CHAPTER 2 - MENDELIAN ANALYSIS

I. MENDEL'S LIFE
A. Born 1822 – Heinzendorf, Moravia (then part of Austria)
B. Novice at 21, priest at 25 in Brunn Augustinian monastery
C. Attended U. of Vienna where he studied chemistry, zoology, botany, entomology, paleontology, and physics
D. Kept bees, mice, amateur meteorologist, kept records all his life (records of sunspots, groundwater level via monastery well, detailed report of tornado), director of institute for deaf mutes, chairman of a mortgage bank, became abbot of monastery
E. Began experiments with garden pea, Pisum sativum in 1854-1856
F. "Experiments in Plant Hybridization" first published in Brunn Society for Natural History in 1866.

II. MENDEL'S EXPERIMENTS
A. Mendel started his experiments during a period in history when the prevailing idea was that the characteristics of an organism were due to the blending of the traits from each parent (blending inheritance). His work suggested otherwise though. Mendel proposed the idea that an element determined a particular characteristic of an organism. This theory that an element/ factor/ gene controlled heredity was called particulate inheritance. Now it is known that that the gene is the fundamental unit of heredity.

B. Mendel's work was not appreciated at the time, and it was rediscovered by Hugo deVries, Carl Correns, and Erik von Tschermak in the early 1900's.

C. General considerations why Mendel's expts. with garden peas were successful
    1. Garden pea which was easily manipulated so that controlled matings could be done. He could easily control pollination.
    2. Pea plants had a short generation time.
    3. Mendel chose easily identifiable characteristics to examine in the pea plants. Later on we will also see that he was lucky in his choices in that they are simple cases of inheritance, uncomplicated by other factors.
    4. Mendel carefully quantitated his results.
    5. Pea plants generated a large number of progeny, which was good for statistical analysis.
    6. Pure breeding lines had been established. Pure lines meant that the trait of interest remains unchanged generation after generation in self fertilized plants

III. CHARACTERS OR TRAITS THAT MENDEL EXAMINED.
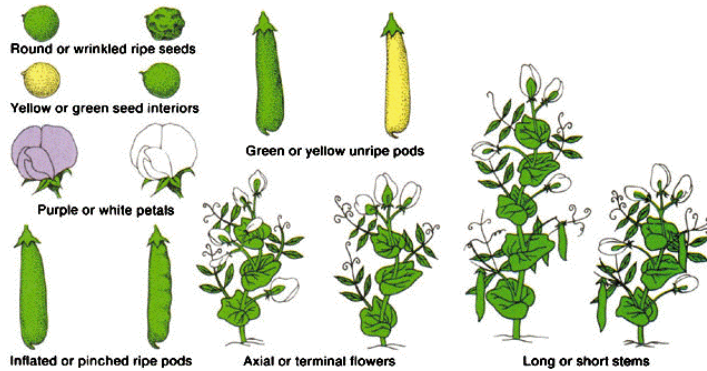
(from An Introduction to Genetic Analysis, 6th ed. By Griffiths et al. W. H. Freeman and Company)

Different forms of each character are called <u>phenotypes</u>.

IV. EXP. SET #1 (THE <u>MONOHYBRID</u> CROSS)
   A. Trait was seed color

   Y=yellow and G=green

   <u>parental generation</u> Y x G   ------------->

   <u>F$_1$ (first filial) generation</u> were all Y ----<u>selfed</u>-----> F$_2$ were 3Y and 1G

   Among the F$_2$ progeny that were selfed
   1/4 Y --------->all Y
   2/4 Y --------->3 Y and 1 G
   1/4 W --------->all G

   Therefore the Y plants in the F$_2$ were really all not the same because they gave different type progenies and the underlying ratio if we consider what type progeny they give was 1 true breeding Y: 2 mixed breeding Y: 1 true breeding G

   B. Mendel found this same pattern for all the traits he examined

C. What did all of Mendel's monohybrid crosses lead him to propose?
   1. <u>The particulate theory of inheritance</u>: Characters are determined by discrete units (genes) that are transmitted from parent to progeny in gametes. Each pea plant has two factors which control each trait.
      a) No blending because F_1 progeny did not have intermediate color
      b) Reappearance of G in F_2 which suggested that even though the parents were Y, they still had the something that gave them the directions to produce G.
   2. One factor of the pair is <u>dominant</u> over the other.
   3. <u>Mendel's First Law or the Principle of Equal Segregation</u>: Each factor independently segregates into the egg/sperm. This is: 2 members of a gene pair segregate (separate) from each other into gametes. 1/2 the gametes carry one member of the pair, the other 1/2 the gametes carry the other.
   4. The union of the gametes is random.

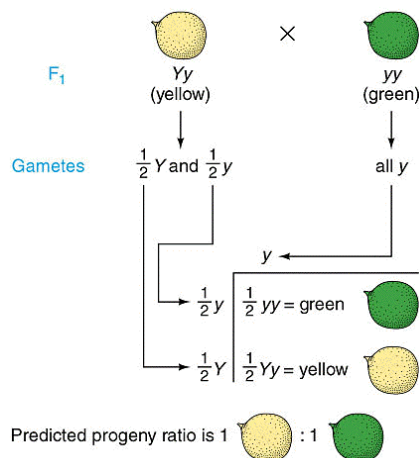D. Testing the Principle of Equal Segregation
   Mendel set up additional crosses to test his model. He made predictions based on the Principle of Equal Segregation and then examined the progeny of the crosses to see if the predictions held up. The most important cross is the <u>test cross</u> (cross to a pure breeding recessive)

   Trait was seed color again (Y=yellow and G=green)

   parental generation Y x G ------------> F_1 generation were all Y
   crossed F_1 Y with G

   Based on Mendel's laws
   1. Trait for seed color was controlled by a pair of factors (genes) were Y = yellow and y = green
   2. Yellow is dominant to green
   3. Equal segregation of factors

F_1 Yy (yellow) × yy (green)

Gametes $\frac{1}{2}Y$ and $\frac{1}{2}y$ all y

y
$\frac{1}{2}y$ | $\frac{1}{2}yy$ = green
$\frac{1}{2}Y$ | $\frac{1}{2}Yy$ = yellow

Predicted progeny ratio is 1 : 1

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***Phenotypic ratio of test cross is 1:1 for Mendelian inheritance

V. RELATING MENDEL'S EXP. TO TODAYS GENETIC TERMS

- <u>gene</u> = the physical unit of heredity allowing information transfer from one generation to the next (i.e. the seed color gene)
- <u>allele</u> = one of two or more alternate forms of a gene (i.e. Y and y are alleles of the gene coding for the seed colors yellow and green, respectively)
- <u>genotype</u> = the specific allele composition of a cell or organism; many times just referring to one gene (yy for plants that produce green seeds)
- <u>phenotype</u> = the appearance of a cell or organism; more specifically, the outward manifestation of the genotype (green or yellow for seed color)
- <u>heterozygous gene pair</u> = a gene pair having two different alleles (i.e. Yy)
- <u>heterozygote</u> = organism that has a heterozygous gene pair
- <u>homozygous gene pair</u> = a gene pair having two of the same alleles (i.e. YY)
- <u>homozygote</u> = organism that has a homozygous gene pair
- <u>homozygous dominant</u> = indicates that gene pair is homozygous for a dominant allele (i.e. YY)
- <u>homozygous recessive</u> = indicates that gene pair is homozygous for a recessive allele (i.e. yy)

VI. EXP. SET #2 (THE DIHYBRID CROSS)
   A. Trait 1 was seed color: (either yellow or green)
      Trait 2 was seed shape: (either round or wrinkled)
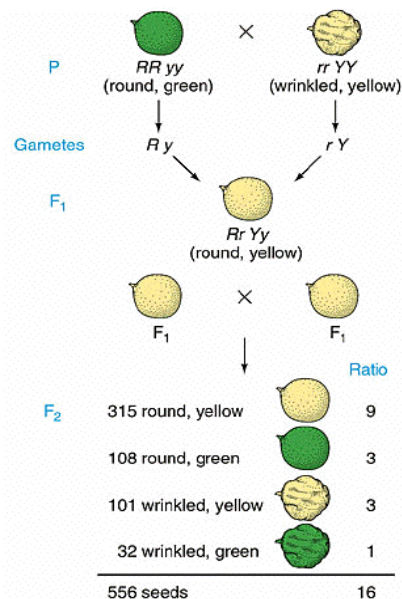
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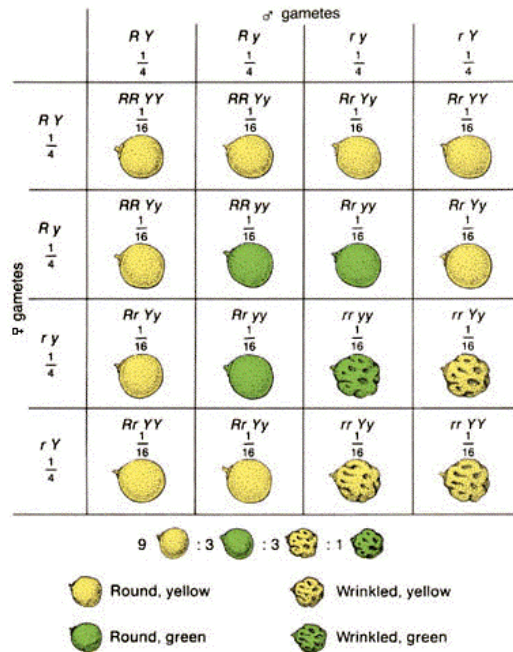
F_1 ratios told Mendel that the dominance of one trait is unaffected by another trait.

At first, the F_2 ratios seemed really confusing; however, Mendel carefully analyzed each trait separately and found that color was 3:1 and shape was 3:1. This is the same phenotypic ratio Mendel saw for the monohybrid crosses! So Mendel proposed the his 2<u>nd</u> law or the Law of Independent Assortment: During

gamete formation the segregation of one gene pair is independent of other gene pairs.
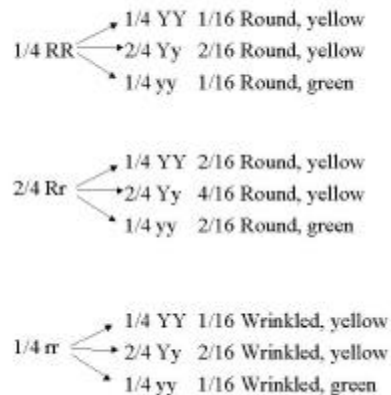
B. There are several ways to graphically represent this. It is important to understand all of them.

   1. Punnett Square

σ gametes

	RY $\frac{1}{4}$	Ry $\frac{1}{4}$	ry $\frac{1}{4}$	rY $\frac{1}{4}$
RY $\frac{1}{4}$	RR YY $\frac{1}{16}$	RR Yy $\frac{1}{16}$	Rr Yy $\frac{1}{16}$	Rr YY $\frac{1}{16}$
Ry $\frac{1}{4}$	RR Yy $\frac{1}{16}$	RR yy $\frac{1}{16}$	Rr yy $\frac{1}{16}$	Rr Yy $\frac{1}{16}$
ry $\frac{1}{4}$	Rr Yy $\frac{1}{16}$	Rr yy $\frac{1}{16}$	rr yy $\frac{1}{16}$	rr Yy $\frac{1}{16}$
rY $\frac{1}{4}$	Rr YY $\frac{1}{16}$	Rr Yy $\frac{1}{16}$	rr Yy $\frac{1}{16}$	rr YY $\frac{1}{16}$

$\female$ gametes

9 : 3 : 3 : 1

Round, yellow   Wrinkled, yellow

Round, green   Wrinkled, green

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   2. Branching method

1/4 RR
  • 1/4 YY   1/16 Round, yellow
  → 2/4 Yy   2/16 Round, yellow
  ▲ 1/4 yy   1/16 Round, green

2/4 Rr
  • 1/4 YY   2/16 Round, yellow
  → 2/4 Yy   4/16 Round, yellow
  ▲ 1/4 yy   2/16 Round, green

1/4 rr
  • 1/4 YY   1/16 Wrinkled, yellow
  → 2/4 Yy   2/16 Wrinkled, yellow
  ▲ 1/4 yy   1/16 Wrinkled, green

3. Statistical method
   Used the laws of Probability (Probability of an event is its frequency of occurrence over a very large number of trials.)

   or Probability = <u># of times event expected to happen</u>
                     # of opportunities for it to happen (or # of trials)

   For instance: With a quarter that you toss there only two possibilities if you toss it lands head side up or tails.   Doing this many times results in very nearly 1/2 times heads and 1/2 times tails. With a die, the probability of rolling a 4 is 1/6.

   a) Product Rule: The probability of two or more independent events (i.e A and B) occurring simultaneously is the product of their separate probabilities. Here the KEY WORD IS AND.

         P(A <u>and</u> B) = P(A)·P(B)

         For example, with a pair of dice the probability of getting two 4's- one on each dice (one 4 AND another 4) is
               P(4) ·P(4) = (1/6) (1/6) = 1/36

         For the genetic cross RrYy   X   RrYy
         The probability of rryy = P(rr) ·P (yy) = (1/4)(1/4) = 1/16

   b) Sum Rule: The probability of one or another of a set of mutually exclusive events occurring is the sum of the probabilities of the separate events. Here the KEY WORD IS OR

         P(A <u>or</u> B) = P(A) + P(B)

         With a pair of dice the probability of getting two 4's <u>or</u> two 6's on each dice is   P(4)·P(4) + P(6)·P(6) = (1/6)(1/6) + (1/6)(1/6) = 1/18

         For the genetic cross RrYy   X   RrYy
         The probability of rryy or RRYY =
         P(rr) ·P (yy) + P(RR) ·P (YY) = (1/4)(1/4) + (1/4)(1/4) = 1/8

C. Testing the hypothesis...... with a test cross
    F_1 RrYy X rryy ---------->
    1 yellow/round RrYy
    1 yellow/wrinkled Rryy
    1 green/round rrYy
   1 green/wrinkled rryy

VII. **MENDELIAN GENETICS IN HUMANS:**
Analysis is done via a <u>pedigree analysis</u> (analysis of a family history to find patterns of genetic transmission of traits.

A. Rare Recessive Traits
(because it is rare, unrelated individuals who do not have affected children are presumed homozygous dominant)
   1. Parents not usually affected because it is rare and recessive.
    2. Unaffected parents can produce affected children if both are heterozygous.
    3. Two affected parents always have affected offspring because they are homozygous recessive.
    4. Approx. 1/4 of siblings of affected individuals are affected provided that parents were heterozygous (based on 3:1 Mendelian ratios).
    5. Recessive traits often result from consanguineous matings.
    6. Examples PKU (phenylketonuria), CF (cystic fibrosis), and albinism
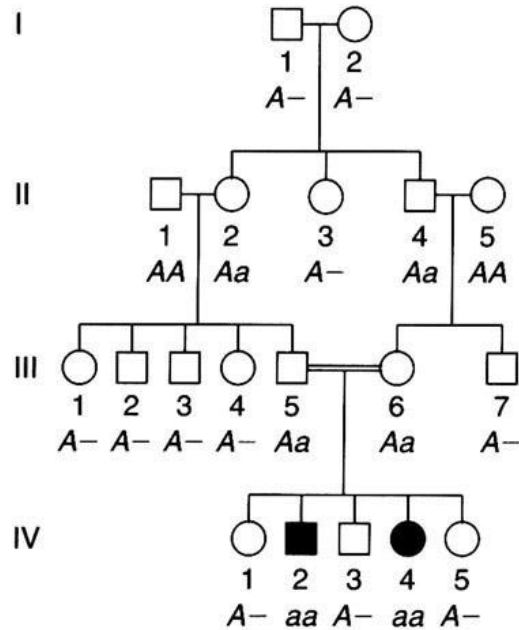
B. Rare, Dominant Traits

Because it is rare, assume affected people of unknown genotype (either AA or Aa) are Aa. An affected person must have at least one A allele because they are affected. The assumption that the second allele is an "a" is made because the A allele is RARE and so it is more likely that they would have an "a" for the second allele vs. an "A"Trait occurs every generation .

    1. At least one parent affected
    2. With one parent affected, approx. ½ of the children are affected (because of Mendelian ratios)
    3. Unaffected individuals cannot produce offspring (because they are homozygous recessive).
    4. Two affected parents can produce unaffected offspring if they are both heterozygotes and pass on the recessive alleles.
    5. Examples include achondroplasia (dwarfism), Huntington's Disease, Polydactyly (extra digits), Brachydactyly (short fingers).

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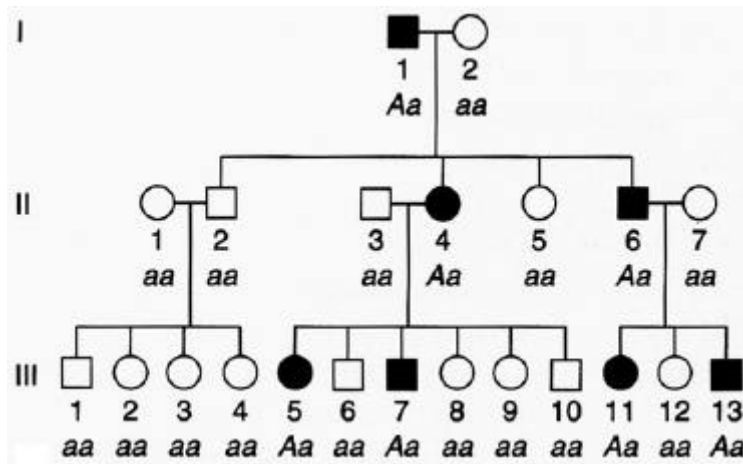
C. Normal phenotypic variation
    1. Continuous – phenotypes arranged around a continuous spectrum.
    2. Discontinuous – phenoytpes arranged into distinct classes
        a) Polymorphism = a set of two or more common alternative phenotypes
        (i.e. PTC tasting)

□, ○ = Tasters (*TT* or *Tt*)
■, ● = Nontasters (*tt*)