

Introduction to Genetics

Pre-AICE Biology (S.III.3,4-5)

IGCSE Biology Ch. 22, 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

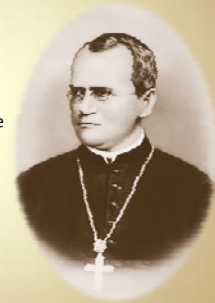
the transmission of genetic information from generation to generation

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

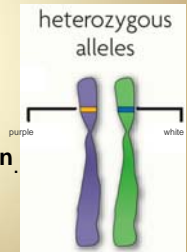


Mendel's Experiments

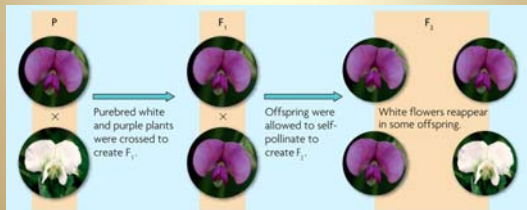


- Mendel drew three important conclusions
- Traits are inherited as discrete units.
 - Organisms inherit two copies of each gene, one from each parent.
 - The two copies segregate during gamete formation.

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

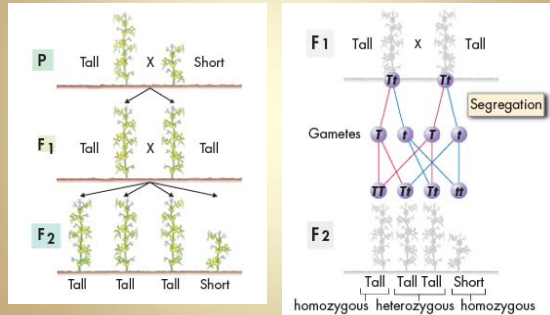
Recessive and Dominant Traits

- 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

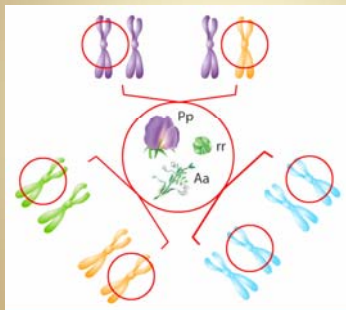
Segregation



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

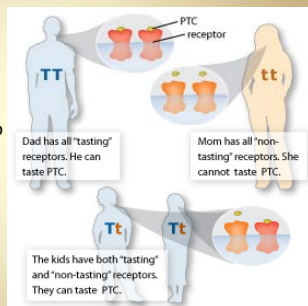
- independent of one another

Support for Mendel's Conclusions

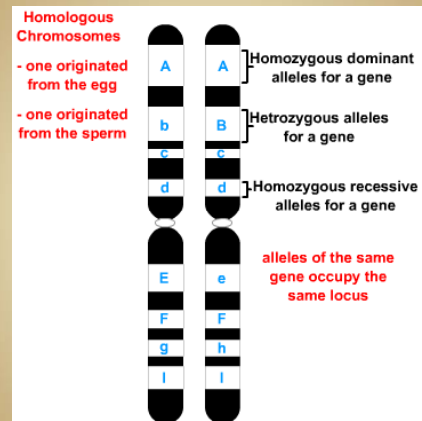
- The **factors** that Mendel studied are **alleles**, or alternative forms of a gene.
- One allele for each trait is passed from each parent to the offspring.

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).

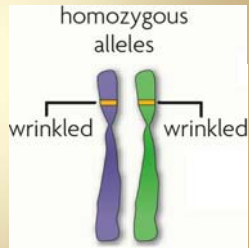


Link



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.



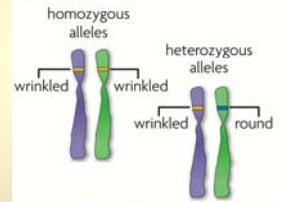
- An allele is any alternative form of a gene occurring at a specific locus on a chromosome.

Each parent donates one allele for every gene.

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

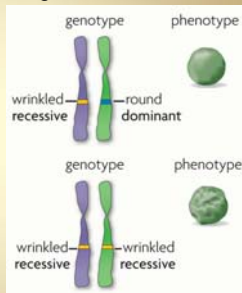
Heterozygous - two alleles that are different at a specific locus.

Homozygous alleles are identical to each other.



Heterozygous alleles are different from each other.

- 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.



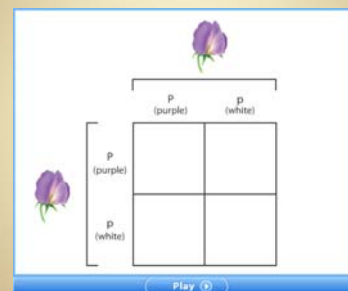
Genotype and Phenotype

- The **genotype** is the genetic makeup of an organism.
 - Aa or aa or AA
- The **phenotype** is the appearance of an organism.
 - What you see

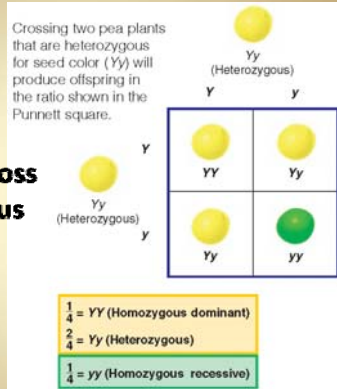
Predicting Results of Monohybrid Crosses

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

Punnett Square with Homozygous Cross



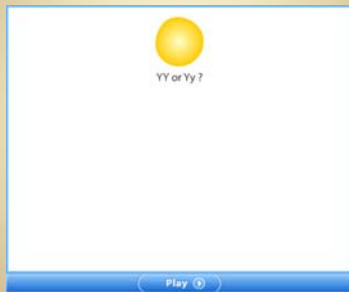
Monohybrid Cross of Heterozygous Plants



Predicting Results of Monohybrid Crosses

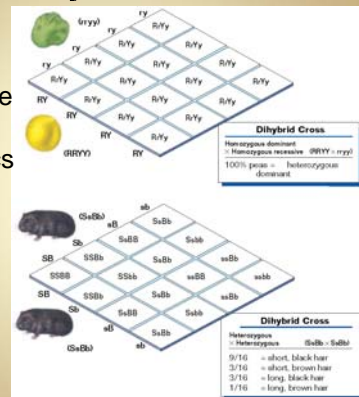
- **Testcross**
 - Example: an individual of unknown genotype is crossed with a homozygous recessive individual
- Testcross can be used to determine the genotype of an individual whose phenotype expresses the dominant trait

Testcross

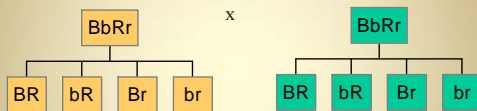


Dihybrid Cross

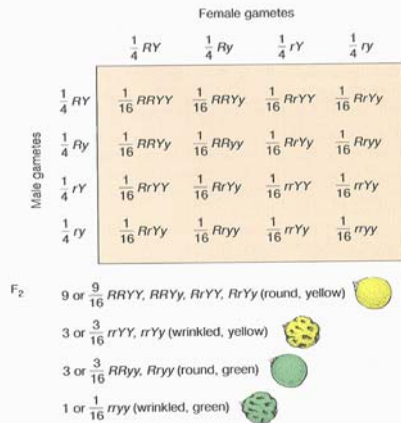
A cross where two characteristics are tracked



Dihybrid Ratios

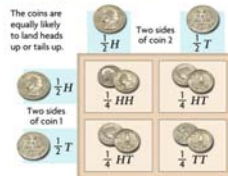


F₂ Yields 9:3:3:1 Ratio



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

$$\text{Probability} = \frac{\text{number of one kind of possible outcome}}{\text{total number of all possible outcomes}}$$

Play

Predicting Results of Monohybrid Crosses

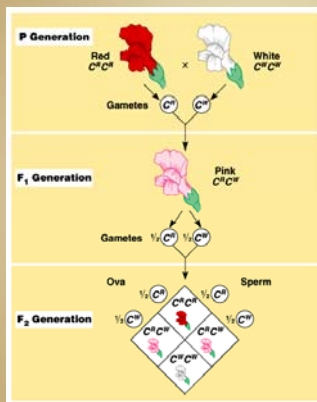
- **Complete dominance**
 - occurs when heterozygous individuals and dominant homozygous individuals are indistinguishable in phenotype.
 - Tt and TT are the same in appearance

- **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



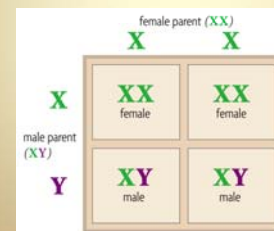
Incomplete dominance



Situation where one allele is not completely dominant over another

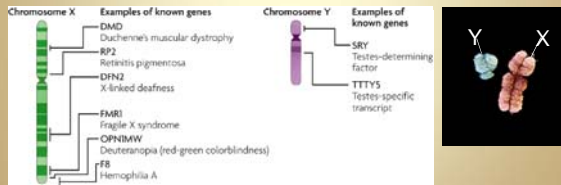
Sex linked traits

- 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.



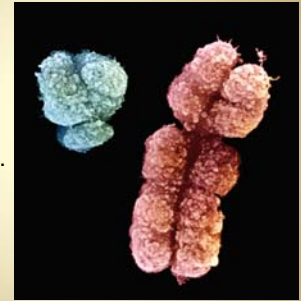
Females can carry sex-linked genetic disorders

- 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.

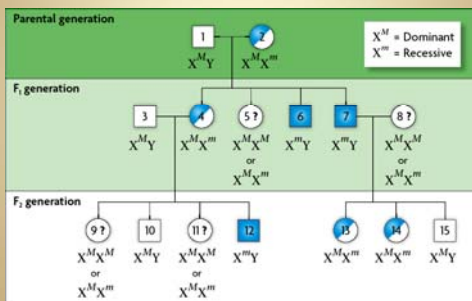


- Male mammals have an XY genotype.

- 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.



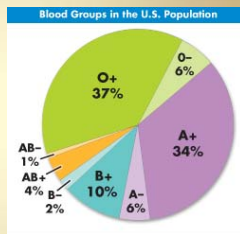
- Female mammals have an XX genotype.

- 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			alleles	blood type
	A	B	O		
A	AA	AB	AO	A+A = A	A+O = A
B	BA	BB	BO	A+B = AB	B+B = B
O	OA	OB	OO	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.

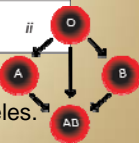
- Codominant alleles will both be completely expressed

– Codominant alleles are neither dominant nor recessive.

– The ABO blood types result from codominant alleles.

- Many genes have more than two alleles.

PHENOTYPE (BLOOD TYPE)	GENOTYPES
A antigen A	$I^A I^A$ or $I^A i$
B antigen B	$I^B I^B$ or $I^B i$
AB both antigens	$I^A I^B$
O no antigens	ii



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

Epistasis



- 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)

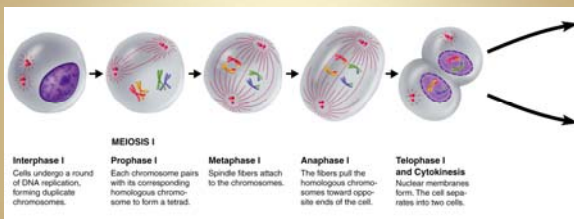
The effect of environment on phenotype

Acidity of soil changes the color of the hydrangea



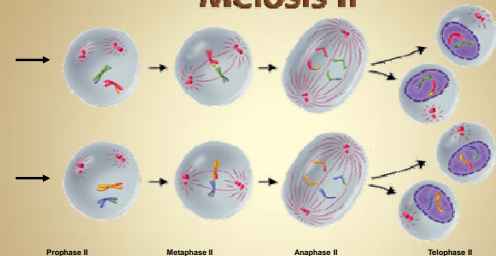
Benjamin Cummings

Meiosis I



Crossing over in prophase I

Meiosis II

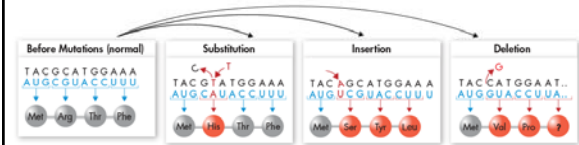


Mutation

- Change in a gene or chromosome

Gene Mutations

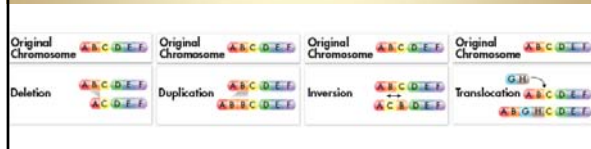
Point mutations include substitutions, insertions, and deletions



Chromosomal Mutations

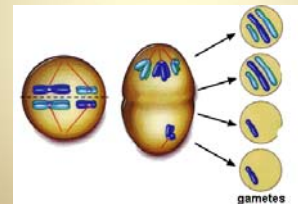
- changes in the number or structure of chromosomes
- can change the location of genes on chromosomes and can even change the number of copies of some genes

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



Nondisjunction

- The failure of homologous chromosomes to separate properly during meiosis



Trisomy

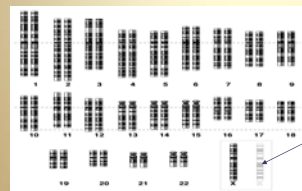
- A gamete with an extra chromosome is fertilized by a normal gamete
- Zygote has an extra chromosome
- Ex. Down syndrome (Trisomy 21)



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
- Haploid daughter cells
 - Genetically different
 - Reduction/division
- 2n
- n
- Somatic cells: body cells
- 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.

3-4 Monohybrid inheritance

Core

- **genotype** - genetic makeup of an organism in terms of the alleles present (e.g. Tt or GG)
- **phenotype** - the physical or other features of an organism due to both its genotype and its environment (e.g. tall plant or green seed)
- **homozygous** - having two identical alleles of a particular gene (e.g. TT or gg). Two identical homozygous individuals that breed together will be pure-breeding
- **heterozygous** - having two different alleles of a particular gene (e.g. Tt or Gg), not pure-breeding
- **dominant** - an allele that is expressed if it is present (e.g. T or G)
- **recessive** - an allele that is only expressed when there is no dominant allele of the gene present (e.g. t or g)

[Revision Checklist](#)

- Calculate and predict the results of monohybrid crosses involving 1 : 1 and 3 : 1 ratios

- Explain codominance by reference to the inheritance of ABO blood groups
 - phenotypes, A, B, AB and O blood groups
 - genotypes I^A, I^B, and I^O

3.5 Variation

Core

- State that continuous variation is influenced by genes and environment, resulting in a range of phenotypes between two extremes, e.g. height in humans
- State that discontinuous variation is caused by genes alone and results in a limited number of distinct phenotypes with no intermediates
 - e.g. A, B, AB and O blood groups in humans
- Define *mutation* as a change in a gene or chromosome
- Describe mutation as a source of variation, as shown by Down's syndrome
- Outline the effects of ionising radiation and chemicals on the rate of mutation

Resources

Genetic Science Learning Center. (2010). PTC: GENES AND BITTER TASTE.

Learn.Genetics. Retrieved January 7, 2010, from

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