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- Genetics - Part 1 - Genes

<u>Mendel</u>

Mendel was an Austrian monk who taught natural science and worked on plant breeding experiments.

He developed a basic understanding of genetics and inheritance.

Mendel's Work

It took him 2 years to select the pea plant as his subject.

He collected data for 10 years.

His sample sizes were large; he tabulated results from 28,000 pea plants.

He replicated his experiments.

He analyzed his data with statistics (probability theory).

Characteristics of Garden Peas:

Peas are easy to grow, and take little space.

They are inexpensive.

They have a short generation time compared to large animals so that a large number of offspring can be obtained in a short amount of time.

They have some distinct characteristics that are easy to recognize. These characteristics can be used when trying to determine patterns of inheritance.

They are easily self-fertilized or cross fertilized.

Traits Studied by Mendel

smooth or wrinkled seeds yellow or green seeds red or white flowers inflated or constricted pods green or yellow pods axial or terminal flowers tall or dwarf plants

Mendels Crosses

Mendel used pure-breeding individuals in the first (P_1) generation.

- $\begin{array}{cc} P_1 & \text{yellow X green} \\ \downarrow \end{array}$
- F_1 all yellow
- \mathbf{F}_2 3/4 yellow, 1/4 green

Mendel's Results for 7 different crosses

P ₁	F ₁	F ₂	F ₂ ratio
smooth X wrinkled seeds	all smooth	5474 smooth 1850 wrinkled	2.96:1
yellow X green seeds	all yellow	6022yellow2001green	3.01:1
axial X terminal flowers	all axial	651 axial 207 terminal	3.14:1
red X white flowers	all red	705red224 white	3.15:1
inflated X constricted pods	all inflated	882 inflated299 constricted	2.95:1
green X yellow pods	all green	428 green 152 yellow	2.82:1
tall X dwarf plants	all tall	787 tall	2.84:1

	277 dwarf
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Conclusions from Mendel's Crosses

The F_1 generation showed only one character that was present in the P_1 . The other character reappeared in the F_2 (25%).

The sex of the parent did not matter.

The traits did not blend.

Mendel concluded that the F_1 plants must contain 2 discrete factors, one for each character. The character that was seen in the F_1 is called *dominant*. The character not seen in the F_1 is called *recessive*.

Letters Can Represent Genes

The characteristics studied by Mendel were due to single genes. On the pair of <u>chromosomes</u> diagrammed below, the letter "A" represents a gene for yellow seeds. The letter "a" on the <u>homologous chromosome</u> represents a gene for green seeds. By convention, upper case letters are used to represent dominant genes and lower case letters are used for recessive genes.

Because individuals are <u>diploid</u>, two letters can be used to represent the genetic makeup of an individual. In the case of seed color, the following three gene combinations are possible: AA, Aa, and aa.



Heterozygote (also called hybrid) refers to an individual that has two different forms of the gene. Example: Aa

Homozygote refers to an individual that has two identical genes. Example: AA or aa

A **hybrid** is a heterozygote. Example: Aa

Meiosis, Gamete Formation

The three diagrams below show <u>metaphase I</u>, <u>anaphase I</u> and <u>telophase I</u> in an "Aa" individual.

As can be seen in the diagrams, an "Aa" individual can produce <u>gametes</u> that have "A" and gametes that have "a".



Principle of Segregation

Mendel's principle of segregation states that paired factors (genes) separate during gamete formation (meiosis). Because the pair of genes (Aa, AA, or aa) separate, one daughter cell will contain one gene and the other will contain the other gene. (See diagram above.)

Gametes

Because pairs of chromosomes separate during meiosis I, gametes are <u>haploid</u>, that is, they carry only one copy of each chromosome. An Aa individual therefore produces two kinds of <u>gametes</u>: A and a.



Below: An "AA" individual produces all "A" gametes. Similarly, an "aa" individual produces all "a" gametes.



Individual (genotype)	Type of gametes produced	
АА	all gametes will contain an "A"	
Aa	1/2 will contain "A" and 1/2 will contain "a"	

all	"a"	gametes
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Punnett Squares

aa

Suppose that an "Aa" individual is crossed with another "Aa" individual. One will produce "A" eggs and "a" eggs. The other will produce "A" sperm and "a" sperm. What are all of the possible combinations of eggs and sperm? A Punnett square can be used to show all of these combinations.

The Punnett square in the diagram below is used to show between two Aa individuals.



Gametes produced by the first parent

The square below is used for this cross: AA X Aa.

	A	а
A	AA	Aa
A	AA	Aa

One half of the offspring produced by this cross will be AA, the other half will be Aa.

The cross can also be written as shown below because the AA parent can produce only one kind of gamete (all A).

A Closer look at Mendel's Crosses (One Gene Locus)

Y = yellow **y** = green

 $\begin{array}{ccc} P_1 & \mathrm{YY} \ \mathrm{X} \ \mathrm{yy} \\ & \downarrow \\ F_1 & \mathrm{Yy} \end{array}$

Yy X Yy \leftarrow A cross between two individuals that are heterozygous for a trait is called a <u>monohybrid cross</u>.

 \mathbf{F}_2 The above cross is illustrated below.

	Y	У
Y	YY	Yy
У	уY	уу

Genotype and Phenotype

The genetic makeup of P_1 plants was different from that of F_1 because the P_1 plants were true breeding and the F_1 plants were not. The genetic makeup of an individual is referred to as its *genotype*. Because the plants are diploid, two letters can be used to write the genotype. In this case, the genotype of the P_1 plants was YY; the genotype of the F_1 plants was Yy.

The characteristics of an individual are its *phenotpye*. This word refers to what the individual looks like so ddjectives are used to write the phenotype. For example, "yellow" or "tall" are phenotypes. The yellow P_1 plants looked like the F_1 ; they had the same phenotype but different genotypes.

An individual with a recessive phenotype has two recessive genes. A dominant phenotype results from either one or two dominant genes. In the cross above, YY or Yy are yellow; yy is green. The phenotype ratio in the F2 is 3 yellow:1 green. The genotype ratio is 1YY:2Yy:1yy.

Genotype	Phenotype
AA or Aa	Yellow
aa	Green

Other Crosses

S = smooth s = wrinkled

$$\begin{array}{ccc} P_1 & SS \ X \ ss \\ & \downarrow \\ F_1 & Ss \end{array}$$

Ss X Ss

	S	S
s	SS	Ss
s	sS	SS

 \mathbf{F}_2 genotype ratio = 1:2:1 (1SS : 2Ss : 1ss)

phenotype ratio = 3:1 (3Smooth : 1 wrinkled)

F = full **f** = constricted

 $\begin{array}{ccc} \mathbf{P_1} & \mathrm{FF} \ \mathrm{X} \ \mathrm{ff} \\ & \downarrow \\ \mathbf{F_1} & \mathrm{Ff} \\ & \mathrm{Ff} \ \mathrm{X} \ \mathrm{Ff} \\ & & \\ &$

 \mathbf{F}_2 genotype ratio = 1:2:1 (1FF : 2Ff : 1ff)

phenotype ratio = 3:1 (3full: 1 constricted)

Alleles and Loci

An **allele** is a gene that has more than one form. Each of the forms is referred to as an allele. For example, the gene for red flowers and the gene for white flowers are two different alleles.

A **locus** (plural: **loci**) is the location of a gene on a chromosome. The gene for red flowers and the gene for white flowers are two different alleles at the same locus. A single chromosome can have a gene for white flowers or a gene for red flowers but not both.

There are two **loci** illustrated below, one is for flower color and the other is for stem length. Flower color has five **alleles** and stem length has two.



Testcross - One Locus

let A = red

a = white

Is a red flower AA or Aa?

Solution: cross it with aa

 P_1 A? X aa

The A? individual can produce these kinds of gametes: "A" and "?"

gametes: A, ? and a

 F_1 Aa and ?a

If the ?a individual is red, then ? = A. If it is white, then ? = a.

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