Gregor Mendel and the Birth of Mendelian Genetics

- Mendel grew up on a small farm in what is today the Czech Republic.
- In 1843, Mendel entered an Augustinian monastery.
- Studied at the U. of Vienna (1851-1853)
    - Influenced by a physicist (mathematical applications) and botanist (plant variation)
- Around 1857, Mendel began breeding garden peas to study inheritance.
- Pea plants have several advantages for genetics.
    - Many varieties with distinct heritable features (character) with different variants (traits).
    - Easy to cultivate, short generation time
    - Had strict control over which plants mated with each other – controlled breeding by removing stamens (male sex organs)
    - Studied simple traits that were "either-or"
    - Studied several generations
- In a typical breeding experiment,Mendel would cross-pollinate (**hybridize**) two contrasting, true-breeding pea varieties.
- The true-breeding parents are the ***P generation*** and their hybrid offspring are the ***F1 generation***.
- Mendel would then allow the F1 hybrids to self-pollinate to produce an F2 generation.
- It was mainly Mendel's quantitative analysis of F2 plants that revealed the two fundamental principles of heredity:
    - The Law of Segregation
    - The Law of Independent Assortment

By the law of segregation, the two alleles for a character are packaged into separate gametes

- Parental cross: Purple X White
- F1 generation = all purple flowers
- F1 plants allowed to self-fertilize
- F2 generation included both purple-flowered and white flowered plants
    - The white trait, absent in the F_1, reappeared in the F_2.
- Based on a large sample size, Mendel recorded 705 purple-flowered F_2 plants and 224 white-flowered F_2 plants from the original cross.
- This cross produced a ***3:1 ratio*** (three purple to one white) of traits in the F_2 offspring.
- Mendel reasoned that the heritable factor for white flowers was present in the F_1 plants, but it did not affect flower color.
    - ***Purple flower is a dominant trait and white flower is a recessive trait.***
- Mendel developed a hypothesis to explain these results that consisted of four related ideas.

1. Alternative version of genes (different **alleles**) account for variations in inherited characters.
    - Different alleles vary somewhat in the sequence of nucleotides at the specific locus of a gene.
    - The purple-flower allele and white-flower allele are two DNA variations at the flower-color locus.

2. For each character, an organism inherits two alleles, one from each parent.

    - A diploid organism inherits one set of chromosomes from each parent.
    - Each diploid organism has a pair of homologous chromosomes and therefore two copies of each locus.
    - These homologous loci may be identical, as in the true-breeding plants of the P generation.
    - Alternatively, the two alleles may differ
        - In the flower-color example, the F_1 plants inherited a purple-flower allele from one parent and a white-flower allele from the other.

3. If 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.
        - Mendel's F_1 plants had purple flowers because the purple-flower allele is dominant and the white-flower allele is recessive.

4. The two alleles for each character segregate (separate) during gamete production.

- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes in meiosis.
    - If an organism has identical allele for a particular character, then that allele exists as a single copy in all gametes.
    - If different alleles are present, then 50% of the gametes will receive one allele and 50% will receive the other.
- The separation of alleles into separate gametes is summarized as Mendel's **law of segregation**.
- Mendel's law of segregation accounts for the 3:1 ratio that he observed in the F_2 generation.
- The F_1 hybrids will produce two classes of gametes, half with the purple-flower allele and half with the white-flower allele.
- During self-pollination, the gametes of these two classes unite randomly.
- This can produce four equally likely combinations of sperm and ovum.

- Genetics has some unique, useful vocabulary.
- An organism with two identical alleles for a character is **homozygous** for that character.
- Organisms with two different alleles for a character is **heterozygous** for that character.
- A description of an organism's traits is its **phenotype**.
- A description of its genetic makeup is its **genotype**.
    - Two organisms can have the same phenotype but have different genotypes if one is homozygous dominant and the other is heterozygous.

By the law of independent assortment, each pair of alleles segregates into gametes independently

- Mendel's experiments that followed the inheritance of flower color or other characters focused on only a single character via **monohybrid** crosses (crosses between only 1 pair of contrasting traits).
- He conduced other experiments in which he followed the inheritance of two different characters, a **dihybrid** cross.
- In one dihybrid cross experiment, Mendel studied the inheritance of seed color and seed shape.
    - The allele for yellow seeds (Y) is dominant to the allele for green seeds (y).
    - The allele for round seeds (R) is dominant to the allele for wrinkled seeds (r).
- Mendel crossed true-breeding plants that had yellow, round seeds (YYRR) with true-breeding plants that had green, wrinkled seeds (yyrr).
- One possibility is that the two characters are transmitted from parents to offspring as a package.
    - The Y and R alleles and y and r alleles stay together.
- If this were the case, the F_1 offspring would produce yellow, round seeds.
- The F_2 offspring would produce two phenotypes in a 3:1 ratio, just like a monohybrid cross.
- This was not consistent with Mendel's results.

- An alternative hypothesis is that the two pairs of alleles segregate independently of each other.
    - ***The presence of one specific allele for one trait has no impact on the presence of a specific allele for the second trait.***
- In our example, the F_1 offspring would still produce yellow, round seeds.
- However, when the F_1's produced gametes, genes would be packaged into gametes with ***all possible allelic combinations.***
    - Four classes of gametes (YR, Yr, yR, and yr) would be produced in equal amounts.
- When sperm with four classes of alleles and ova with four classes of alleles combined, there would be 16 equally probable ways in which the alleles can combine in the F_2 generation.
- These combinations produce four distinct phenotypes in a 9:3:3:1 ratio.
- This was consistent with Mendel's results.
- Mendel repeated the dihybrid cross experiment for other pairs of characters and always observed a 9:3:3:1 phenotypic ratio in the F_2 generation.
- Each character appeared to be inherited independently.
- The independent assortment of each pair of alleles during gamete formation is now called Mendel's **law of independent assortment.**