Topic 3 and 10 Genetics and Meiosis

Topic 3.4 and 10.2 Inheritance
Essential idea: The inheritance of genes follows patterns AND Genes may be linked or unlinked and are inherited accordingly.

Understandings

- 3.4.U1 Mendel discovered the principles of inheritance with experiments in which large numbers of pea plants were crossed.
- 3.4.U2 Gametes are haploid so contain only one allele of each gene.
- 3.4.U3 The two alleles of each gene separate into different haploid daughter nuclei during meiosis.
- 3.4.U4 Fusion of gametes results in diploid zygotes with two alleles of each gene that may be the same allele or different alleles.
- 3.4.U5 Dominant alleles mask the effects of recessive alleles but co-dominant alleles have joint effects.

Understandings

- 3.4.U6 Many genetic diseases in humans are due to recessive alleles of autosomal genes, although some genetic diseases are due to dominant or co-dominant alleles.
- 3.4.U7 Some genetic diseases are sex-linked. The pattern of inheritance is different with sex-linked genes due to their location on sex chromosomes.
- 3.4.U8 Many genetic diseases have been identified in humans but most are very rare.
- 3.4.U9 Radiation and mutagenic chemicals increase the mutation rate and can cause genetic diseases and cancer.

Understandings

- 10.2.U1 Gene loci are said to be linked if on the same chromosome.
- 10.2.U2 Unlinked genes segregate independently as a result of meiosis.
- 10.2.U3 Variation can be discrete or continuous.
- 10.2.U4 The phenotypes of polygenic characteristics tend to show continuous variation.
- 10.2.U5 Chi-squared tests are used to determine whether the difference between an observed and expected frequency distribution is statistically significant.

Applications

- 3.4.A1 Inheritance of ABO blood groups. The expected notation for ABO blood group alleles: O = i, A=IA, B = IB.
- 3.4.A2 Red-green colour blindness and hemophilia as examples of sex-linked inheritance.
- 3.4.A3 Inheritance of cystic fibrosis and Huntington's disease.
- 3.4.A4 Consequences of radiation after nuclear bombing of Hiroshima and accident at Chernobyl.

- 10.2.A1 Morgan's discovery of non-Mendelian ratios in Drosophila.
- 10.2.A2 Completion and analysis of Punnett squares for dihybrid traits.
- 10.2.A3 Polygenic traits such as human height may also be influenced by environmental factors.

Skills

- 3.4.S1 Construction of Punnett grids for predicting the outcomes of monohybrid genetic crosses.
- 3.4.S2 Comparison of predicted and actual outcomes of genetic crosses using real data.
- 3.4.S3 Analysis of pedigree charts to deduce the pattern of inheritance of genetic diseases.

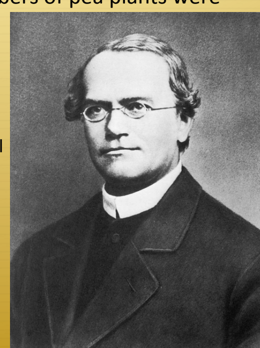
- 10.2.S1 Calculation of the predicted genotypic and phenotypic ratio of offspring of dihybrid crosses involving unlinked autosomal genes.
- 10.2.S2 Identification of recombinants in crosses involving two linked genes.
- 10.2.S3 Use of a chi-squared test on data from dihybrid crosses.

I. Mendel and the principles of inheritance

A. Mendel discovered the principles of inheritance with experiments in which large numbers of pea plants were crossed-

Johann Gregor Mendel (1822-1884)

A. Because of his work with pea plants Mendel is considered the father of modern genetics.
B. He planted 1000s of seeds per trial and carried out many trials to be sure of his results.
C. His published work (1865) is now considered important, but at the time was ignored for 30 years.



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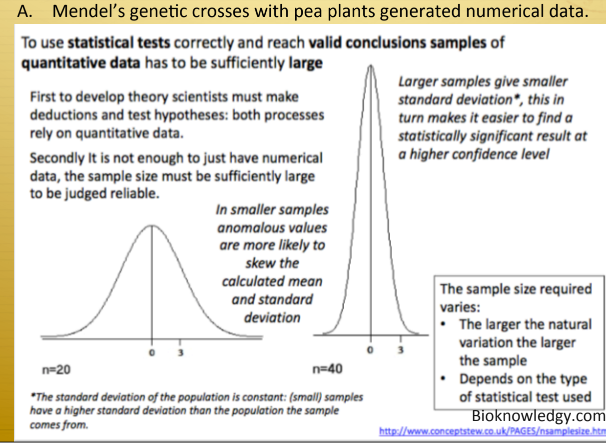
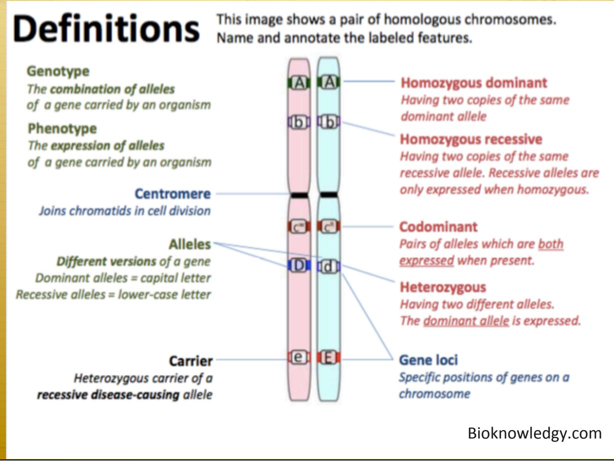
II. Nature of Science: Making quantitative measurements with replicates to ensure reliability

A. Mendel's genetic crosses with pea plants generated numerical data.

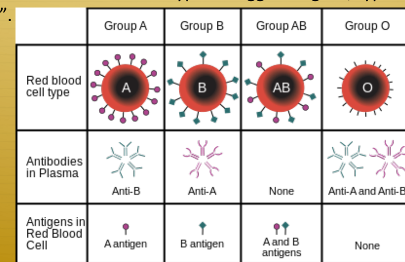


III. Basic Mendelian Genetics



IV. Dominant, Recessive and Co-dominant alleles

A. Inheritance of ABO blood groups.
1. The ABO blood type classification system uses the presence or absence of certain antigens on red blood cells to categorize blood into four types.
2. Distinct molecules called agglutinogens (a type of antigen) are attached to the surface of red blood cells.
3. There are two different types of agglutinogens, type "A" and type "B".



	Group A	Group B	Group AB	Group O
Red blood cell type	A	B	AB	O
Antibodies in Plasma	Anti-B	Anti-A	None	Anti-A and Anti-B
Antigens in Red Blood Cell	A antigen	B antigen	A and B antigens	None

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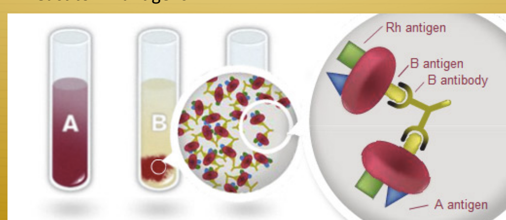
IV. Dominant, Recessive and Co-dominant alleles

3. Antibodies (immunoglobulins) are specific to antigens and are used by the immune system to recognize 'foreign' antigens. If you are given the wrong blood type your body might react fatally as the antibodies cause the blood to clot!
4. Blood type O is known as the universal donor, as it has no antigens against which the recipient immune system can react. Type AB is the universal recipient, as the blood has no antibodies which will react to AB antigens.



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IV. Dominant, Recessive and Co-dominant alleles

B. The ABO blood type is controlled by a single gene, the ABO gene. This gene has three different alleles:

i O allele (no antigen is produced)
I^A A allele (type "A" antigen is produced)
I^B B allele (type "B" antigen is produced)

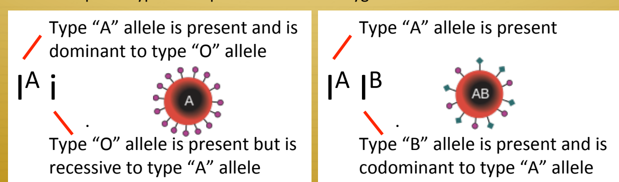
I^A — Allele variant
Gene (lower case for "recessive" alleles

Genotype	Phenotype	Antigen Produced
ii	O	none
I^AI^A or I^Ai	A	Type A antigen
I^BI^B or I^Bi	B	Type B antigen
I^AI^B	AB	Both A & B antigen

IV. Dominant, Recessive and Co-dominant alleles

C. Dominant alleles mask the effects of recessive alleles but co-dominant alleles have joint effects-

1. Dominant alleles have the same effect on the phenotype whether it is present in the homozygous or heterozygous state
2. Recessive alleles only have an effect on the phenotype when present in the homozygous state
3. Codominant alleles are pairs of different alleles that both affect the phenotype when present in a heterozygote



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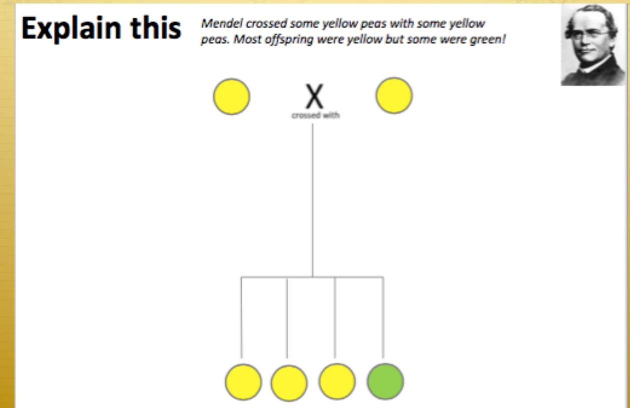
V. Punnett Grids AND Segregation of Alleles



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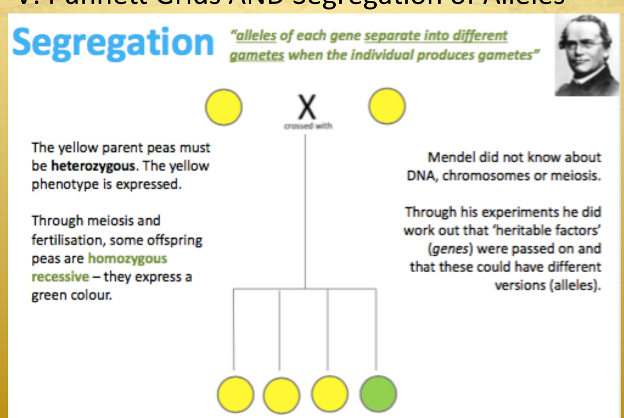

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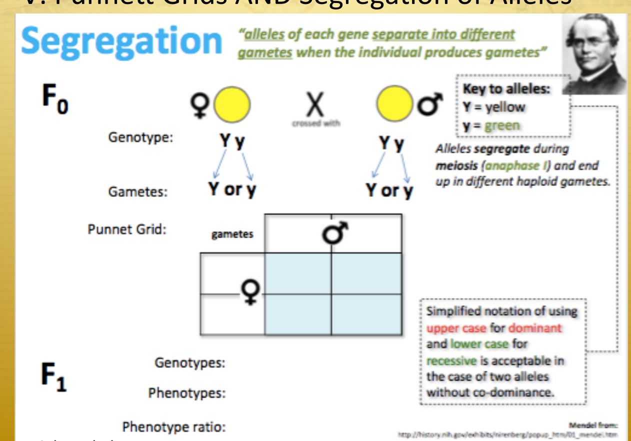

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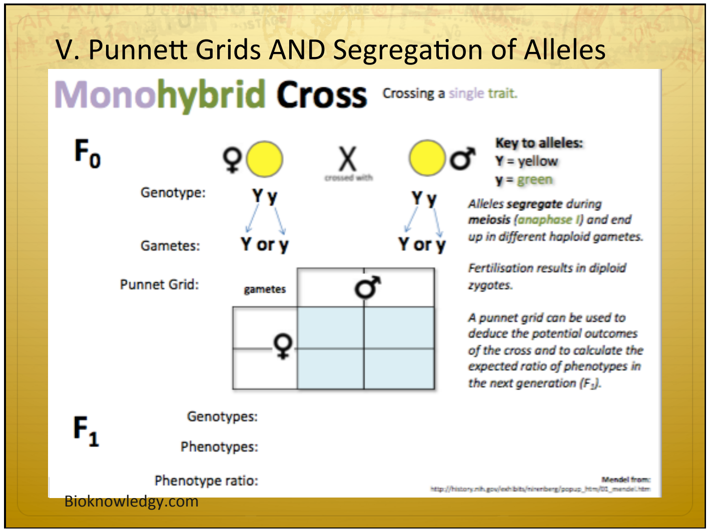
V. Punnett Grids AND Segregation of Alleles
Monohybrid Cross
Crossing a single trait.
F0
X
crossed with
Key to alleles:
Y = yellow
y = green
Genotype:
Y y
Y y
Gametes:
Y or y
Y or y
Alleles segregate during meiosis (anaphase I) and end up in different haploid gametes.
Fertilisation results in diploid zygotes.
Punnet Grid:
gametes
A punnet grid can be used to deduce the potential outcomes of the cross and to calculate the expected ratio of phenotypes in the next generation (F1).
F1
Genotypes:
Phenotypes:
Phenotype ratio:
Mendel from:
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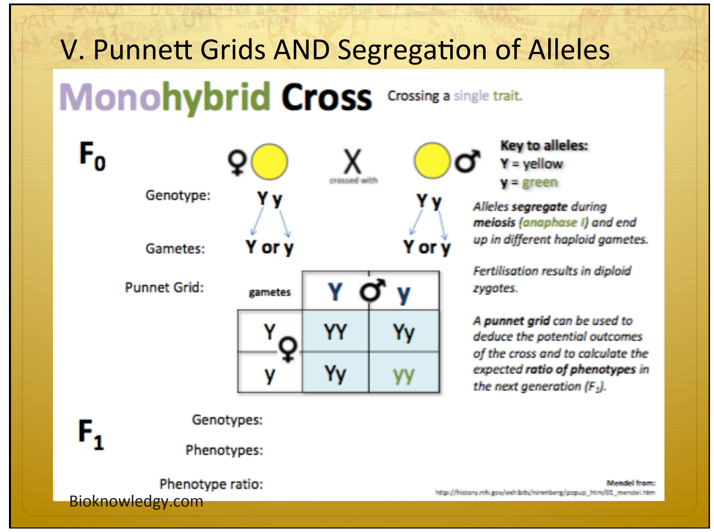
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Fertilisation results in diploid zygotes.
Punnet Grid:
gametes
Y y
Y YY Yy
y Yy yy
A punnet grid can be used to deduce the potential outcomes of the cross and to calculate the expected ratio of phenotypes in the next generation (F1).
F1
Genotypes:
Phenotypes:
Phenotype ratio:
Mendel from:
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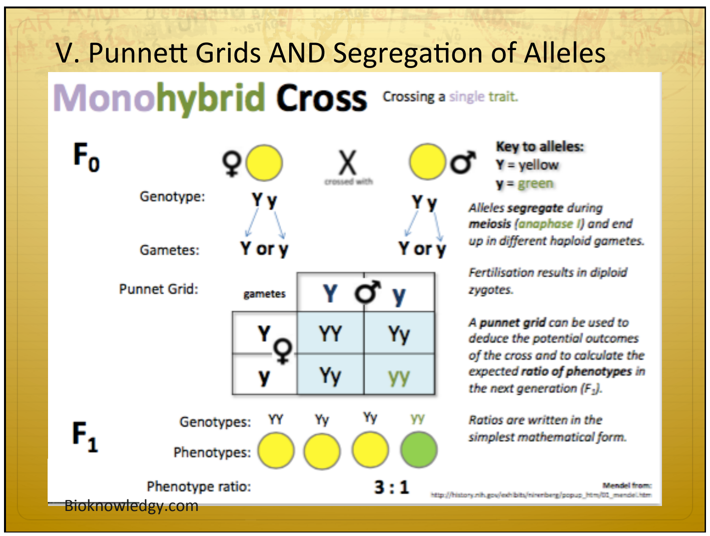
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Fertilisation results in diploid zygotes.
Punnet Grid:
gametes
Y y
Y YY Yy
y Yy yy
A punnet grid can be used to deduce the potential outcomes of the cross and to calculate the expected ratio of phenotypes in the next generation (F1).
F1
Genotypes: YY Yy Yy yy
Phenotypes:
Ratios are written in the simplest mathematical form.
Phenotype ratio: 3 : 1
Mendel from:
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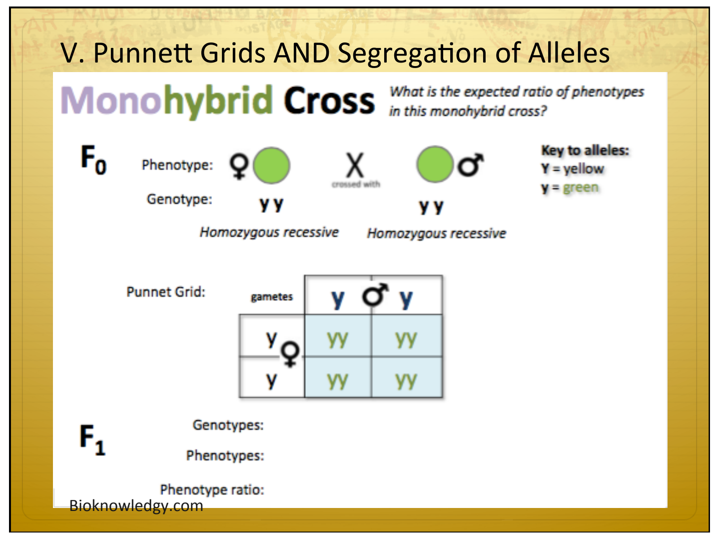
V. Punnett Grids AND Segregation of Alleles
Monohybrid Cross
What is the expected ratio of phenotypes in this monohybrid cross?
F0
Phenotype:
X
crossed with
Key to alleles:
Y = yellow
y = green
Genotype:
y y
y y
Homozygous recessive
Homozygous recessive
Punnet Grid:
gametes
y y
y yy yy
y yy yy
F1
Genotypes:
Phenotypes:
Phenotype ratio:
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V. Punnett Grids AND Segregation of Alleles

Monohybrid Cross

What is the expected ratio of phenotypes in this monohybrid cross?

F_0 Phenotype: ♀ (yellow) X (crossed with) (yellow) ♂

Genotype: Y Y — *Homozygous dominant*; Y y — *Heterozygous*

Key to alleles: Y = yellow; y = green

Punnet Grid:

gametes	Y	y
Y	YY	Yy
Y	YY	Yy

F_1 Genotypes:

Phenotypes:

Phenotype ratio:

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VI. Chi-squared Tests Are Used to Determine Whether the Difference Between an Observed and Expected Frequency Distribution is Statistically Significant

A. Used to test whether an observed frequency is significantly different to an expected frequency.

1. Null Hypothesis (H_0) = there is no significant difference between the observed and expected results
2. Alternate Hypothesis (H_1) = there is a significant difference between the observed and expected results

First calculate the value of X^2

	Collected data: Observed (O)	Collected data: Expected (E)	Calculating χ^2: (O - E)	Calculating χ^2: $(O-E)^2$	$\frac{(O-E)^2}{E}$
yellow					
green					
Total:				Total (χ^2) =	

And then compare it with the critical value at the desired level of certainty

Critical Values table

D.F. (n-1)	p (certainty) 0.1 (90%)	0.05 (95%)	0.01 (99%)
1	2.70554	3.84146	6.6349
2	4.60517	5.99146	9.21034
3	6.25139	7.81473	11.34487
4	7.77944	9.48773	13.2767
5	9.23636	11.0705	15.08627
6	10.64464	12.59159	16.81189
7	12.01704	14.06714	18.47531
8	13.36157	15.50731	20.09024
9	14.68366	16.91898	21.66599
10	15.98718	18.30704	23.20925

(how sure do you want to be?)

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VI. Chi-squared Tests Are Used to Determine Whether the Difference Between an Observed and Expected Frequency Distribution is Statistically Significant

Calculating X^2

probability x sample size
e.g. 0.75 x 50 = 37.5

actual number recorded → Observed (O); expected number → Expected (E); squaring (O-E) factors out any negatives

possible phenotypes	Observed (O)	Expected (E)	(O - E)	$(O-E)^2$	$\frac{(O-E)^2}{E}$
yellow					
green					
Total:	50	50		Total (χ^2) =	

← add them up!

These must be the same (represent total sample size)

keep this value to compare to the critical values table → X^2 = ______

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VI. Chi-squared Tests Are Used to Determine Whether the Difference Between an Observed and Expected Frequency Distribution is Statistically Significant

Do we accept or reject H_0?

X^2 = ______

Degrees of Freedom = no. possible outcomes - 1

2 possibilities (yellow or green)
∴ d.f. = 1

Critical Values table

D.F. (n-1)	p (certainty) 0.1 (90%)	0.05 (95%)	0.01 (99%)
1	2.70554	3.84146	6.6349
2	4.60517	5.99146	9.21034
3	6.25139	7.81473	11.34487
4	7.77944	9.48773	13.2767
5	9.23636	11.0705	15.08627

In Biology, we normally aim for p=0.05 (95% confident)

Compare X^2 with the critical value:

If χ^2 > C.V. we **reject** the null hypothesis *(we are 95% sure that the observed results are different from the expected results)*

If χ^2 < C.V. we **accept** the null hypothesis *(we are 95% sure that the observed results are the same as the expected results)*

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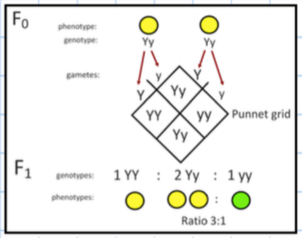
VI. Chi-squared Tests Are Used to Determine Whether the Difference Between an Observed and Expected Frequency Distribution is Statistically Significant

Worked Example: Mendel's Peas

In a cross between two heterozygous peas, we expect a 3:1 yellow:green ratio in the offspring, as shown in the punnet grid.

We can test this experimentally and use the to determine whether the results collected differ significantly from the expected ratio.

Null hypothesis (H_0):
"there is no significant difference between observed and expected results"



Sample size = 50 (50 offspring will be collected - modeled as coin flips)

A ratio of 3:1 can also be expressed in terms of probability:

There is a 0.75 chance of the offspring being yellow
0.25 chance of the offspring being green.

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VI. Chi-squared Tests Are Used to Determine Whether the Difference Between an Observed and Expected Frequency Distribution is Statistically Significant

Results:

Total Observed: 35 15

Calculating χ^2

	Collected data		Calculating χ^2		
	Observed (O)	Expected (E)	(O - E)	$(O - E)^2$	$\frac{(O-E)^2}{E}$
yellow	35	37.5*			
green	15	12.5#			
Total:	50	50		Total (χ^2) =	

* Expected result for Yellow = 0.75 x 50 = 37.5

\# Expected result for green = 0.25 x 50 = 12.5

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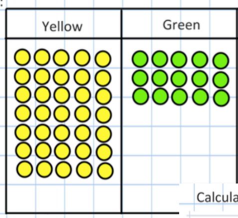
VI. Chi-squared Tests Are Used to Determine Whether the Difference Between an Observed and Expected Frequency Distribution is Statistically Significant

Results:



Total Observed: 35 15

Calculating χ^2

	Collected data		Calculating χ^2		
	Observed (O)	Expected (E)	(O - E)	$(O - E)^2$	$\frac{(O-E)^2}{E}$
yellow	35	37.5*	-2.5	6.25	0.1667
green	15	12.5#	2.5	6.25	0.5
Total:	50	50		Total (χ^2) =	0.6667

squaring (O-E) factors out negative values

X^2

* Expect

\# Expect

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VI. Chi-squared Tests Are Used to Determine Whether the Difference Between an Observed and Expected Frequency Distribution is Statistically Significant

Do we accept or reject H_0?

$X^2 = 0.6667$

2 possibilities (yellow or green)
∴ d.f. = 1

Critical Values table

D.F. (n-1)	p (certainty) (how sure do you want to be?) 0.1 (90%)	0.05 (95%)	0.01 (99%)
1	2.70554	3.84146	6.6349
2	4.60517	5.99146	9.21034
3	6.25139	7.81473	11.34487
4	7.77944	9.48773	13.2767
5	9.23636	11.0705	15.08627

Compare X^2 with the critical value:

0.6667 < 3.84146

∴

we accept the null hypothesis
(we are 95% sure that the observed results are the same as the expected results)

The data collected in this example DO fit the ration 3:1 yellow:green.

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VII. Test Cross

Test Cross *Used to **determine the genotype** of an **unknown** individual. The unknown is crossed with a known **homozygous recessive**.*

F_0 Phenotype: ♀ X (crossed with) ♂

Genotype: R ? (*unknown*) r r (*Homozygous recessive*)

Key to alleles:
R = Red flower
r = white

Possible outcomes:

F_1 Phenotypes:

Unknown parent = RR

gametes	♂	
♀		

Unknown parent = Rr

gametes	♂	
♀		

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VII. Test Cross

Test Cross *Used to **determine the genotype** of an **unknown** individual. The unknown is crossed with a known **homozygous recessive**.*

F_0 Phenotype: ♀ X (crossed with) ♂

Genotype: R ? (*unknown*) r r (*Homozygous recessive*)

Key to alleles:
R = Red flower
r = white

Possible outcomes:

F_1 Phenotypes: **All red** / **Some white, some red**

Unknown parent = RR

gametes ♀ \ ♂	r	r
R	Rr	Rr
R	Rr	Rr

Unknown parent = Rr

gametes ♀ \ ♂	r	r
R	Rr	Rr
r	rr	rr

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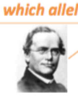
VIII. Punnett Squares for Dihybrid Traits (10.2)

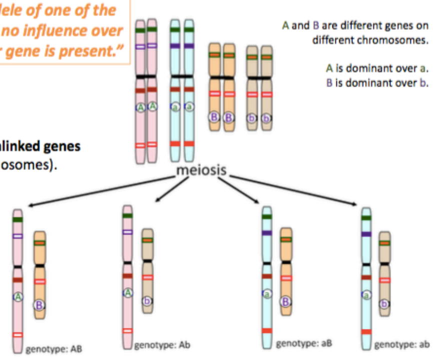
Review: 10.1.U7 Independent assortment of genes is due to the random orientation of pairs of homologous chromosomes in meiosis I.

Mendel's Law of Independent Assortment

"The presence of an allele of one of the genes in a gamete has no influence over which allele of another gene is present."



This only holds true for **unlinked genes** (genes on different chromosomes).



All four genotypes are possible therefore **10.2.U2 Unlinked genes segregate independently as a result of meiosis.**

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VIII. Punnett Squares for Dihybrid Traits (10.2)

Dihybrid Crosses *Consider two traits, each carried on separate chromosomes (the genes are unlinked).*

In this example of *Lathyrus odoratus* (sweet pea), we consider two traits: pea colour and pea surface.

What is the predicted phenotype ratio for a cross between two pea plants which are heterozygous at both loci?

Key to alleles:
Y = yellow
y = green
S = smooth
s = rough

F_0 Phenotype: *Heterozygous at both loci* X (crossed with) *Heterozygous at both loci*

Genotype:

Punnet Grid:

gametes				

F_1

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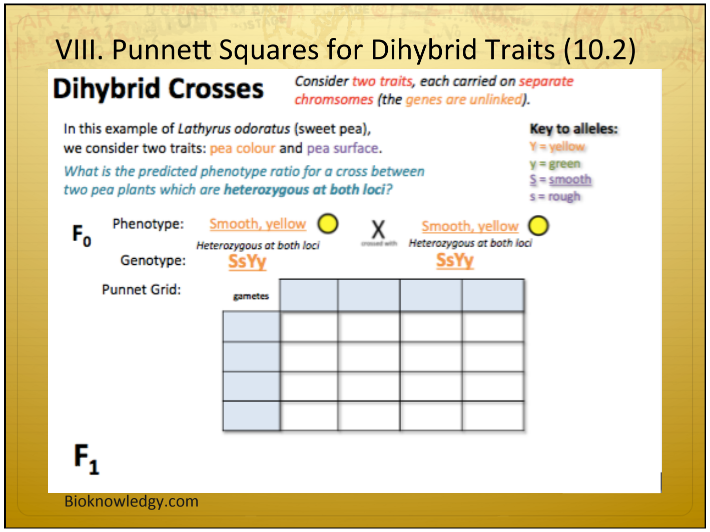
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Dihybrid Crosses
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What is the predicted phenotype ratio for a cross between two pea plants which are heterozygous at both loci?
Key to alleles:
Y = yellow
y = green
S = smooth
s = rough
F0
Phenotype: Smooth, yellow X Smooth, yellow
Heterozygous at both loci
Heterozygous at both loci
Genotype: SsYy SsYy
Punnet Grid:
gametes
F1
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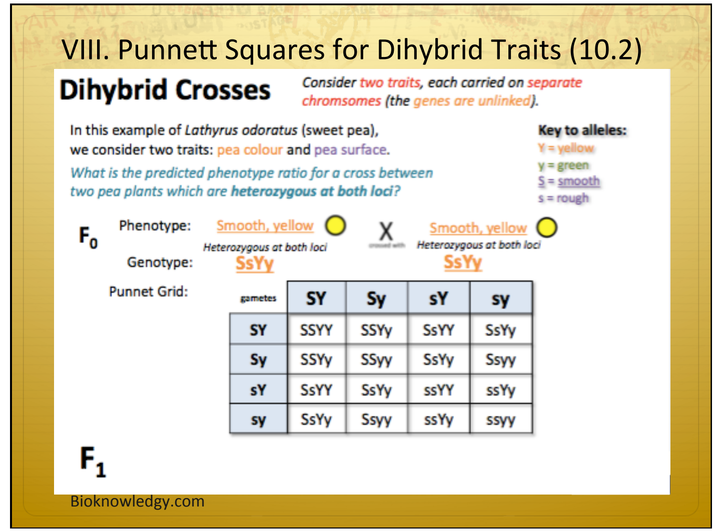
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Key to alleles:
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F0
Phenotype: Smooth, yellow X Smooth, yellow
Heterozygous at both loci
Heterozygous at both loci
Genotype: SsYy SsYy
Punnet Grid:
gametes SY Sy sY sy
SY SSYY SSYy SsYY SsYy
Sy SSYy SSyy SsYy Ssyy
sY SsYY SsYy ssYY ssYy
sy SsYy Ssyy ssYy ssyy
F1
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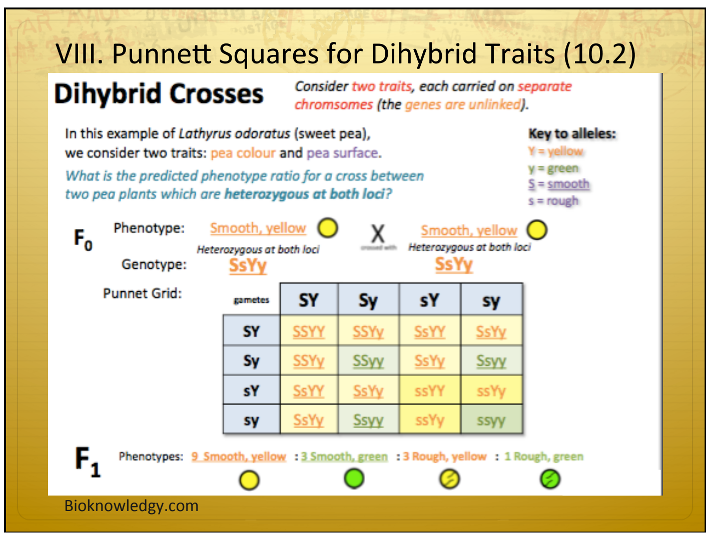
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Key to alleles:
Y = yellow
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S = smooth
s = rough
F0
Phenotype: Smooth, yellow X Smooth, yellow
Heterozygous at both loci
Heterozygous at both loci
Genotype: SsYy SsYy
Punnet Grid:
gametes SY Sy sY sy
SY SSYY SSYy SsYY SsYy
Sy SSYy SSyy SsYy Ssyy
sY SsYY SsYy ssYY ssYy
sy SsYy Ssyy ssYy ssyy
F1
Phenotypes: 9 Smooth, yellow : 3 Smooth, green : 3 Rough, yellow : 1 Rough, green
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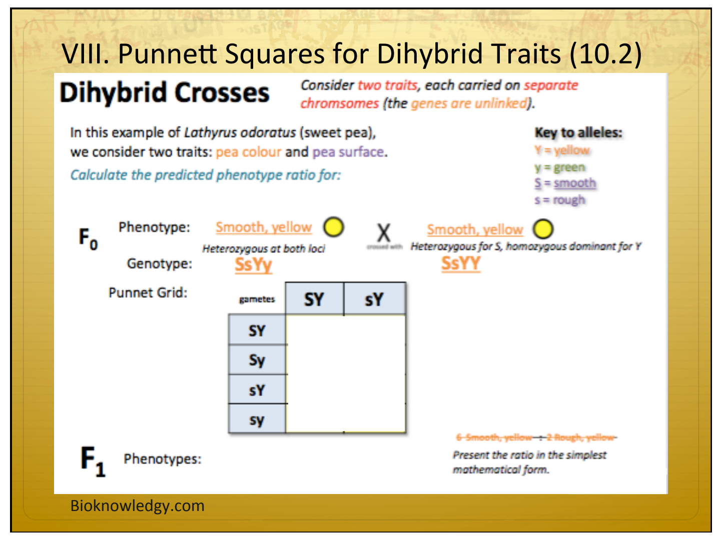
VIII. Punnett Squares for Dihybrid Traits (10.2)
Dihybrid Crosses
Consider two traits, each carried on separate chromsomes (the genes are unlinked).
In this example of Lathyrus odoratus (sweet pea), we consider two traits: pea colour and pea surface.
Calculate the predicted phenotype ratio for:
Key to alleles:
Y = yellow
y = green
S = smooth
s = rough
F0
Phenotype: Smooth, yellow X Smooth, yellow
Heterozygous at both loci
Heterozygous for S, homozygous dominant for Y
Genotype: SsYy SsYY
Punnet Grid:
gametes SY sY
SY
Sy
sY
sy
F1
Phenotypes:
6 Smooth, yellow : 2 Rough, yellow
Present the ratio in the simplest mathematical form.
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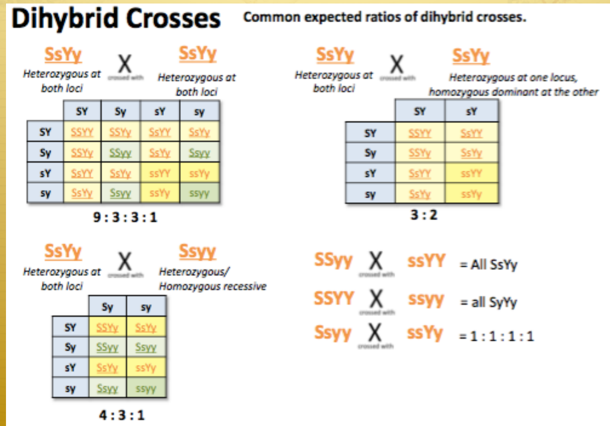
VIII. Punnett Squares for Dihybrid Traits (10.2)



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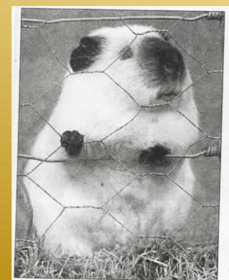
VIII. Punnett Squares for Dihybrid Traits (10.2)

A. Completion and analysis of Punnett squares for dihybrid traits AND Calculation of the predicted genotypic and phenotypic ratio of offspring of dihybrid crosses involving unlinked autosomal genes-



Pen pal: Sooty after his one-night stand

Guinea pig harem says 'hello Sooty'

A GUINEA pig called Sooty had a night to remember after escaping from his pen and tunnelling into a cage of 24 females.

He romanced each of them in turn and was yesterday the proud father of 43 offspring.

Staff at Little Friend's Farm in Pontypridd, South Wales, have now secured Sooty's pen — and begun looking for homes for the guinea pigs.

His owner, Carol Feehan, 42, said: "I'm sure a lot of men will be looking at Sooty with envy.

"We knew that he had gone missing after wriggling through the bars of his cage.

"We looked for him everywhere but never thought of checking the pen where we keep 24 females. We did a head count and found 25 guinea pigs — Sooty was fast asleep in the corner.

"He was absolutely shattered. We put him back in his cage and he slept for two days."

Key to alleles*:
C = color c = albino
A = agouti a = black
R = round ears
r = pointy ears
L = long whiskers
l = short whiskers
S = soft fur
s = rough fur
N = sharp nails n = smooth nails

Funny Shit

* C and A genes are real. The rest are made up for this story.

VIII. Punnett Squares for Dihybrid Traits (10.2)

Sooty has soft fur and sharp nails.
In one of his matings with a rough-furred, smooth-nailed female, the following guinea piglets are produced:
6 x rough fur, sharp nails; 3 x soft fur sharp nails.

Deduce Sooty's Genotype

F_0 Phenotype: ♀ *Rough fur, smooth nails* X ♂ *Soft fur, sharp nails*

Genotype:

Punnet Grid:

F_1 Phenotypes:

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Deduce Sooty's Genotype

F_0 Phenotype: ♀ *Rough fur, smooth nails* X ♂ *Soft fur, sharp nails*

Genotype: ssnn

Punnet Grid:

Possible Gametes				
All sn				

F_1 Phenotypes:

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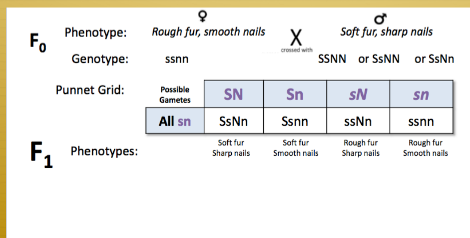

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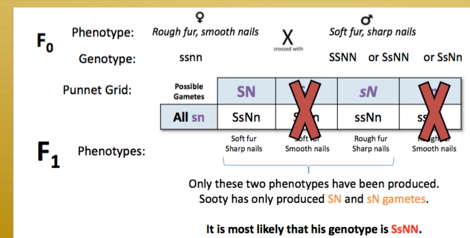

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VIII. Punnett Squares for Dihybrid Traits (10.2)

DBQ: Using the Chi-Square Test
Warren and Hutt (1936) test-crossed a double heterozygote for two pairs of alleles in hens: one for the presence (Cr) or absence (cr) of a crest and one for white (I) or non-white (i) plumage.
For their F_1 cross, there were a total of 754 offspring:

337 were white, crested;
337 were non-white, non-crested;
34 were non-white, crested;
46 were white, non-crested

1. Construct a contingency table of observed values.
2. Calculate the expected values assuming independent assortment.
3. Determine the number of degrees of freedom.
4. Find the critical region for chi-square at a significance level of 5%.
5. Calculate chi-squared.
6. State the two hypotheses H_0 and H_1 and evaluate them using the calculated value for chi-squared.

VIII. Punnett Squares for Dihybrid Traits (10.2)

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	O	E	O – E	$(O - E)^2$	$\frac{(O - E)^2}{E}$
white, crested					
non-white, non-crested					
non-white, crested					
white, non-crested					
Total				X^2 =	

VIII. Punnett Squares for Dihybrid Traits (10.2)

3. Determine the number of degrees of freedom.
   df = categories – n =

4. Find the critical region for chi-square at a significance level of 5%.
   p =

5. Calculate chi-squared.
   $X^2 = 468.3$

6. State the two hypotheses H_0 and H_1 and evaluate them using the calculated value for chi-squared.

df	0.05	0.01
1	3.84	6.64
2	5.99	9.21
3	7.82	11.34
4	9.49	13.28
5	11.07	15.09
6	12.59	16.81
7	14.07	18.48
8	15.51	20.09
9	16.92	21.67
10	18.31	23.21
11	19.68	24.72
12	21.03	26.22
13	22.36	27.69
14	23.68	29.14
15	25.00	30.58