Mendelian Genetics

Ch. 2

The historical puzzle of inheritance

- Artificial selection has been an important practice since before recorded history
 - Selection of animals for domestication
 - Selective breeding of plants
- 19th century Breeders desire precise techniques for controlled matings in plants and animals in order to produce desired traits in many of offspring
- Breeders could not explain why traits would sometimes disappear and then reappear in subsequent generations.

Animal Mating Mysteries





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Important genetics questions in the early 1800's

What traits are inherited?

How are traits inherited?

What is the role of chance in heredity?

Gregor Mendel



Important Facts

Monk Founder of Genetics "Father of Genetics"



Mendelian Genetics

But first, let's introduce a few terms

- Mendelian factors are now called genes
- <u>Alleles</u> are different versions of the same gene
- An individual with two identical alleles is termed <u>homozygous</u>
- An individual with two different alleles, is termed <u>heterozygous</u>
- <u>Genotype</u> refers to the specific allelic composition of an individual
- <u>Phenotype</u> refers to the outward appearance of an individual

Genotype and Phenotype

- Hereditary traits are under the control of genes (Mendel called them factors).
- <u>Genotype</u> is the genetic makeup of an organism, a description of the genes it contains.
- Phenotype is the characteristics that can be observed in an organism.
- Phenotype is determined by interaction of genes and environment.
 - Genes provide potential, but environment determines whether that potential is realized (Figure 2.1).

Fig. 11.1 Influences on the physical manifestation (phenotype) of the genetic blueprint (genotype)

Genotype (genetic constitution)

Actions of other genes and their products Environmental influences and random developmental events

Phenotype (expression of physical trait)

Alternative forms of traits are alleles

- Each trait carries two copies of a unit of inheritance, one inherited from the mother and the other from the father
- Alternative forms of traits are called alleles



2.1 MENDEL'S LAWS OF INHERITANCE

- His work, entitled "Experiments on Plant Hybrids" was published in 1866
- Like many great scientific discoveries, it was ignored for 34 years
- In 1900, Mendel's work was rediscovered by three botanists working independently
 - Hugo de Vries of Holland
 - Carl Correns of Germany
 - Erich von Tschermak of Austria

Keys to Mendel's Experiments

- The garden pea was an ideal organism for study because:
 - Vigorous growth
 - Self fertilization
 - Easy to cross fertilize
 - Produces large number of offspring each generation
- Mendel analyzed traits with discrete alternative forms (one of two options)
 - purple vs. white flowers
 - yellow vs. green peas
 - round vs. wrinkled seeds
 - long vs. short stem length
- Mendel established pure/true breeding lines to conduct his experiments.
 - (traits remain constant from generation to generation)

Monohybrid Crosses and Mendel's Principle of Segregation

Terminology used in breeding experiments:

- Parental generation is the P generation.
 - True breeding unless stated otherwise
- Progeny of P generation is the first filial generation,
 - designated F1.
- When F1 interbreed or are "selfed",
 - the second filial generation, F2, is produced.
 - F2 is always F1 x F1
- Subsequent interbreeding produces F3, F4, and F5 generations.

Monohybrid Crosses and Mendel's Principle of Segregation

- A monohybrid cross involves true-breeding strains that differ in a single trait.
- To determine whether both parents contribute equally to the phenotype of a particular trait in offspring
 - a set of reciprocal crosses is performed.
- In Mendelian genetics, offspring of a monohybrid cross will exactly resemble only one of the parents.
 - This is the principle of uniformity in F1 (Figure 2.5).
 - Complete dominance



Monohybrid Crosses and Mendel's Principle of Segregation

- Mendel noted that traits that disappear in the F1 reappear in the F2.
- The F2 has a ratio of about three individuals with the phenotype of the F1 to one individual with the "reappearing" phenotype
 - 3:1 ratio
 - 3/4 to 1/4
- Mendel reasoned that information to create the trait was present in the F1 in the form of "factors"
 - now called genes (Figure 2.6).

Monohybrid Crosses and Mendel's Principle of Segregation

By convention, letters may be used to designate alleles,

- with the dominant a capital letter (*S*)
- and the recessive in lowercase (*s*).
- Individuals with identical alleles are said to be homozygous for that gene
 - (e.g., genotypes *SS* and *ss*)
 - because all their gametes will have the same allele for this trait.
- Individuals with different alleles are heterozygous
 - (e.g., *Ss*)
 - because 1/2 of their gametes will contain one allele, and 1/2 the other (Figure 2.7).





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Monohybrid Crosses and Mendel's Principle of Segregation

- When Mendel had conducted experiments for the seven different traits in garden peas (Table 2.1), he made these conclusions:
 - 1. Results of reciprocal crosses are always the same.
 - 2. The F1 resembled only one of the parents.
 - 3. The trait missing in the F1 reappeared in about 1/4 of the F2 individuals.

Punnett Squares

- A <u>Punnett square</u> is a grid that enables one to predict the outcome of simple genetic crosses
 - proposed by the English geneticist, Reginald Punnett
- I. Write down the genotypes of both parents
 - Male parent = Tt
 - Female parent = *Tt*
- 2. Write down the possible gametes each parent can make.
 - Male gametes: *T* or *t*
 - Female gametes: *T* or *t*

3. Create an empty Punnett square



4. Fill in the Punnett square with the possible genotypes of the offspring

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- 5. Determine the relative proportions of genotypes and phenotypes of the offspring
 - Genotypic ratio
 - TT : Tt : tt
 - **1** : 2 : 1
 - Phenotypic ratio
 - Tall : dwarf
 - 3 : 1

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The Principle of Segregation

- The first Mendelian law, the principle of segregation, states:
 - "Recessive characters, which are masked in the F1 from a cross between two true-breeding strains, reappear in a specific proportion in the F2."
- This is because alleles segregate during anaphase I of meiosis
 - and progeny are then produced by random combination of the gametes.

Confirming the Principle of Segregation:

Mendel observed that plants with the recessive phenotype are all true breeding.

Not true for the dominant phenotype

- When plants with the dominant phenotype are self crossed
 - 1/3 are true-breeding,
 - and 2/3 produce progeny with both phenotypes
 - (Figure 2.10).

Q ð F₁ generation F₁ phenotypes Smooth Smooth seeds seeds Diploid F₁ genotypes Ss Ss Haploid × F₁ gametes S $F_1 \circ d$ gametes 1/2 1/2 F₂ generation SS **F**₁♀ gametes 1/2

F2 genotypes: 1/4 SS, 1/2 Ss, 1/4 ss

F₂ phenotypes: ³/₄ smooth seeds, ¹/₄ wrinkled seeds

Fig. 2.10b Mendel's first law, principle of segregation of Mendelian factors: Production of the F₂ generation

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Confirming the Principle of Segregation:

You can determine whether an individual with the dominant phenotype is homozygous or heterozygous by self crossing the individuals (Figure 2.11).

Fig. 2.11 Determining the genotypes of the F_2 smooth progeny of Figure 2.10 by selfing the plants grown from the smooth seeds



Confirming the Principle of Segregation: Testcrosses

- A more common way to determine whether an individual with the dominant phenotype is homozygous or heterozygous is to perform a testcross
 - By definition you must cross the individual with one that is homozygous recessive (Figure 2.11).

Testcrosses

- A test cross allows one to determine the alleles carried by the F1 parent:
- because the test cross parent can only contribute the recessive allele is ALWAYS homozygous recessive!
- The phenotypes of the resulting progeny allow you to determine the genotype of the F1 parent.



Testcrosses

- Used to determine the genotype of a phenotypically dominant individual
 - is a round pea plant heterozygous (Rr)
 - or homozygous (RR)?
- Cross with individual with known genotype... a phenotypically recessive individual
 - in this case, wrinkled pea plant (rr)
- Test for hidden recessive genes with <u>homozygous</u> <u>recessive</u> tester strain *phenotype* of progeny from test cross indicates the *genotype* of tested parent

> DEFINITION:

- > A **test cross** is a cross of the F_1 plant to a <u>homozygous recessive</u> plant
- Test crosses help establish gentoype

Expected Results of a Testcross

- > Yellow pea is dominant to green pea
- Cross A = If homozygous
 - YY X yy (tester)
 - expect 100% of progeny are dominant phenotype
 - progeny will all be Yy
- > Cross BN = If heterozygous
 - Yy X yy (tester)
 - expect 50% of progeny are recessive phenotype (yy)
 - 50% are **Yy**
- This works for all 7 of Mendel's chosen traits
- (and many others)



- If Mendel had a homozygous tall plant what would the genotype look like from this individual?
- 1) tall
- 2) short
- 3) TT
- 4) Tt

- If Mendel had a homozygote recessive plant for height what would the phenotype look like from this individual?
- 1) tall
- 2) short
- **3)** TT
- **4)** Tt

- If homozygous recessive for plant height what would gametes look like?
- 1) all TT
- 2) all tt
- 3) half T and half t
- 4) all t
 - 5) all T

- Assume that in guinea pigs, dark brown fur (B) is dominant to black fur (b). If you mate a black guinea pig with a homozygous brown guinea pig, what proportion of the progeny will be homozygous?
 - -1.100%
 - -2.50%
 - -3.35%
 - 4. none

Homework Problems

Chapter 2

1, 4, 5, 6, 7, 10, 11