Introduction to Genetics

Biology Honors

Miller Biology Ch. 11

SC.912.L.16.1-2; SC.912.L.16.16-17

Genetics

The study of how characteristics are transmitted from parents to offspring

Inheritance

the transmission of genetic information from generation to generation

Determines:
Blood type, eye color, hair color, skin color, and so much more!

Gregor Mendel

- Born in 1822
- 1st scientist of his time to obtain successful results from inheritance studies
  - Due to his methods – quantitative approach to analyze results
  - He formulated hypotheses to explain his results and experimental tests to support them
  - Many tests are still used today

Gregor Mendel: Father of Genetics

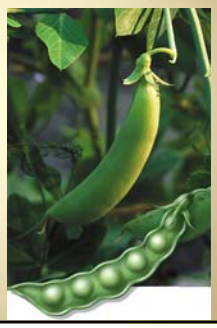
POSTULATES:

- Unit factors (genes) occur in pairs
- Dominance/ Recessiveness
- Segregation of alleles into gametes in equal frequencies
- Independent Assortment of alleles from different gene pairs into gametes

Patterns of Inheritance

Mendel made three key decisions in his experiments.

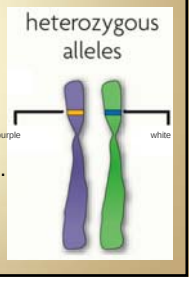
- use of purebred plants
- control over breeding
- observation of seven "either-or" traits



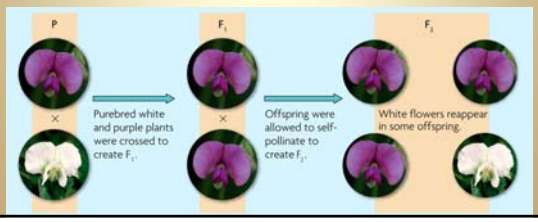
Mendel's Experiments

Mendel drew three important conclusions
- Traits are inherited as discrete units.
- Organisms inherit two copies of each gene, one from each parent.
- The two copies segregate during gamete formation.

The last two conclusions are called **the law of segregation**.



- Mendel allowed the resulting plants to self-pollinate.
  - Among the F_1 generation, all plants had purple flowers
  - F_1 plants are all heterozygous
  - Among the F_2 generation, some plants had purple flowers and some had white



Mendel's Conclusions

Mendel's Results and Conclusions

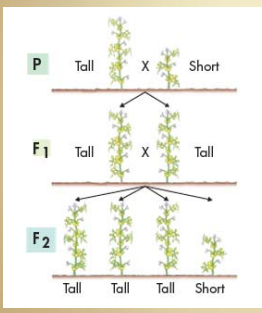
Recessive and Dominant Traits

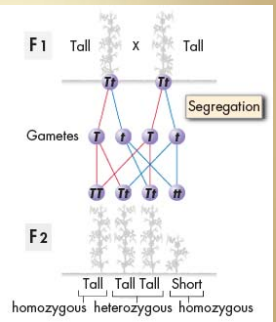
- Mendel concluded that inherited characteristics are controlled by factors that occur in pairs.
- Through experimentation Mendel found one factor in a pair masked the other
  - The trait that masked the other was called the **dominant** trait.
  - The trait that was masked was called the **recessive** trait.

Mendel's Results and Conclusions

- **The Law of Segregation**
  - a pair of factors is segregated, or separated, during the formation of gametes
  - Meiosis

Segregation

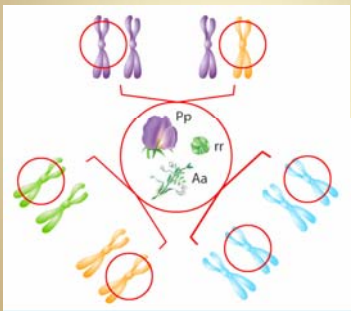




Mendel's Results and Conclusions

- **The Law of Independent Assortment**
  - …states that factors for individual characteristics are distributed to gametes **independent of one another**.
  - …is observed only for genes that are located on separate chromosomes or are far apart on the same chromosome.

Independent Assortment

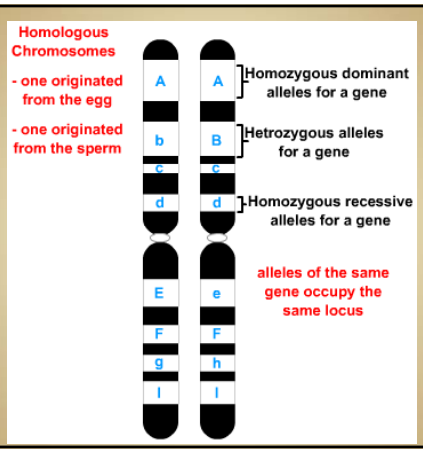


Factors affecting flower color, seed texture and plant height are located on different chromosomes

- independent of one another

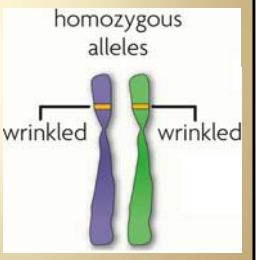
Support for Mendel's Conclusions

- The ***factors*** that Mendel studied are **alleles,** or alternative forms of a gene.
- One allele for each trait is passed from each parent to the offspring.



The same gene can have many versions

- A gene is a piece of DNA that directs a cell to make a certain protein.
- Each gene has a locus, a specific position on a pair of homologous chromosomes.

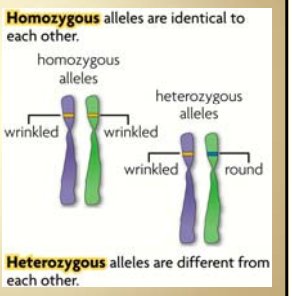


• An allele is any alternative form of a gene occurring at a specific locus on a chromosome.

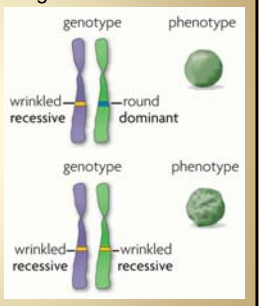
Each parent donates one allele for every gene.

Homozygous - two alleles that are the same at a specific locus.

Heterozygous - two alleles that are different at a specific locus.



- Alleles can be represented using letters.
  - Dominant alleles are represented by uppercase letters; recessive alleles by lowercase letters.
  - A dominant allele is expressed as a phenotype when at least one allele is dominant.
  - A recessive allele is expressed as a phenotype only when two copies are present.



Genotype and Phenotype

The **genotype** is the genetic makeup of an organism.
- Aa or aa or AA
  - Dominant alleles are represented by uppercase letters;
  - recessive alleles by lowercase letters.

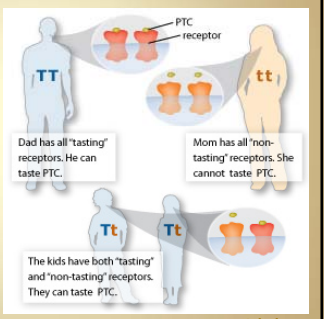
The **phenotype** is the appearance of an organism.
- What you see
  - Color, size, shape, etc.

PTC Taste Test

• The ability to taste PTC shows a dominant pattern of inheritance.

• A single copy of a tasting allele (**T**) conveys the ability to taste PTC.

• Non-tasters have two copies of a non-tasting allele (**t**).

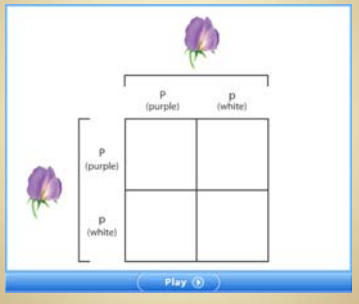


Link

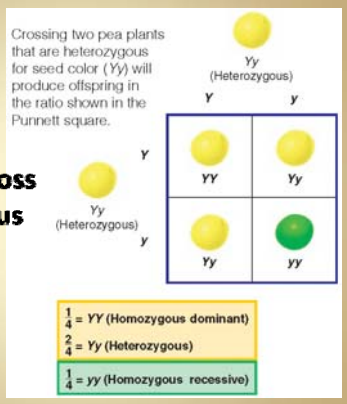
Predicting Results of Monohybrid Crosses

- A **Punnett square** can be used to predict the outcome of genetic crosses.
- A cross in which <u>one characteristic</u> is tracked is a **monohybrid cross.**

Punnett Square with Homozygous Cross



Monohybrid Cross of Heterozygous Plants



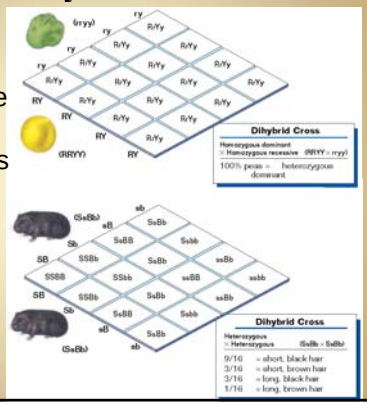
Predicting Results of Monohybrid Crosses

- **Testcross**
  - Example: an individual of unknown genotype is crossed with a homozygous recessive individual
  - Testcross can be used to determine the genotype of an individual whose phenotype expresses the dominant trait

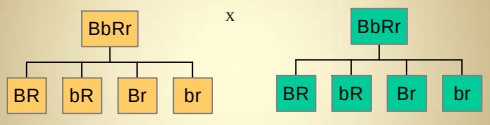
Testcross

Dihybrid Cross

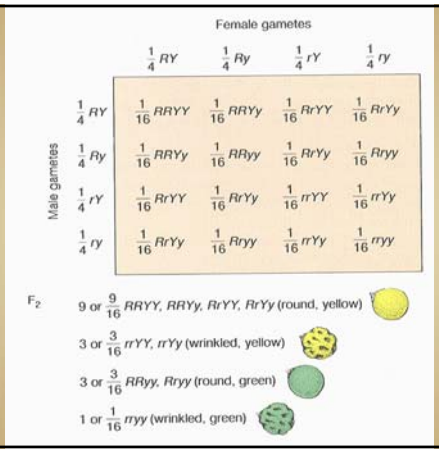
A cross where two characteristics are tracked



Dihybrid Ratios

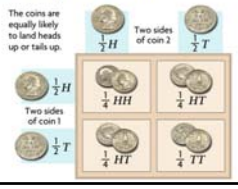


F_2 Yields 9:3:3:1 Ratio

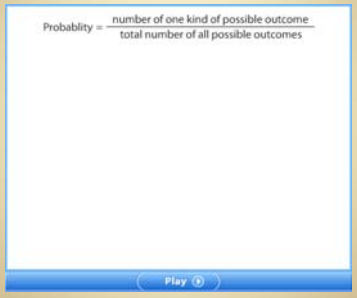


Probability

- the likelihood that a specific event will occur.
- may be expressed as a decimal, a percentage, or a fraction.
  - Probability predicts an average number of occurrences, not an exact number of occurrences.
  - Probability = $\frac{\text{number of ways a specific event can occur}}{\text{number of total possible outcomes}}$
  - Probability applies to random events such as meiosis and fertilization.



Calculating Probability

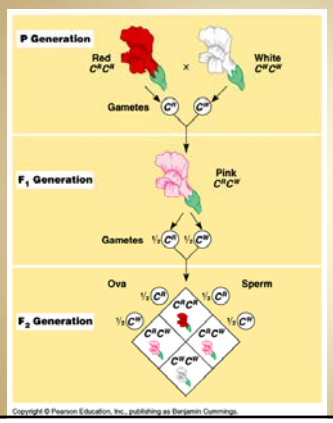


Predicting Results of Monohybrid Crosses

- **Complete dominance**
  - occurs when heterozygous individuals and dominant homozygous individuals are indistinguishable in phenotype.
  - Tt and TT are the same in appearance

- **Incomplete dominance**
  - occurs when two or more alleles influence the phenotype and results in a phenotypic **intermediate** between the dominant and recessive trait.
    - Heterozygous = different color than dominant
      - Red x White = Pink

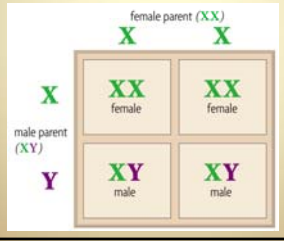
Incomplete dominance



Situation where one allele is <u>not</u> completely dominant over another

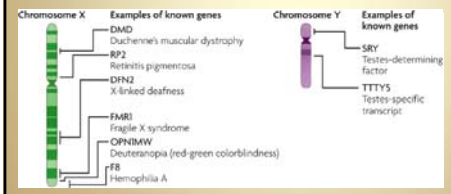
Sex linked traits

- Genes on sex chromosomes are called sex-linked genes.
  - Y chromosome genes in mammals are responsible for male characteristics.
  - X chromosome genes in mammals affect many traits.



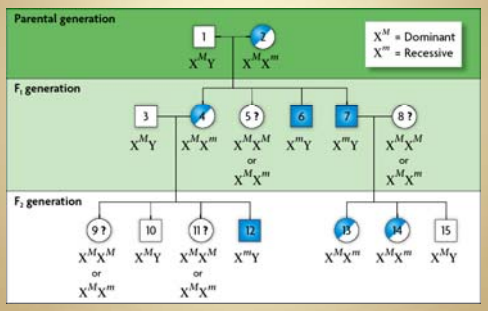
Females can carry sex-linked genetic disorders

- Males (XY) express all of their sex linked genes.
- Expression of the disorder depends on which parent carries the allele and the sex of the child.



- Male mammals have an XY genotype.
  - All of a male's sex-linked genes are expressed.
  - Males have no second copies of sex-linked genes.

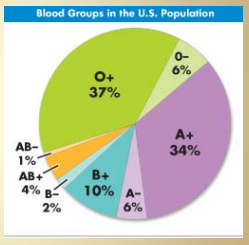
- If the phenotype is more common in males, the gene is likely sex-linked.



- Female mammals have an XX genotype.
  - Expression of sex-linked genes is similar to autosomal genes in females.
  - X chromosome inactivation randomly "turns off" one X chromosome.

Multiple Alleles

- A single gene can have many possible alleles.
- A gene with more than two alleles is said to have multiple alleles.
- Many genes have multiple alleles, including the human genes for blood type. This chart shows the percentage of the U.S. population that shares each blood group.



- **Codominance** occurs when both alleles for a gene are expressed in a heterozygous offspring.
  - *Blood type*

father \ mother	A	B	O
A	AA	AB	AO
B	BA	BB	BO
O	OA	OB	OO

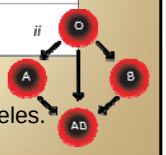
alleles	blood type
A+A	A
A+O	A
A+B	AB
B+B	B
B+O	B
O+O	O

The **A and B alleles** code for enzymes that produce the type A and B antigens respectively.

A third version of this gene, the **O allele**, codes for a protein that is not functional and does not produce surface molecules.

- **Codominant** alleles will both be completely expressed
  - Codominant alleles are neither dominant nor recessive.
  - The ABO blood types result from codominant alleles.

PHENOTYPE (BLOOD TYPE)		GENOTYPES
A	antigen A	I^AI^A or I^Ai
B	antigen B	I^BI^B or I^Bi
AB	both antigens	I^AI^B
O	no antigens	ii



- Many genes have more than two alleles.

Polygenic Traits

- An interaction of two or more genes.
- Shows a wide range of phenotypes
- Skin color in humans – more than 4 genes

Order of dominance: brown > green > blue.

GENE NAME	DOMINANT ALLELE	RECESSIVE ALLELE
BEY1	brown	blue
BEY2	brown	blue
GEY	green	blue

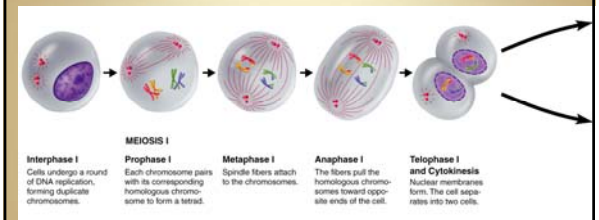
Epistatisis

- Polygenic trait
- Interferes with the expression of other genes
- Mice: 5 different genes
  - 2 general color
  - 1 effects of shading
  - 1 spots
  - 1 overshadows the expression of all others (albinism)

The effect of environment on phenotype

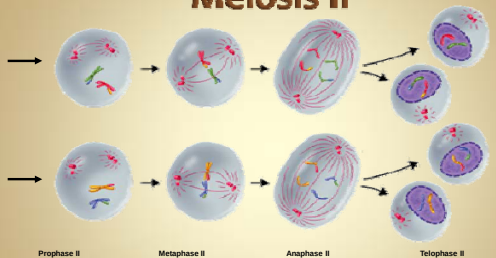
Acidity of soil changes the color of the hydrangea

Meiosis I



Crossing over in prophase I

Meiosis II



Meiosis I results in two haploid (N) daughter cells, each with half the number of chromosomes as the original.

The chromosomes line up in a similar way to the metaphase stage of mitosis.

The sister chromatids separate and move toward opposite ends of the cell.

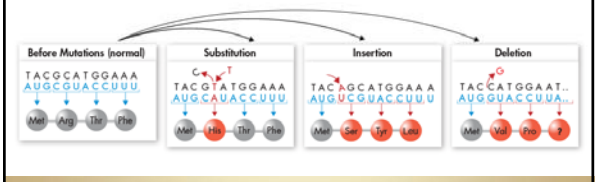
Meiosis II results in four haploid (N) daughter cells.

Mutation

- Change in a gene or chromosome

Gene Mutations

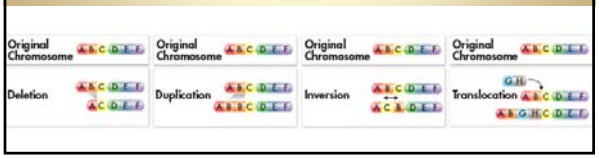
Point mutations include substitutions, insertions, and deletions



Chromosomal Mutations

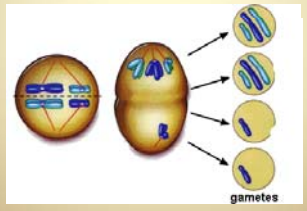
- changes in the number or structure of chromosomes
- can change the location of genes on chromosomes and can even change the number of copies of some genes

4 types of chromosomal mutations:
deletion, duplication, inversion, and translocation



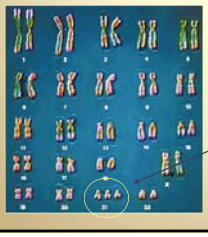
Nondisjunction

- The failure of homologous chromosomes to separate properly during meiosis



Trisomy

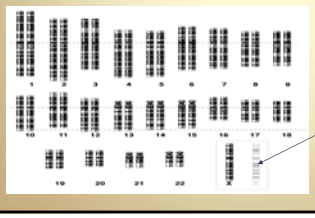
- A gamete with an extra chromosome is fertilized by a normal gamete
- Zygote has an extra chromosome
- Ex. Down syndrome (Trisomy 21)



Extra chromosome

Monosomy

- A gamete with a missing chromosome is fertilized by a normal gamete
- Zygote lacks a chromosome
- Ex. human females with only a single X-chromosome



Missing chromosome

Mitosis vs. Meiosis

- Diploid daughter cells
  - Genetically identical
- 2n
- Somatic cells: body cells

- Haploid daughter cells
  - Genetically different
  - Reduction/division
- n
- Sex cells: sexual reproduction

See chart on pg. 326 Miller

Strands

SC.912.L.16.1
- Use Mendel's laws of segregation and independent assortment to analyze patterns of inheritance.

SC.912.L.16.2
- Discuss observed inheritance patterns caused by various modes of inheritance, including dominant, recessive, codominant, sex-linked, polygenic, and multiple alleles.

SC.912.L.16.16
- Describe the process of meiosis, including independent assortment and crossing over. Explain how reduction division results in the formation of haploid gametes or spores.

SC.912.L.16.17
- Compare and contrast mitosis and meiosis and relate to the processes of sexual and asexual reproduction and their consequences for genetic variation.

Resources

Genetic Science Learning Center. (2010). PTC: GENES AND BITTER TASTE. *Learn.Genetics*. Retrieved January 7, 2010, from http://learn.genetics.utah.edu/content/begin/traits/ptc/

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