October 10 Lecture notes – Genetics of heredity

Review: Meiosis, diagrammed on blackboard.
Meiosis: The division of a single nucleus (and the cell that contains it) into four daughter nuclei (and cells that contain them). The four daughter cells are haploid (1n) gametes (i.e., egg or sperm).

Heredity: The transmission of genetic features (characters) from one generation to the next.

Character: A heritable feature that varies among individuals (e.g., flower color).

Trait: A specific variant of a character (e.g., red or white flowers).

For thousands of years (at least), humans have noticed that breeding pairs of organisms pass along parental traits to their offspring. We even started taking advantage of this by mating male (♂: symbol for male) and female (♀: symbol for female) individuals with desired traits (or **phenotypes**) – this process is called **selective breeding.**

Selective breeding: Human mediated sexual reproduction of two members of a species, the object of which is to maintain or enhance certain traits (phenotypes)

Species: a group of organisms capable of interbreeding and producing fertile offspring.

Sexual reproduction: Creation of new individuals from the union of gametes produced by existing individuals.

Phenotype – The expressed (i.e., "visible") traits of an organism
-contrast with -
Genotype: The genetic makeup of an organism

Examples given of selective breeding:
Domesticated dogs from gray wolves (Eastern Europe, ~ 15,000+ y.b.p.)
Cattle from wild bull-like ancestor Aurochs (Europe, ~ 8,000 y.b.p.)
Corn from wild ancestor teosinte (Mexico, ~8,700 y.b.p.)

Of course, as humans conducted selective breeding for certain phenotypes, we had no idea of the underlying mechanisms for HOW the traits were transferred from mom and dad to puppies, calves, corn seeds...etc…

That is, until a genius named **Gregor Mendel** came along….

- Gregor Mendel (1822-1884) – his critical work occurred in the 1860's, was a monk and worked in an abbey in what was then Austria
- would become known as the father of genetics.
- Discovered the genetic basis for heredity (see net page)
- was a total and complete badass (!!) and made some incredible insights using just his mind, some simple garden tools, and the scientific method.

By examining the heredity of ***traits*** for certain ***characters*** (flower color, seed shape, etc) in pea plants, here's what Mendel figured out about how traits are passed along from parents to offspring (modern definitions are inset beneath Mendel's conclusions).

1) The "heritable characters" (**genes**) that are passed from parents to progeny exist in two different variations within each parent – these alternative variations are called **alleles**.

> **Gene** (Mendel called them "heritable factors"): A discrete segment of DNA that codes for a particular trait (often a single protein determines an individual character – such flower color). Genes are the basic unit of heredity in organisms.
>
> **Allele**: Different versions (nucleotide sequences) of a gene that code for specific traits (e.g., gene for flower color may have alleles for red or white, or other colors).

2) For each "heritable character" (gene), an individual inherits two alleles – one each from each parent ('mom' and 'dad'). Alleles can be identical or different.

> **Homozygous** – having two identical alleles for a particular trait
> (e.g., flower color: maternal allele = red; paternal allele = white)
>
> **Heterozygous** – having two different alleles for a particular trait
> (e.g., gene for flower color: maternal allele = red; paternal allele = red)

3) If the two inherited alleles are different, then one is *expressed* in (determines) the phenotype and is said to be the **dominant allele**, while the other does not influence the phenotype and is said to be the **recessive allele**

> **Dominant allele**: expressed as phenotype in either homozygous dominant or heterozygous condition
>
> **Recessive allele**: expressed as phenotype ONLY in homozygous recessive condition

4) A sperm or egg (gamete) carry only one allele for each character because allele pairs segregate during formation of gametes (Mendel's "Principle of Segregation").

> **Gamete**: A haploid egg or sperm
>
> **Zygote**: The diploid fertilized egg, which results from the union of a sperm and an egg cell nucleus
>
> **Embryo**: A multicellular diploid eukaryote in early stages of development, from first cell division of the zygote to birth/hatching/germination.

Q: So, how did he Mendel make these discoveries?
A: He drew diagrams of his puzzle (always draw a picture or diagram if you are "stuck" on a problem!!) His diagrams are called **Punnett squares**

Punnett square: a matrix used to determine the genotypic and phenotypic ratios of zygotes (and later, offspring) produced by the union of two known gametes. Note: (simplified Punnett squares appear at the end of these notes)

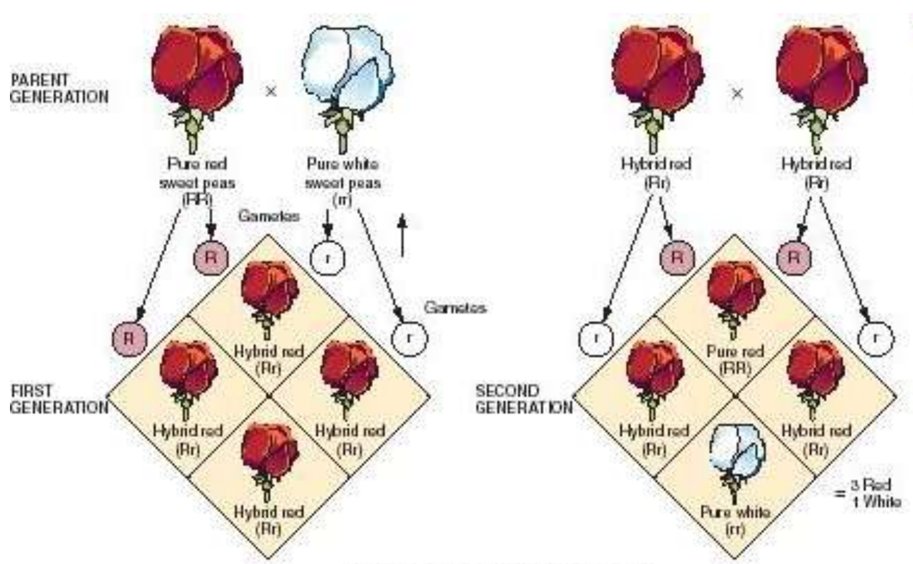
Notation: dominant alleles written as CAPITAL letter, recessive as lowercase letter. Placing a circle around allele symbols is a good habit…I recommend it!!

Mendel always started his experiments with '**true breeding'** individuals. **'True breeding'** individuals would produce offspring identical to the parent when self-pollinated – they are **homozygous.**

Example 1:
In this first example, Mendel starts with a parent generation of pure breeding red and white flowers (*character = flower color; trait = red or white*), and performs a simple cross. The red flower has identical dominant alleles RR coding for red flower color (the red flowered individual is homozygous dominant) and the white flower has recessive alleles rr and therefore will be white (it is homozygous recessive).

When Mendel performs the cross, alleles from an individual parent separate from each other (gamete formation via meiosis!!), then recombine independently with the alleles from the other parent (zygote formation via fertilization!!). The ratio of offspring with particular genotypes and phenotypes of the first generation are self-evident below (they are all heterozygous, and so they all have the phenotype of the dominant allele – red flower).

In the second generation cross, two heterozygous individuals are crossed and by the same mechanisms produce the four new offspring – this time the ratio is 1:2:1 for the genotypes RR : Rr : rr. However, the phenotype ratio is 3:1 red to white!! Simple but elegant – that's how Mendel did it!

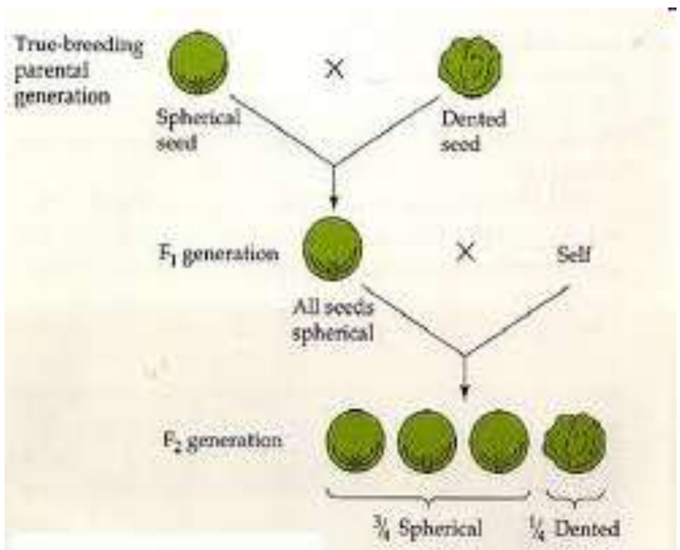


Mendel's First Law: The Law of Segregation

<u>Example 2</u>:

Mendel also examined other characters of pea plants, such as seed shape. The two traits he examined were smooth peas and wrinkled (or 'dented') peas (peas are the seeds of pea plants…but you knew that….I wonder what disperses them?).

See if you provide the correct alleles and genotypes for the peas pictured below. You should be able to produce a Punnett Square for this cross – now and on exams. Puzzle time people! Remember, we will always start with pure-breeding parents (homozygotes) unless otherwise noted…)



Chromosomal basis for heredity:
What Mendel could not "see" or know is where the alleles were physically stored in cells. The answer of course, is on the chromosomes!! Even though he couldn't see the chromosomes, Mendel NAILED the manner in which genes and alleles exist upon homologous chromosomes (two copies of each gene – his observation #1, above) - AND the fact that prior to sexual reproduction the two alleles in an individual parent are separated into egg and sperm, then one allele is combined with a complementary allele from the other parent of the opposite sex. In realizing the alleles are separated he predicted the existence of the process of meiosis! Incredible!

Think about homologous chromosomes and relate Mendel's findings to the meiosis sketch we drew in class.

We'll discuss dihybrid crosses in lab.

Dihybrid cross: A mating of parents that differ in two characters (e.g., seed color and seed shape).

Mendel constructed dihybrid crosses as an experiemtal test of a specific question that he had, specifically: are multiple genes from an individual parent distributed into alleles (and therefore inherited) as a "package" (i.e., the genes always "travel" together as gametes are formed), or are they inherited separately? The result of his dihybrid crosses confirmed his hypothesis that different alleles are inherited seperatley and indeppendetly of each other. From this observation, he posited his principle of independent assortment.

Independent assortment: the principle, originated by Gregor Mendel, stating that when two or more characteristics are inherited, individual hereditary factors assort **independently** during gamete production, giving different traits an equal opportunity of occurring together.

The chromosomal basis for this is random alignment of paternal and maternal chromosomes at Metaphase I and II in meiosis.

You do not need to be able to conduct dihybrid crosses on lecture assessments.

<u>A few final tidbits about human genetics (we'll put these into play during our genetics lab this week). You need to know these terms for lecture assessments.</u>

Homologous chromosomes: Structurally identical chromosomes that code for the same genes – the two different alleles are on homologous chromosomes.

> All of our cells (except for our gametes and red blood cells!) have two homologous chromosomes, one from the egg (the maternal chromosome) and one from the sperm (the paternal chromosome) that originally fused to create us. We have 23 different types of chromosomes, but two homologues of each so that makes 46 total (23 x 2 = 46).

Autosomal traits – alleles are located on non-sex chromosomes

Sex linked traits – alleles are located on sex chromosomes (X and Y)

Females – have two X chromosomes (XX)

Males – have one X and one Y chromosome (XY)

Punnett square excersizes: **BE ABLE TO SOLVE THESE PUZZLES!!**

Example 1: Homozygous dominant parent (AA) x homozygous recessive parent (aa)

Parents: AA x aa

	A	**A**
a	**Aa**	**Aa**
a	**Aa**	**Aa**

Offspring (F_1 generation) **Aa Aa Aa Aa**

Example 2: Homozygous dominant parent (AA) x heterozygous parent (Aa)

Parents: AA x Aa

	A	**A**
A	**AA**	**AA**
a	**Aa**	**Aa**

Offspring (F_1 generation) **AA AA Aa Aa**

Example 3: heterozygous parent (Aa) x heterozygous parent (Aa)

Parents: Aa x Aa

	A	**a**
A	**AA**	**Aa**
a	**Aa**	**aa**

Offspring (F_1 generation) **AA Aa Aa aa**

Understand the GENOTYPES and PHENOTYPES of each of the potential offspring!!