**: DF represents the number of alternate possibilities. In other words, total possibilities-1. In our coin example we can get heads or tails, so we have 2 possibilities – 1 = 1 degree of freedom. If we were looking at a 6 sided die, we would have 6 - 1 = 5 degrees of freedom.

χ^2 critical values

	probability					
df	.995	.90	.50	.10	.05	.025
1	.000	.016	.455	2.706	3.841	5.024
2	.010	.211	1.386	4.605	5.991	7.378
3	.072	.584	2.366	6.251	7.815	9.348
4	.207	1.064	3.357	7.779	9.488	11.143

*Line up the predetermined acceptable P value with the appropriate number of degrees of freedom. If your chi square value is less than this number, accept the null hypothesis. If the chi square value is greater than or equal to this number, the null hypothesis must be rejected and an alternative hypothesis must be created.

χ^2 Example Problem:

From a cross between two plants, one completely heterogyzous tall green (SsYy) and the other heterozygous tall yellow (Ssyy), the following results were observed:

52 tall green
55 tall yellow
26 short green
27 short yellow

A. Show by Punnett square (or other labeled diagrams) the cross between the two original plants and the expected phenotypic ratio of offspring.

B. Use χ^2 calculations to determine whether or not the variations in the observed results could be due to chance.

1) *Set up the problem to determine the expected ratio:*

SsYy x Ssyy

Show possible gametes from each parent: SsYy can produce <u>SY</u>, <u>Sy</u>, <u>sY</u>, and <u>sy</u>
Ssyy can produce <u>Sy</u> and <u>sy</u>

The Punnett square should be 4 x 2 with the following phenotypic ratio:

	SY	**Sy**	**sY**	**sy**
Sy	SSYy	SSyy	SsYy	Ssyy
sy	SsYy	Ssyy	ssYy	ssyy

Expected ratio: 3 tall green, 3 tall yellow, 1 short green, 1 short yellow

USING THE OBSERVED RESULTS, DETERMINE WHAT WAS WOULD BE <u>EXPECTED</u> FROM THE 3:3:1:1 RATIO (Hint: 3:3:1:1 is actually <u>8</u> parts)

Total the results and divide by <u>8</u> to determine what one part would be.
160/8 = 20
Three parts would be 3(20) = 60.
Therefore, the EXPECTED results from the cross would be 60:60:20:20.
You already have the <u>observed</u> results.

$$X^2 = \frac{(52-60)^2}{60} + \frac{(55-60)^2}{60} + \frac{(26-20)^2}{20} + \frac{(27-20)^2}{20} = \frac{64 + 25 + (3)36 + (3)49}{60} = \frac{334}{60} = 5.73$$

Degrees of freedom = 3
Critical value (from chi-square table) = 7.815

Conclusion: 5.73 is less than 7.815 and within the limits of chance; therefore, the observed results could fit the 3:3:1:1 ratio.

Multiple Choice

1. Genes S and T are not linked. If there is a 50% probability that allele S is in a gamete and there is a 50% probability that allele T is in a gamete, what is the probability that both are in the same gamete?

   (A) 5%
   (B) 25%
   (C) 50%
   (D) 75%
   (E) 100%

B	There is a 25% chance that S and T will both be found in the same gamete. (.5)(.5) = .25

2. The trait for tall pea plants is (T) and the trait for short pea plants is (t). The trait for smooth peas is (S) and the trait for wrinkled is (s). Two plants are crossed yielding an F_1 generation with 612 tall plants with smooth peas and 188 short plants with wrinkled peas. Which of the following is the most likely genotype of the parent generation?

   (A) ttss x ttss
   (B) TTss x TTss
   (C) TtSs x TtSs
   (D) TTSS x ttss
   (E) TtSS x Ttss

C	The F_1 generation represents ~ 3:1 ratio typical of heterozygote crosses. TtSs crossed with TtSs will give a 3:1 ratio.

3. A male and female have 3 offspring all of which are female. The couple is now pregnant again. What is the likelihood that the next child will be a female?
   (A) 1/16
   (B) ¼
   (C) 1/3
   (D) ½
   (E) ¾

D	Each birth is independent of the previous birth. There is always a 50% chance that the next <u>individual</u> birth will be a female. Be careful!- see next question

4. What is the likelihood that a couple will have 4 offspring all of which are female?
   (A) 1/16
   (B) ¼
   (C) 1/3
   (D) ½
   (E) ¾

A	Each individual birth represents a ½ chance of producing a female. The likelihood that four consecutive births will all be female is $(½)^4$ or 1/16.

5. A child is born with the blood type B. The mother of the child is blood type O. Which of the following statements is most correct?

   (A) The father must be type B
   (B) The father could be type AB
   (C) The father passed along a recessive allele
   (D) The mother could have passed along a type B allele
   (E) The mother could have passed along a dominant allele

B	The child received a recessive *i* allele from his mother as she is *ii*. This is a recessive allele. The father could be type B but could also be type AB. In this case only answer choice that is feasible is choice B.

6. If traits X, Y, and Z are consistently inherited together, which of the following best explains why this is the case?

   (A) These 3 traits are all dominant.
   (B) The parents both carry the dominant form of each trait.
   (C) These 3 traits are located on different chromosomes
   (D) These 3 traits are located close to each other on the same chromosome.
   (E) These 3 traits are located far from each other on the same chromosome.

D	The genes that code for these traits appear to be linked. Linked genes are located near one another on the same chromosome so that they are not often separated during the process of crossing over in meiosis. The closer they are to each other, the less likely they are to separate. Dominance is irrelevant in this scenario.

7. Hemophilia is a sex-linked recessive trait. A male hemophiliac and phenotypically normal female have a girl that is a hemophiliac. All of the following statements are correct EXCEPT:

(A) The daughter inherited a recessive gene from each parent.
(B) The daughter inherited a dominant allele from her mother.
(C) The mother is a carrier of hemophilia
(D) The genotype of the mother is $X^H X^h$
(E) The genotype of the father is X^hY

B	The daughter did not inherit a dominant allele from her mother. If this were the case the daughter would not be a hemophiliac. All other statements are true.

<u>Questions 8-10</u> refer to the following genetic terms

(A) Codominance
(B) Epistasis
(C) Multiple alleles
(D) Pleitotropy
(E) Incomplete Dominance

8. A red flower and a white flower produce a pink flower

E	Incomplete dominance shows as a phenotypic blending of traits.

9. A red flower and a white flower produce a red and white streaked flower

A	In codominant inheritance both alleles are expressed.

10. A single gene coding for multiple phenotypes

D	Pleiotropy is when a single gene codes for multiple phenotypes.

Free Response

1. Imagine an isolated species of caterpillars with five distinct color variations living in a prescribed area. Many counts of these caterpillars revealed that the colors were equally balanced. In July a count of 50caterpillars revealed:

10 green
10 yellow
10 brown
10 white
10 red

Also in July a species of bird that preys on caterpillars was introduced in the area of the caterpillar population. After two months the caterpillar population had dropped, and when the count was done, the color variations were no longer balanced. The following results were obtained from a random count of 25 caterpillars in early September:

10 green
5 yellow
7 brown
1 white
2 red

A. Using chi-square, determine if the color variations found in the September count could be due to chance, or if some other factor influenced results.

χ^2 **critical values**						
	probability					
df	.995	.90	.50	.10	.05	.025
1	.000	.016	.455	2.706	3.841	5.024
2	.010	.211	1.386	4.605	5.991	7.378
3	.072	.584	2.366	6.251	7.815	9.348
4	.207	1.064	3.357	7.779	9.488	11.143

1 pt for each of the following

Mathematics
_original ratio is 1:1:1:1:1
_choosing 5 as the correct expected value for each color OR showing such mathematically
_using the correct observed values for all 5 colors in the chi square equation
_correct answer of 10.8 or rounded value of ~11

$$X^2 = \frac{(10-5)^2}{5} + \frac{(5-5)^2}{5} + \frac{(7-5)^2}{5} + \frac{(1-5)^2}{5} + \frac{(2-5)^2}{5} = \frac{54}{5} = 10.8$$

Data Interpretation
_the critical value at 0.05 is 9.49
_there are 4 degrees of freedom.
_the number is greater than the critical value AND that this means the results are not due to chance

B. What was the most likely influence in the results revealed in the September count?

1 pt for each of the following
_birds fed more frequently on caterpillars with conspicuous colors _camouflaged caterpillars are not as easily detected by the birds and therefore avoided consumption

C. Propose a possibility of what may happen to the caterpillar color variations if the birds and caterpillars remain in the area for five years and explain the biological significance of this outcome.

1 pt for each of the following
_Populations will shift such that the vast majority of caterpillars will be green or brown OR the yellow, white, and red colors will become increasingly rare in the population _Adaptations such as camouflaging colors provide an advantage to the caterpillars

Free Response

2. Chromosomal alterations can have significant evolutionary impact.

A. Observe the collected recombination data below. Recent experiments show that gene C is more closely linked with D than with A. Reconstruct a gene map and determine the map distance between C and D.

GENE	CROSSOVER FREQUENCY
A-B	7%
A-C	18%
C-D	?
B-D	35%

1 pt for each of the following
_gene map order is BACD _map distance between C and D is 10 map units

B. Discuss the events involved in crossing over during meiosis and describe the evolutionary significance of this event.

1 pt for each of the following
_synapsis is the process of bringing together homologous chromosomes during prophase of meiosis I _crossing over occurs when genetic material is exchanged between homologous chromosomes _crossing over produces genetic variation _genetic variation allows for a mixture of adaptations that may prove beneficial for the survival of an organism.

C. Describe 2 of the following 4 chromosomal alterations. In light of evolution, are individual alterations such as these beneficial or detrimental? Include a hypothetical or literal example in your answer

- Deletion
- Duplication
- Inversion
- Translocation

1 pt for each of the following
Chromosomal Alteration *2 pt max* _Deletion is the removal of a chromosomal segment _Duplication is the repetition of a chromosomal segment _Inversion is the reversal of a chromosomal segment _Translocation is the movement of a chromosomal segment from one chromosome to another nonhomologous chromosome. **Evolution** _genetic diversity on the whole is beneficial to the population as it increases likelihood of survival of the population _a genetic alteration could be harmful, neutral, or beneficial. _additional point for a sensible example of a situation in which the alteration is beneficial, neutral, or beneficial