A. Outline:
Transmission Genetics 1
Material from Human Genetics, 7th edition by Ricki Lewis

B. Reading assignment:
   - Lewis Human Genetics 7th edition: Chapter 4 pp 75-85;
   - Cartoon Guide pp 37-55

C. Suggested practice questions:
   - Chapter 4 Review questions 3a, 3c; Applied questions 1-3

Transmission Genetics 1
Genetics is the study of inheritance and variation in organisms.

Why do traits sometimes skip a generation? Occasionally traits that skip a generation are visible in humans, some children have traits present in one of their grandparents, but not in either parent!

Traits that skip a generation can be more obvious in rapidly breeding organisms, including plants. Gregor Mendel studied such traits using pea plants. Mendel studied traits that are governed by a single gene. His findings helped us understand the principles that govern the transmission or inheritance of traits regulated by single genes. His work provided the foundation for the study of genetics.

Reproduction in Plants

Camerarius (1665-1721) - showed that flowers have sex organs and that some plants can self-fertilize, a process called selfing

- Male parts called anthers - contain pollen (sperm)
- Female part called stigma - contains ovary and egg - where pollen attaches and migrates to ovary and egg
Johann Gregor Mendel (1822-1884)
- a monk who lived in Austria in the mid 1800s

- bred *Pisum sativa* - Pea plant - excellent genetic system because it *exists* in stable varieties which *form hybrids*, contain male and female parts (anthers and stigma), and can self-fertilize

- *master of scientific method* - careful observation and record keeping

- studied stable traits each with two easily distinguishable forms
  - tall vs. short
  - smooth vs. wrinkled
  - plump vs. pinched pods
  - yellow vs. green peas
  - white vs. purple flowers
  - unripe pod color
  - seed albumin color
  - axial vs. terminal position of flowers

- he could prevent *selfing* and control parentage by:
  - snipping off anthers
  - dusting stigma with pollen from the desired parent (father)
  - tying a bag over the flowers to keep out other pollen
One of the questions Mendel asked and answered: Why do traits disappear in one generation only to reappear in the next?

Mendel performed pea plant crosses and from the results of his crosses, he described the transmission of traits, now called the Laws of Inheritance.

Mendel hypothesized that plants transmitted distinct units of inheritance - he was correct and we call now call the units of inheritance GENES.

Mendel studies traits that have two distinct forms, i.e. tall and short.

Mendel started his experiments with true-breeding plants. Tall plants that always gave rise to tall progeny and short plants that always gave rise to short progeny.

Parental (P₁) cross: Cross of 2 true-breeding plants, tall and short. We know now that the parents have the genotypes TT (tall) and tt (short). The genetic cross can be written in this manner: TT x tt

The resulting progeny are called the first filial generation or the F₁.

All of the F₁ progeny exhibit one of the traits only. The observed trait is the dominant trait (tall) and the hidden or masked trait is the recessive trait (short). The F₁ progeny have the genotype Tt, receiving the T allele from the tall parent and the t allele from the short parent. They exhibit the tall phenotype.

Mendel found that the cross of TT x tt gives rise to all tall progeny in the F₁ generation, 4 tall:0 short. Tallness masks shortness in the F₁
This representation of the parental gametes and the possible progeny genotypes is called a Punnett square or crossing square - named after Reginald C. Punnett a Brit who, in 1917, introduced this method to predict genotypic and phenotypic ratios of progeny from a genetic cross.

The phenotype of these progeny is tall. However, in the next set of crosses Mendel found that the F\textsubscript{1} progeny, although tall, had a different genotype than the original TT parent.

These hybrid progeny (Tt) hide the expression of one of the traits - this is the basis of dominance. In the next cross the self cross, you will see that these Tt tall plants are not true breeding - they can give rise to tall and short offspring.

**Self cross Figure 4.5**

From a self-cross we expect 75% of the progeny or offspring to express the dominant or Tall phenotype and 25% to exhibit the recessive or short phenotype. The self-cross, a type of monohybrid cross, gives rise to 3 Tall:1 short phenotypic ratio and a 1TT:2Tt:1tt genotypic ratio.
Mendel concluded that:
- Tallness is dominant over shortness
- Tall allele T is dominant
- Short allele t is recessive

TT tall plant - homozygous dominant
Tt tall plant - heterozygous or hybrids
tt short plant - homozygous recessive

In the space below draw the Punnet square depicting a cross between a non-
true breeding tall (Tt) and a short pea plant (tt). What are the possible
parental gametes? Progeny genotypes and phenotypes?

Unlike the progeny of the true-breeding pea plants, both traits show up in
the progeny if one parent is not true breeding. In this case, you would
expect half the progeny to be tall (Tt) and half to be short (tt).

Figure 4.4 Mendel’s first law - gene segregation

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Mendel developed a method to distinguish between tall plants that true breeding and those that are non-true-breeding, called the test cross.

He performed a test cross by crossing an individual of unknown genotype expressing the dominant trait to an individual that is homozygous recessive for the trait being studied (in his case, the short trait).

Examples of test crosses: to determine if a plant exhibiting the dominant trait, tall, is true breeding (TT genotype) or not true breeding (Tt genotype).

Example 1: TT x tt - all progeny are Tt and exhibit the tall trait

Because all the progeny are tall, then the tall parent must be of genotype TT - the tall parent is true breeding or TT. Mendel found that the cross of TT x tt gives rise to all tall progeny in the F₁ generation, 4 tall:0 short.

Example 2: Tt x tt - 2 progeny are Tt and exhibit the tall phenotype and 2 progeny are tt and exhibit the short phenotype. In this case, the tall parent must be Tt able to contribute a T allele to half the progeny and a t allele to the other half of the progeny.

Key concepts: When using true-breeding plants or organisms, the F₁ generation displays the phenotype of one parent (the dominant trait) but because in the F₂ generation the recessive or missing trait reappears, thus the F₁ progeny must inherit information from both parents.

P₁ - TT x tt
F₁ - all progeny are Tt
Mendel allowed the F₁ progeny (heterozygous hybrids, Tt) to self - this cross is a monohybrid cross (a cross following one trait using parents that are hybrids, i.e. Tt).
For the monohybrid cross Tt x Tt, the crossing square looks like this.

\[
\begin{array}{c|c}
T & T \\
\hline
T & TT & Tt \\
\hline
T & Tt & tt \\
\end{array}
\]

F₂ progeny
- genotypic ratio: 1TT:2Tt:1tt
- phenotypic ratio: 3 tall:1 short

One quarter of the offspring are short (homozygous recessive) and 3/4 of the offspring are tall. These results indicated that the F₁ progeny were not true breeding but were heterozygotes. The F₁ progeny had the genotype Tt with one T allele from the TT parent and one t allele from the tt parent.

**Recap:** genetic nomenclature
- \( P_1 \) - parental generation
- \( F_1 \) - first filial generation, \( F_2 \) - second filial generation, etc
- Dominant allele denoted in upper case, T
- Recessive allele is denoted in lower case, t
- Allele - versions of the same gene or DNA sequence
- Genotype - indicates the alleles an organism has
- Homozygous - has the same alleles
- Heterozygous - has two different alleles
- Phenotype - indicates the observed trait
- Genotype and phenotype - distinguish the observed form and the underlying alleles present.
- wild type - the most common version in the general population
- wild type phenotype - most common phenotype
- mutant phenotype - phenotype different from wild type
- wild type allele - most frequent allele associated with the common phenotype
- mutant allele - allele associated with the mutant phenotype