Genetic Terminology

Essential to the mastery of genetics is a thorough knowledge and understanding of the vocabulary of this science. New terms will be introduced and defined routinely in the lectures.

☞ You are required to know all terms defined in lecture.

EXPLORE THE COURSE WEB SITE
http://fire.biol.wwu.edu/trent/trent/index.html
In the 19th century Mendel articulated the fundamental laws of heredity that apply to all eukaryotic organisms.

Many biologists in Mendel’s time were interested in heredity, but Mendel succeeded in elucidating these basic rules of heredity where others failed.

What experimental strategies (still in use today) did Mendel use to discover the basic laws of heredity?
• Inspired choice of an appropriate Model Organism

• Use of modern scientific methodology and careful quantitative analysis of his data: Mendel collected and examined large numbers of progeny from each genetic cross and repeated his experiments to determine if the same results were consistently obtained

• Used a simple symbolism to represent abstract hereditary determinants (genes and alleles)

• Applied the basic rules of probability to explain his data (progeny phenotypes and ratios)
What do we mean by the term model organism?

What does this term imply about the research done on this organism or about the knowledge gained from studying this organism?

(Note this question is not asking about specific features that make good model organisms)
model system:
a label we apply to species that we
http://www.dnalc.org/ddnalc/resources/model_organisms.html
(i) study in detail

(ii) use as a basis for constructing a
general understanding of how biological
processes work
What are the favorite model organisms of geneticists?

1. Start with the classic and still trendy multicellular eukaryote?

2. Classic and still trendy single-celled eukaryote?

Hint:

3. Trendy, classy, and approaching classic (Nobel winning animal)?

4. Not classic yet, but a very trendy weedy plant?

5. Classic prokaryotic model?
Some Favorite Eukaryotic Model Organisms

Drosophila melanogaster

Caenorhabditis elegans: free-living roundworm

Saccharomyces cerevisiae: bakers and brewers yeast

Arabidopsis thaliana: small flowering plant in the mustard family

What question comes to mind when you view these organisms?
This is a seemingly funky collection of organisms….

Why have these particular organisms been picked by geneticists for intensive study?

Speculate on what features would make a good model organism for a geneticist:
Features of good models

• can be raised in the lab easily and inexpensively
• produce large numbers of progeny (why is this important?)
• short generation time
• phenotypic variants are readily available
• some aspect of the habit or life-cycle of the organism lends itself to scientific inquiry
• an element of serendipity has entered into the choice of some models

Model Organisms Guides

http://www.nih.gov/science/models/

http://www.loci.wisc.edu/outreach/text/model.html
model system:
a label we apply to species that we

(i) study in detail

(ii) use as a basis for constructing a
general understanding of how biological
processes work  HUH?

Okay so HOW or WHY can what we
learn about worms or flies or a plant tell
us about how a human functions or what
kinds of genes we have?
We can justify the use of these organism based on practical arguments (limited time, space, money, etc.)

But how do we establish the biological (theoretical) credibility for using studying these organisms?

This argument is based on knowledge of history of life on earth

In other words, Why is a fly or a worm a credible model system for other organisms?

Why do biologists care so much about the genome projects of non-human animals?
Because of our common evolutionary lineage, all organisms share fundamental biological processes that are controlled by genes that are conserved among distantly related organisms.
Using the common pea as a model organism, Mendel elucidated basic principles of inheritance that could be applied to all eukaryotic organisms

One of the reasons that Mendel’s experiments were so successful is that he chose an appropriate organism to work with:

• After investigating and discarding other candidates, Mendel chose the pea plant *Pisum sativum* for practical reasons.

• Peas are cheap and easy to propagate

• They have a relatively short reproductive cycle. This facilitated the collection of data from a large number of plants

• Reproduction of the pea plant can be artificially manipulated

• A large number of different varieties of this species were available from the local “seedsman”.

So how did Mendel actually approach his study of hereditary determinants?

If you are a molecular geneticist, you know you are looking at a gene when you examine a stretch of DNA and see that it codes for protein and has a promoter in front of it
>gi|455025|gb|U01317.1|HUMHBB Human beta globin region on chromosome 11
TCTTATTTGTGTAATAAGAAGTTTGAGCAAGTTGATCTCTCAATATTGCTTACCAAGTCTG
TGATCCAAATATTATTCTAATACCTTCGGCAAAAGGAGGTGTTTTGATTAGCAATT
GTACGTGTTAGTGGGCGCAAGAGGATATATATCTTAGAGGGGCTGAGGGTTTTGA
TGCCCAACTCTCAAAGCCAGTGCACAGCAAGCCAGGACCAGGTACGCTGCTCTATTAG
TAGCAGCTGACACTCAGCTCAGGAGAAGTCTGAGTGCCTTTGAGGCAAGGTGAAC
GTATGAGTTGGGCTGCCGCTGCCCTGAGGAGACAGAACGACTTGGTTGAGAAG
TTGGTGGTGAGGCCCTGGGCTGGTTGAAAGGTTACAAGGTTTAAGGAGACGAG
AAGGACACATAGAAACTGGGCAATGATCTGAGACAGAGAACGACTCTTGGGTTTAG
GTAGTGGCAACACCAGTCTACTGGGCAAGAGATATCTTAGAGGGGCTGAGGGTTTG
GCTACACCTTGGAGCAACAGCAGGTTCTTGAGTCCTCGACTCAGTCTGACTCT
GTATGGGCAACCCCAAGCAGGTTCTTGAGTCTGCTCGACTCAGTCTGACTCT
GGAGAAGGTGTTTGAGGCAAGGTTTACATGATCTGAGACAGAGAACGACTTTGGG
TAGTTGCAATTGCTAATTTTTCTAATTATTTATATAGGGAATGTGGGAGGTCAG
TGCATTTAAAACATAAAGAAATGAAGAGCTAGTTCAAACCTTGGGAAAATACACTA
TA

AAACATTTATTTTCATTGCA

ATGATGTATTTAAATTATTTCTGAATATTTTACTAAAAAGGGAATGTGGGAGGTCAG
TGCATTTAAAACATAAAGAAATGAAGAGCTAGTTCAAACCTTGGGAAAATACACTA
TA

AAACATTTATTTTCATTGCA

ATGATGTATTTAAATTATTTCTGAATATTTTACTAAAAAGGGAATGTGGGAGGTCAG
TGCATTTAAAACATAAAGAAATGAAGAGCTAGTTCAAACCTTGGGAAAATACACTA
TA
But, how do you know that you are “looking” at a gene if you are a classical geneticist and don’t know the molecular language of DNA?

How did Mendel identify the existence of a gene?
In classical genetic methodology, the existence of a gene controlling a trait is inferred from phenotypic variation between individual organisms or groups of organisms.

In other words, using traditional genetic methodology, a gene is defined by specific phenotypic differences.
A large number of different lines or varieties of peas were available to Mendel from his local “seedsmen.”

- For each trait, his lines exhibited one of two alternative variations or forms. For example, for the trait of flower color, each of Mendel’s lines were **true-breeding** for either purple flowers or white flowers.

- These lines then differed with respect to the **phenotype** of flower color.
- Note that some of the phenotypes that Mendel examined are properties of the peas (seeds) and others are properties of the plant.

This variation in phenotype provided Mendel with the raw material for his classical genetic studies.
Before starting his experimental analysis, Mendel subjected his different varieties of pea plants to a two year trial to confirm that he was working with true-breeding or pure lines.

What is a true-breeding line and why is it important?
Reproduction of the pea plant can be artificially manipulated

- In the pea plant the male and female reproductive parts are present in the same flower and are enclosed in a compartment which prevents pollination from outside sources.
- So accidental fertilization by foreign pollen can not easily occur in this organism.
- Pea plants will self-pollinate unless the anthers of the flower are removed by the experimenter before they release pollen.
- **self-pollinate = self = self-fertilize:** pollen from a given flower fertilizes the ovules from the same flower or from a different flower on the same plant

Mendel was able to artificially cross-pollinate (= cross) two different pea plants by removing the immature anthers from the flowers of one plant and then
• Truebreeding line: a pure line, when selfed or cross-pollinated within the same breeding line, will only give rise to progeny identical to the parents.

• This important step demonstrated that the outcome of selfing (or crossing within) the various strains was predictable and consistent.

• Therefore, deviations following cross-pollination between different strains would be scientifically significant.
What were the questions about heredity that Mendel’s research addressed?

What are the basic mechanisms of heredity? The question is so broadly phrased that it must be broken down into a series of more specific questions in order to be approached in a logical, systematic way:

• Do males and females contribute equally to the appearance of their offspring?
• Are parental traits irreversibly blended in the offspring?
• Are there basic rules, which can be described mathematically, for the transmission of hereditary elements from one generation to another?

The first two questions reflected the confusing thinking about heredity that existed in the mid-19th century
The third question reflects Mendel’s training in physics and mathematics:

• 19th Century physics attributed everything in nature to the action of a small number of laws, the starting point of which was the existence of certain indivisible patterns of matters

• The laws of nature were written in the language of mathematics and the task of the researcher was to discover the existence and activity of these indivisible particles of matter

Crosses and data on BOARD
Inheritance of a single trait: round or wrinkled seeds

♦ Mendel crossed a true-breeding line that had round seeds with a true-breeding line with wrinkled seeds

**PARENTAL**

♦ All of the progeny seeds resulting from this cross were round. These progeny are called the *F1* (for first filial) generation. [Subsequent generations are indicated as *F2*, *F3*, and so on.]

♦ Mendel then did a reciprocal cross and obtained the same result

♦ *What was the significance of the results of the reciprocal crosses?*
Mendel planted the F1 seeds and allowed the F1 plants to self-pollinate

In this **F2 generation**, he observed 5474 round and 1,850 wrinkled seeds, which represents a ratio of round: wrinkled equal to 2.96 to 1.

This is essentially a 3 to 1 ratio.

In other words, about 3/4 of the F2 seeds were round and 1/4 were yellow. No seeds of intermediate phenotype were observed.
Mendel had made the *striking observation* that even though the wrinkled form had disappeared in the F1 generation, it reappeared in the F2 generation.

**THERE WAS NO IRREVERSIBLE BLENDING OF THE TRAITS**

To describe this result, he designated the round phenotype as the **dominant** phenotype since it appeared in the F1 and the wrinkled phenotype as **recessive**.
Mendel repeated these experiments with the six other traits and obtained similar results:

1. The results of reciprocal crosses were the same

2. One form of the trait was always dominant. The F1 progeny only showed the dominant trait and did not exhibit any intermediate phenotype.

3. In the F2 generation, the ratio of plants with the dominant and recessive phenotypes was 3 to 1.
FROM THESE AND OTHER DATA

♦ Mendel proposed that the hereditary determinants for each trait are discrete and do not become blended together in the F1, *but maintain their integrity from generation to generation.*

♦ Mendel also proposed that, for each trait examined, the pea plant contains two copies of the hereditary determinant (gene) controlling the trait, one copy coming from each parent.
Write up genotypes of crosses

**MENDEL USED A SIMPLE SYMBOLISM TO CONNECT THEORY AND OBSERVATIONS.**

- Mendel developed a simple symbolism to show how his *principle of segregation* could be used to explain his observations.
- He designated the **genotype** or genetic composition of a given pea plant in the following manner.
- Each pair of genes, was symbolized by a different letter.
- The dominant form of the gene was indicated by an upper-case letter and the recessive form by a lower-case letter.
- These alternative forms of a gene are called **alleles**.
- Returning to the experiments on seed form, the dominant round allele is designated by **R** and the recessive wrinkled allele by **r** (Figure 4).
- The genotype of the true-breeding parental round line is **RR** indicating that it carries two copies of the round allele in each cell.
- Such pure lines are said to be **homozygous** (*homo* -meaning “identical”) for the round allele.
- The genotype of the parental wrinkled line is **rr** indicating it is homozygous for the recessive wrinkled allele.
- The F1 round progeny contain a dominant allele for round contributed by the round parent and a recessive allele for wrinkled contributed by the wrinkled parent. T
- The F1 plants are **Rr** or **heterozygous** (*hetero* -meaning “different”) for the two different alleles.