

# Biology 101

## Patterns of Inheritance

*Genetics* = science of heredity.

Heredity: passing of characteristics from parents to offspring.

Wildtype vs. Variant

### Roots of Genetics

Hippocrates and Aristotle

### The Father of Modern Genetics

- Began in 1860s with **Gregor Mendel**
- Discovered fundamental principles of genetics
- Parents pass on to offspring discrete heritable factors (genes)

### Terms to Know

Self-fertilize	Hybrids	P-generation
Cross-fertilize	Monohybrid cross	F1-generation
Cross	Dihybrid cross	F2-generation

### Principle of Segregation

- Mendel's hypotheses
- Alleles = alternate forms of genes
- Dominance and recessiveness

Pairs of genes, on homologous chromosomes, separate during gamete formation, and are paired again at fertilization, with one half coming from each parent.

## More Terms

Homozygous

Heterozygous

Phenotype

Genotype

## *Genes and Chromosomes*

Gene: a segment of DNA that codes for a heritable trait

- located at **gene loci** or **loci**
- chromosomes are 1000's of genes

## Principle of Independent Assortment

- Each trait's (gene's) alleles segregate, or split to opposite sides of the cell, independently from one another during gamete formation (shown with a dihybrid cross)
- Useful with a **test cross**: a cross of a unknown genotype with a homozygous recessive to identify the unknown genotype

## Principle of Dominance

- Trait has two different alleles, one is dominant and one is recessive, called complete dominance
- The dominant allele masks the effects of the recessive
- Anytime you have the dominant allele, that phenotype is expressed

*The Relationship of Genotype to Phenotype is Not a Simple One*

**NOW!! EXCEPTIONS TO MENDEL'S PRINCIPLES**

**Incomplete Dominance:**

One allele in a pair is not fully dominant over the other.

*ex. snapdragons*

**Codominance:**

A pair of nonidentical alleles specify two phenotypes, both expressed at the same time in the heterozygote.

*ex. blood type*

**Multiple Alleles:**

A gene may have more than two alleles.

*ex. blood type*

**Pleiotropy:**

A single gene affects many phenotypic characteristics.

*ex. growth hormone*

**Epistasis:**

One set of alleles (a gene) may mask or inhibit the expression of another gene's alleles.

*ex. coat color in Labradors*

**Polygenic Inheritance:**

Continuous variation in a trait, the additive effects of two or more genes on a single phenotypic trait.

*ex. human height*

**Linked Genes:**

Genes located close together on the same chromosome tend to not separate during crossing over and to be inherited together.

## **Problems of Genes**

Pedigree = family history of genetic traits

*Mutation* = a change in the base code of DNA for a gene

Genetic Abnormality vs. Genetic Disorder  
(genetic "disease" not correct)

Autosomal recessive disorders + Autosomal dominant disorders

Carriers = heterozygotes for a recessive disorder

## **Kinds of Mutations**

- 1) Base Substitutions = replacing a base with another
- 2) Base Insertions = adding extra bases to a sequence
- 3) Base Deletions = removing bases from a sequence

## **Chromosomal Aberrations**

- 1) Deletions = loss of whole sections of chromosome
- 2) Duplications = section of chromosome copied
- 3) Inversions = piece of chromosome reversed
- 4) Translocation = part of one chromosome is put into another

## **Changes in Chromosome Number**

Aneuploidy = loss of whole chromosome

Polyploidy = addition of whole chromosome or chromosomes

## Sex Linked Genes

Autosomes = pairs 1-22

Sex Chromosomes = pair 23

## Sex Chromosomes

- Contain genes that determine gender
- Two of them; X and Y
- XX is female, XY is male
- Sex-linked genes are those found on the sex chromosome but are unrelated to sex determination
- Most sex-linked genes are found on X chromosome (80%)
- Passed on maternally

! Most disorders occur in males! Why? There are no such things as male carriers for sex-linked traits.

*ex. colorblindness and hemophilia*

## Some final notes on probability

Mendel's crosses and rules reflect chance, not certainty. Genetic crosses show only the odds of getting a particular genotype at any one time, not what must be.

## Genes, Natural Selection and Adaptation

Some mutations are good. Mutations, genetic recombination and crossing over provide novel new gene combinations.

Genome = the total of all genes in an individual

Gene Pool = the total collection of genes in a population

## The Hardy-Weinberg Equilibrium

Equilibrium law. The frequency of each allele in the gene pool will remain constant. If the frequency of one is known, the other can be determined.

$$P + q = 1$$

$$p^2 + 2pq + q^2 = 1$$

$p$  and  $q$  frequency of alleles for a gene,  $p = A$        $q = a$   
 $p^2$ ,  $2pq$ ,  $q^2$  are frequencies of genotypes in a population

## Molecular Biology of the Gene: Revisited

Genes = short pieces of DNA, are *encoded* in DNA

- ~30,000 in humans
- Located on chromosomes (loci)
- Many on one chromosome, but lots of empty space
- Have a beginning and end

What are exons and intron?

What is Junk DNA? Junk DNA and aging

Many believe that the extra DNA has regulatory functions

### Structure of DNA

Made up of nucleotides

Double stranded

Helix

Sugar-phosphate backbone

Nitrogen bases, face inward

Hydrogen bonds

Ladder-like shape

Complimentary base pairing

- 4 bases: adenine (A), guanine (G), cytosine (C), thymine (T)
- Base sequence determines nature of gene [ATTACGTATCG]
- What is a codon?
- The Genetic Code
- Restriction enzymes and gene swapping