Mendelian Genetics
Gregor Mendel
Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments.

Advantages of pea plants for genetic study:
There are many varieties with distinct heritable features, or characters (such as flower color); character variants (such as purple or white flowers) are called traits.
Mating can be controlled
Each flower has sperm-producing organs (stamens) and egg-producing organ (carpel)
Cross-pollination (fertilization between different plants) involves dusting one plant with pollen from another

Law of segregation
When Mendel crossed contrasting, true-breeding white- and purple-flowered pea plants, all of the F1 hybrids were purple.
When Mendel crossed the F1 hybrids, many of the F2 plants had purple flowers, but some had white.
Mendel discovered a ratio of about three to one, purple to white flowers, in the F2 generation.

Mendel’s model
Mendel developed a hypothesis to explain the 3:1 inheritance pattern he observed in F2 offspring.
Four related concepts make up this model.
These concepts can be related to what we now know about genes and chromosomes.

First
Alternative versions of genes account for variations in inherited characters.
For example, the gene for flower color in pea plants exists in two versions, one for purple flowers and the other for white flowers.
These alternative versions of a gene are now called alleles.
Each gene resides at a specific locus on a specific chromosome.

Second
For each character, an organism inherits two alleles, one from each parent.
Mendel made this deduction without knowing about the role of chromosomes.
The two alleles at a particular locus may be identical, as in the true-breeding plants of Mendel’s P generation.
Alternatively, the two alleles at a locus may differ, as in the F1 hybrids.
Third
If the two alleles at a locus differ, then one (the dominant allele) determines the organism’s appearance, and the other (the recessive allele) has no noticeable effect on appearance. In the flower-color example, the F1 plants had purple flowers because the allele for that trait is dominant.

Fourth
The two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes. Thus, an egg or a sperm gets only one of the two alleles that are present in the organism. This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis.

Useful Terms
Homozygous – An organism with two identical alleles controlling for a particular gene.
Heterozygous – An organism with two different alleles for a particular gene.
Heterozygotes are not true-breeding
Phenotype – Physical appearance of an individual.
Genotype – The genetic make-up of an individual; the genotype is responsible for the phenotype.

Mendel derived the law of segregation by following a single character. The F1 offspring produced in this cross were monohybrids, individuals that are heterozygous for one character. A cross between such heterozygotes is called a monohybrid cross.

Mendel identified his second law of inheritance by following two characters at the same time. Crossing two true-breeding parents differing in two characters produces dihybrids in the F1 generation, heterozygous for both characters. A dihybrid cross, a cross between F1 dihybrids, can determine whether two characters are transmitted to offspring as a package or independently.

Probability governs Mendelian inheritance
Mendel’s laws of segregation and independent assortment reflect the rules of probability. When tossing a coin, the outcome of one toss has no impact on the outcome of the next toss. In the same way, the alleles of one gene segregate into gametes independently of another gene’s alleles.

Multiplication & Addition Rules:
Monohybrid Crosses
The multiplication rule states that the probability that two or more independent events will occur together is the product of their individual probabilities.
Probability in an F1 monohybrid cross can be determined using the multiplication rule.
Segregation in a heterozygous plant is like flipping a coin: Each gamete has a chance of carrying the dominant allele and a chance of carrying the recessive allele.

We can apply the multiplication and addition rules to predict the outcome of crosses involving multiple characters.
A dihybrid or other multicharacter cross is equivalent to two or more independent monohybrid crosses occurring simultaneously.
In calculating the chances for various genotypes, each character is considered separately, and then the individual probabilities are multiplied.

Inheritance of characters by a single gene may deviate from simple Mendelian patterns in the following situations:
When alleles are not completely dominant or recessive
When a gene has more than two alleles
When a gene produces multiple phenotypes

Degrees of Dominance
Complete dominance occurs when phenotypes of the heterozygote and dominant homozygote are identical.
In incomplete dominance, the phenotype of F1 hybrids is somewhere between the phenotypes of the two parental varieties.
In codominance, two dominant alleles affect the phenotype in separate, distinguishable ways.

Relationship between dominance & phenotype
A dominant allele does not subdue a recessive allele; alleles don’t interact that way.
Alleles are simply variations in a gene’s nucleotide sequence.
For any character, dominance/recessiveness relationships of alleles depend on the level at which we examine the phenotype.

Tay-Sachs disease is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain
At the organismal level, the allele is recessive
At the biochemical level, the phenotype (i.e., the enzyme activity level) is incompletely dominant
At the molecular level, the alleles are codominant

Multiple Alleles
Most genes exist in populations in more than two allelic forms.
For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme (I) that attaches A or B carbohydrates to red blood cells: IA, IB, and i. The enzyme encoded by the IA allele adds the A carbohydrate, whereas the enzyme encoded by the IB allele adds the B carbohydrate; the enzyme encoded by the i allele adds neither.

Pleiotropy
Most genes have multiple phenotypic effects, a property called pleiotropy. For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis and sickle-cell disease.

Epistasis
In epistasis, a gene at one locus alters the phenotypic expression of a gene at a second locus. For example, in Labrador retrievers and many other mammals, coat color depends on two genes.

Environmental impact on phenotype
Another departure from Mendelian genetics arises when the phenotype for a character depends on environment as well as genotype. The norm of reaction is the phenotypic range of a genotype influenced by the environment. For example, hydrangea flowers of the same genotype range from blue-violet to pink, depending on soil acidity. Norms of reaction are generally broadest for polygenic characters. Such characters are called multifactorial because genetic and environmental factors collectively influence phenotype.

Many human traits follow Mendelian patterns
Humans are not good subjects for genetic research because:
Generation time is too long
Parents produce relatively few offspring
Breeding experiments are unacceptable
However, basic Mendelian genetics endures as the foundation of human genetics.

Recessively Inherited Disorders
Albinism
Cystic Fibrosis
Sickle-Cell Anemia
Dominantly Inherited Disorders
Achondroplasia
Huntington’s Disease
FATAL!!!! Late Onset
Review