

## Human Genetics

Gregor Mendel



1822 - 1884

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## Mendel's Laws of Heredity

- Law of Segregation
- Law of Independent Assortment

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Law of Segregation –Part 1:

### Alternative versions of genes account for variations in inherited characters

**ALLELES**- Alternative versions of the same gene

**GENOTYPE**- The set of alleles that an organism has for a trait

**PHENOTYPE**- The physical characteristics/traits that result from the genotype. Phenotype is always a combination between genes and environment.

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Law of Segregation –Part 2:

**An organism inherits two alleles for each trait, one from each parent**

**Homologous chromosomes:**

**Genotype:**  $PP$   $aa$   $Bb$

**Homozygous for the dominant allele**    **Homozygous for the recessive allele**    **Heterozygous**

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Law of Segregation- Part 3:

If the two alleles differ, then one, the **dominant allele**, is fully expressed in the organism's appearance; the other, the **recessive allele**, has no noticeable effect on the organism's appearance

Key:  
 E = Unattached earlobes (dominant allele)  
 e = Attached earlobes (recessive allele)

unattached earlobe    attached earlobe    unattached earlobe

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Law of Segregation - Part 4

**The two genes for each character segregate during gamete production**

**Figure 18.3 Gametogenesis.**  
 Because the pairs of chromosomes separate during meiosis, which occurs during gametogenesis, the gametes have only one allele for each trait.

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## Law of Independent Assortment

- the emergence of one trait will not effect the emergence of another
- More on this in lab...

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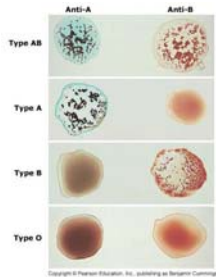
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## Beyond Mendel



**Co-dominance**- both alleles are expressed



**Incomplete Dominance**- intermediate forms occur

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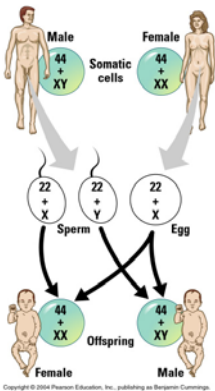
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## Sex-Linked Traits

The X and Y chromosomes carry genes for things besides gender.



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# How Did Mendel Work it Out?

He studied sexual reproduction in Pea plants




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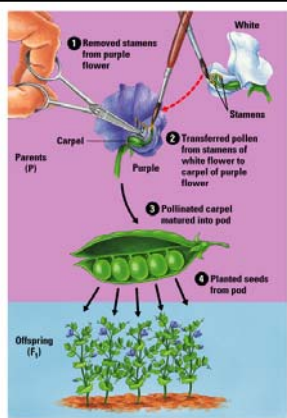
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How *Do* Plants Have Sex, anyway?



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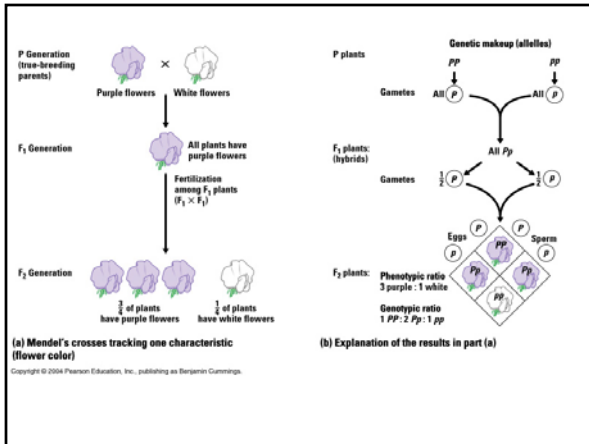
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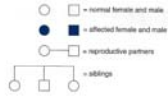
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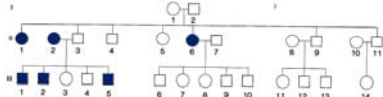
Figure 17.4 Pedigree chart symbols.



**Pedigree Chart Analyses**

For each of the following pedigree charts, determine how a genetic disorder is passed. Is the inheritance pattern autosomal dominant, autosomal recessive, or X-linked recessive? Also, decide the genotype of particular individuals in the chart. Remember that the genotype indicates the dominant and recessive alleles present and the phenotype is the actual physical appearance of the trait in the individual. A pedigree chart indicates the phenotype, and you can reason out the genotype.

1. Study the following pedigree chart:



- What is the inheritance pattern for this genetic disorder?
- What is the genotype of the following individuals? Use *A* for the dominant allele and *a* for the recessive allele.
  - Generation I, individual 1 \_\_\_\_\_
  - Generation II, individual 1 \_\_\_\_\_
  - Generation III, individual 8 \_\_\_\_\_

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