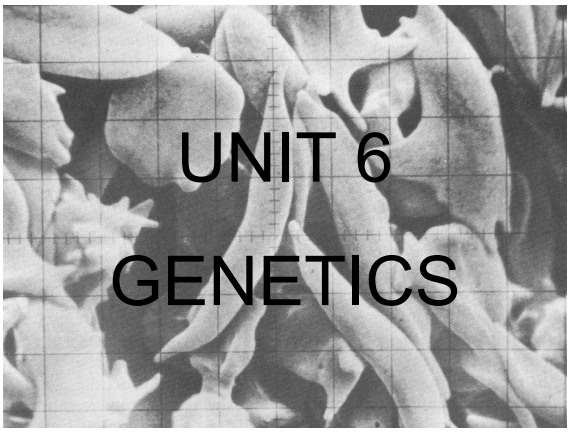


III. Mendel and Heredity (6.3)

A. **Mendel laid the groundwork for genetics**

1. **Traits** are distinguishing characteristics that are inherited.

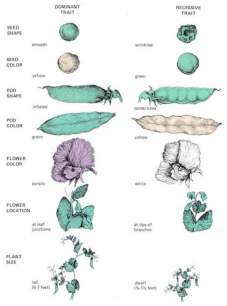
2. **Genetics** is the study of **biological inheritance patterns** and variation.

3. Gregor Mendel showed that traits are inherited as **discrete units.**

4. Many in Mendel's day thought traits were blended.

B. Mendel's data revealed **patterns of inheritance**

1. Mendel studied plant variation in a monastery garden



2. Mendel made <u>three key decisions</u> in his experiments

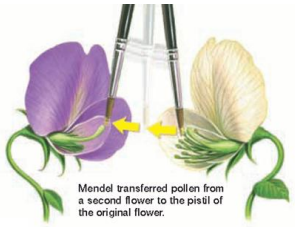
a. **Control over breeding**

b. Use of **purebred** plants

c. Observation of "**either-or**" traits (only appear two alternate forms)

3. Experimental design

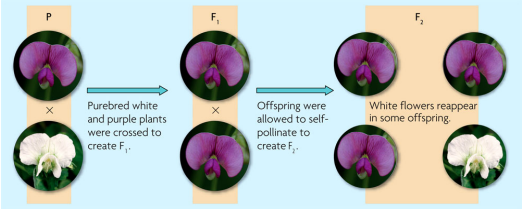
a. Mendel chose **pea plants** because reproduce quickly and could control how they mate



b. Crossed purebred white-flowered with purebred purple-flowered pea plants.

1). Called **parental**, or **P generation**

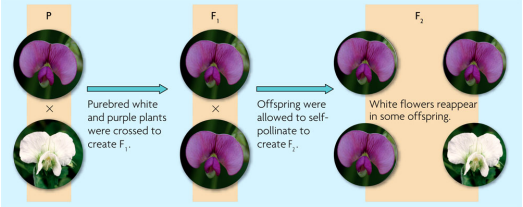
2). Resulting plants (first filial or **F1 generation**) all had purple flowers



c. Allowed F1 generation to self-pollinate

1). Produced **F2 generation** that had both plants with purple and white flowers)

2). Trait for white had been “hidden”, it did not disappear.

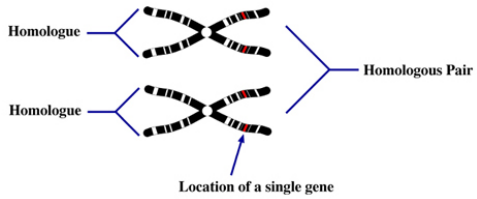


d. He began to observe **patterns**- Each cross yielded similar ratios in F2 generation (**3/4 had purple, and 1/4 white**)

FIGURE 6.10 MENDEL'S MONOHYBRID CROSS RESULTS			
F_2 TRAITS	DOMINANT	RECESSIVE	RATIO
Pea shape	5474 round	1850 wrinkled	2.96:1
Pea color	6022 yellow	2001 green	3.01:1
Flower color	705 purple	224 white	3.15:1
Pod shape	882 smooth	299 constricted	2.95:1
Pod color	428 green	152 yellow	2.82:1
Flower position	651 axial	207 terminal	3.14:1
Plant height	787 tall	277 short	2.84:1

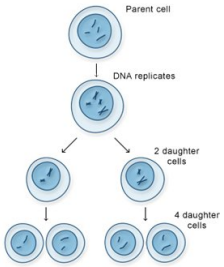
4. Mendel made three important conclusions

a. **Traits are inherited as discrete units** (explained why individual traits persisted without being blended or diluted over successive generations)



b. Two other key conclusions collectively called the **law of segregation**

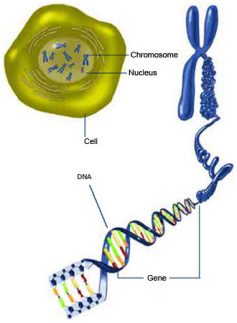
1). Organisms **inherit two copies** of each **gene**, one from each parent



2). **Organisms donate only one copy** of each gene in their gametes (two copies of each gene segregate, or separate, during gamete formation

IV. Traits, Genes, and Alleles (6.4)

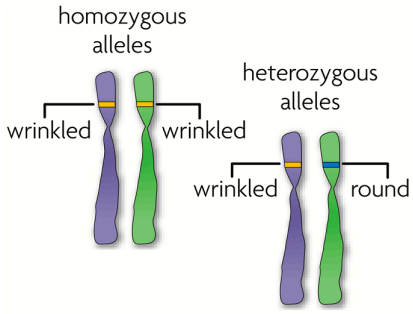
A. The **same gene** can have **many versions**



1. **gene**- a "piece" of DNA that provides a set of instructions to a cell to make a certain **protein**. *(Proteins are either structural or functional)*

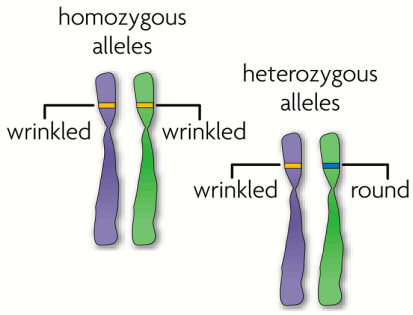
a. Most genes exist in many forms (called **alleles**)

b. You have <u>two alleles</u> for each gene`



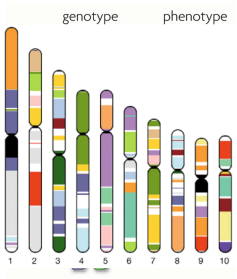
2. **Homozygous**- means two of <u>same</u> allele

3. **Heterozygous**- two <u>different</u> alleles



B. Genes influence the development of traits

1. **Genome**- is all the organisms genetic material

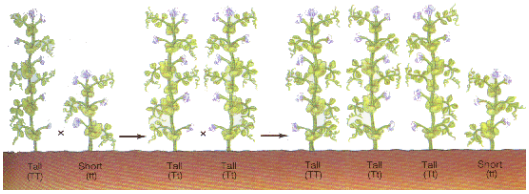


2. **Genotype**- refers to ...netic makeup of a specific ... of genes

Phenotypes- physical

C. Dominant and Recessive Alleles

1. **Dominant alleles**- allele that is expressed when two different alleles or two dominant alleles are present (use capital letter to represent. *(I.e. capital T)*

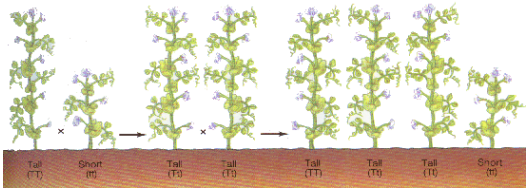


2. **Recessive alleles**- only expressed if have two copies of recessive present (use small-case letter to represent)

3. **Homozygous dominant = TT**

4. **Heterozygous = Tt**

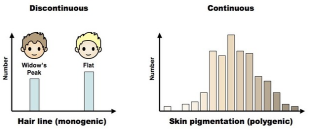
5. **Homozygous recessive = tt**



D. Alleles and Phenotypes

1. Both homozygous dominant and heterozygous genotypes yield a dominant phenotype. *(I.e. TT and Tt)*

2. Most traits occur in a **range** and <u>do not</u> <u>follow</u> simple dominant-recessive patterns

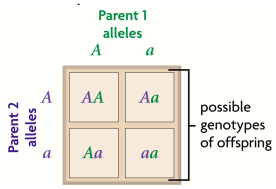


V. Traits and Probability (6.5)

A. **Punnett squares** illustrate genetic crosses

1. Used to **predict possible genotypes** resulting from a cross

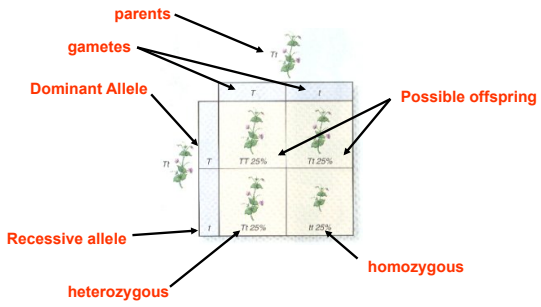
a. Axes of grid represent possible **gamete** genotypes of each



b. Boxes show **genotypes** of **offspring**

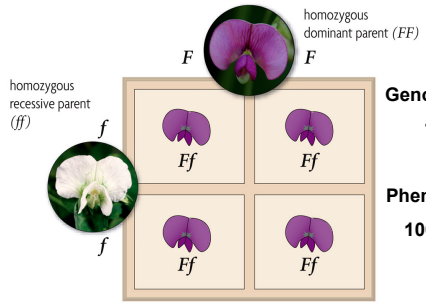
c. Can determine **ratio** of genotypes in each generation

Punnett Square



B. **Monohybrid cross involves one trait**

1. **Homozygous dominant X Homozygous recessive**



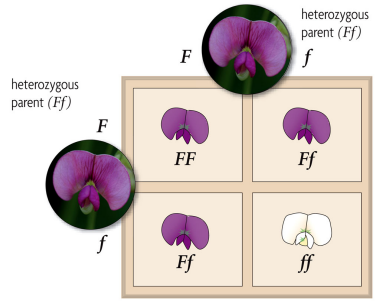
Genotypic ratio

100% Ff

Phenotypic ratio

100% purple

2. **Heterozygous X Heterozygous**



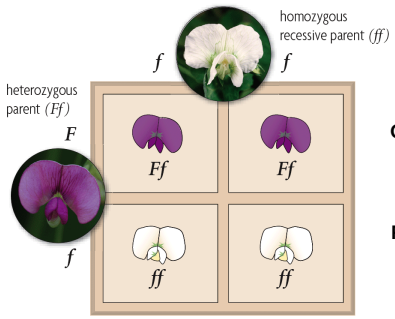
Genotypic ratio

1:2:1

Phenotypic ratio

3:1

3. **Heterozygous X Homozygous recessive**



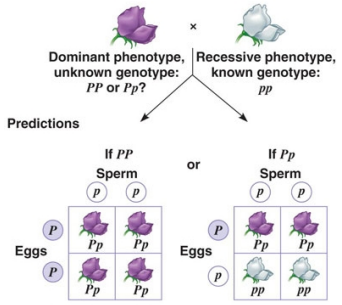
Genotypic ratio

1:1

Phenotypic ratio

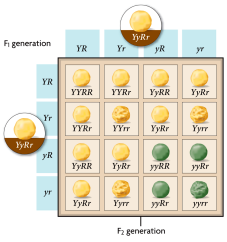
1:1

C. **Test Cross**- a cross between organism with an and an organism with a **unknown genotype recessive phenotype**

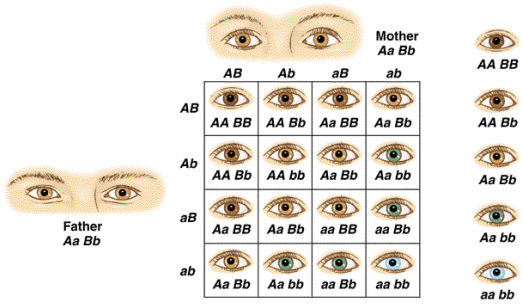


D. **Dihybrid** cross involves <u>two traits</u>

1. Mendel also conducted dihybrid crosses- wondered if both traits would always appear <u>together</u> or if they would be expressed <u>independently</u> of each other

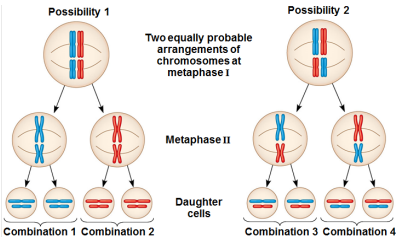


2. Mendel discovered phenotypic ratio in F2 generation as always **9:3:3:1** regardless of combination traits he used



3. Mendel's dihybrid crosses led to his second law,the **law of independent assortment.**

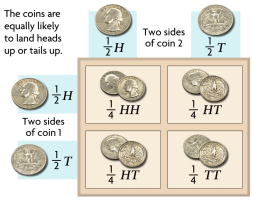
4. The law of independent assortment states that **allele pairs** separate independently of each other **during meiosis**



E. Heredity patterns can be calculated with probability

1. **probability** - the likelihood that a particular event will happen

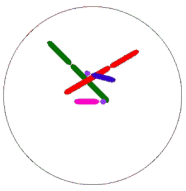
2. Probability applies to random events such as **meiosis** and **fertilization**



VI. Meiosis and Genetic Variation (6.6)

A. **Sexual reproduction** creates **unique gene combinations**

1. Sexual reproduction creates unique combination of genes

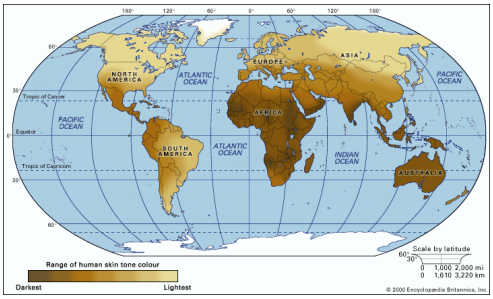


a. **independent assortment** of chromosomes in meiosis

b. **random fertilization of gametes**

2. 2^{23} possible sperm or egg cells produced

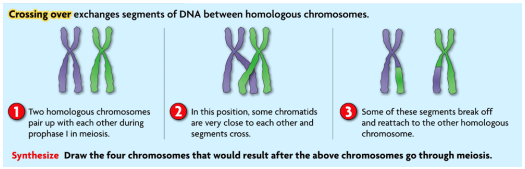
2^{23} X 2^{23} = about 70 trillion different combinations of chromosomes



B. Crossing over during meiosis increases genetic diversity

1. **crossing over** - exchange of chromosome segments between homologous chromosomes during Prophase I of Meiosis I

2. Results in **new combination of genes**

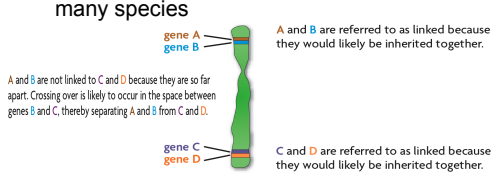


C. **Linked genes** - genes located on the same chromosome inherited together.

1. **Closer together** they are **high** chance of inheriting together

2. If **genes far apart**, **crossing-over may separate them**

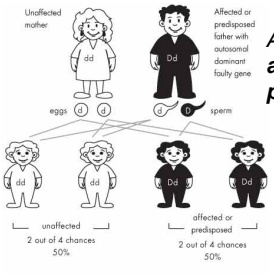
3. **Gene linkage** used to build **genetic map** of many species



UNIT 6: GENETICS

Chapter 7: Extending Mendelian Genetics

I. Chromosomes and Phenotype (7.1)

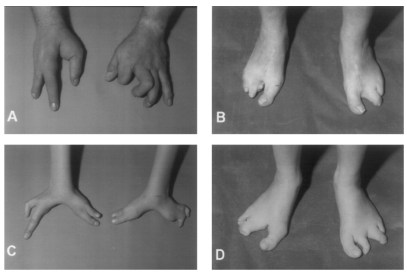


*A. <u>Two</u> copies of each **autosomal** gene affect **phenotype***

*1. Most human **traits** are <u>result</u> of **autosomal genes***

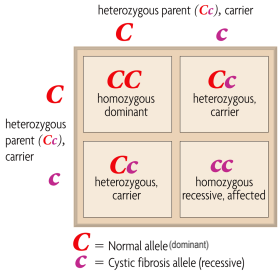
*2. Many human **genetic disorders** also <u>caused by</u> **autosomal genes***

a. Chance of having disorder can be predicted
b. Use same principles as Mendel did



B. Disorders Caused by **Recessive Alleles**

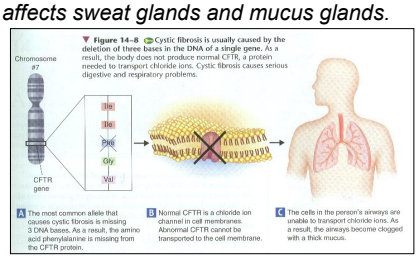
1. Some **disorders** <u>caused</u> by **recessive alleles** on autosomes



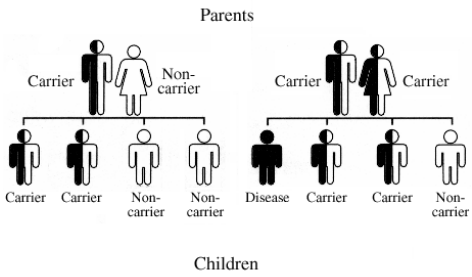
*2. Must have **two copies** of **recessive allele** to have disorder*

*a. Disorders often appear in offspring of parents who are **heterozygous***

*b. **Cystic Fibrosis**- recessive disorder that affects sweat glands and mucus glands.*

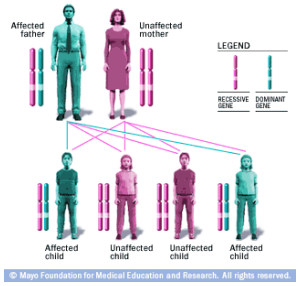


3. A person who is <u>heterozygous</u> for disease is called a **carrier**- does not show disease symptoms



*C. Disorders Caused by **Dominant Alleles***

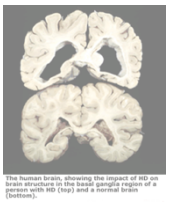
*1. **Less common** than recessive disorders*

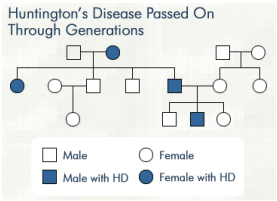


2. **Huntington's Disease**- damages nervous system and usually appears during adulthood.

a. **75% chance** if both parents **heterozygous**

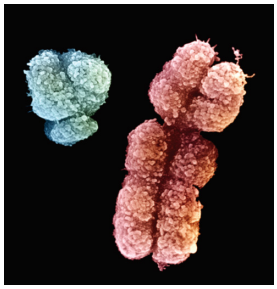
b. Since disease strikes later in life, person can have children before disease appears. Allele is passed on even though disease is fatal





E. Males and Females can differ in sex-linked traits

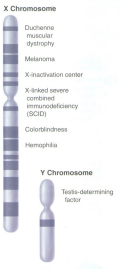
*1. Mendel figured out much about **heredity**, but did <u>not</u> know about **chromosomes***



*a. Mendel only studied **autosomal** traits*

*b. Expression of genes on **sex chromosomes** <u>differs</u> from **autosomal genes***

2. Sex-linked Genes



a. Genes located on sex-chromosomes called **sex-linked genes**

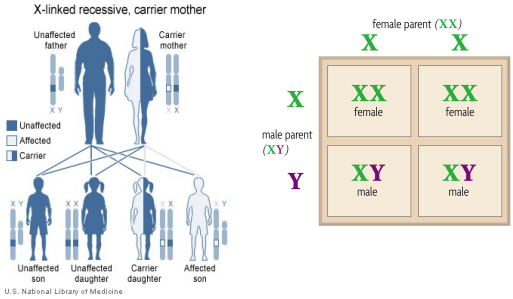
b. Many species have specialized sex chromosomes

1). In mammals and some other animals, individuals with **XX are female** and **XY are male**

2). **X** chromosome <u>much larger</u> than the **Y**

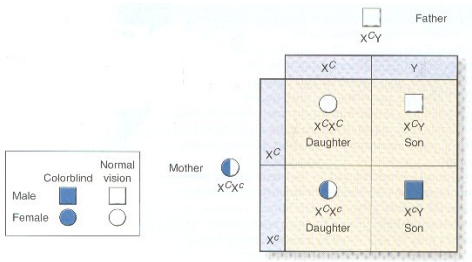
3. Expression of Sex-Linked Genes

a. **Males** only have <u>one copy</u> of each chromosome (**XY**)



1). **Express all alleles** on each chromosome

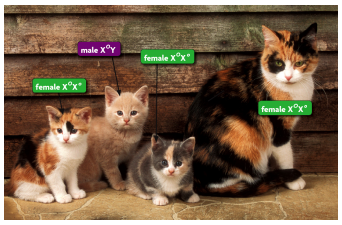
2). No second copy of another allele to mask effects of another allele (**all recessive alleles expressed**)



b. In each cell of female, one of two X-chromosome is randomly "**turned off**".

1). Called **X Chromosome Inactivation**

2). Creates **patchwork** of two types of cells



II. Complex Patterns of Inheritance (7.2)

A. Phenotypes can depend on interactions of alleles

1. Many traits are result from alleles with **range of dominance**, rather than a strict dominant and recessive relationship

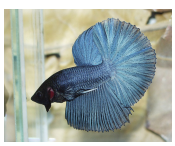
2. In many cases, phenotypes result from multiple genes

B. ***Incomplete Dominance***

1. Neither allele completely dominant

2. ***Heterozygous phenotype*** *somewhere between homozygous phenotypes ("blending")*

Phenotype	Genotype	Phenotype	Genotype	Phenotype	Genotype
green	B_1B_1	Steel blue	B_2B_2	Royal blue	B_1B_2



C. ***Codominance***

1. ***Both traits are expressed completely***

2. Can sometimes look like "blending" of traits, but actually show ***mixture of both***


red

X


white

=


roan

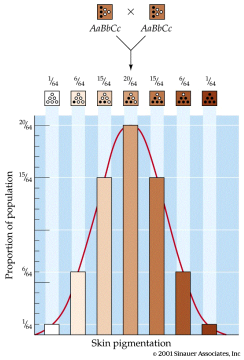
3. **Human blood type** is example of codominance

a. Also has 3 different alleles- trait also considered a **multiple-allele trait**

PHENOTYPE (BLOOD TYPE)		GENOTYPES
A	antigen A	I^AI^A or I^Ai
B	antigen B	I^BI^B or I^Bi
AB	both antigens	I^AI^B
O	no antigens	ii

b. When alleles are neither dominant of recessive (in both incomplete and codominance) use **upper case letters** with either **subscripts** or **superscripts**)

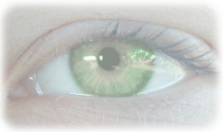
D. Many genes may interact to produce one trait



1. ***Polygenic traits****- two or more genes determine trait*

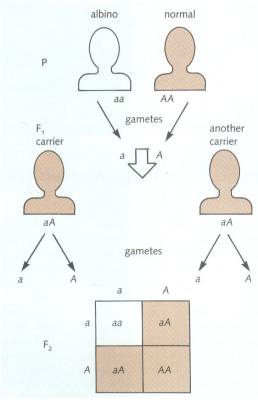
a. ***Skin color*** *result of four genes that interact to produce range of colors*

b. ***Human eye color*** *shows at least 3 genes (hypothesize that are still genes undiscovered as well)* ***Order of dominance: brown > green > blue.***



GENE NAME	DOMINANT ALLELE	RECESSIVE ALLELE
BEY1	brown	blue
BEY2	brown	blue
GEY	green	blue

2. ***Epistasis****- when one gene overshadows all of the others. Albinism is caused by this type of gene*



Albinism is a autosomal recessive trait. Because the allele is recessive, individuals who are heterozygous for the trait express their normal skin color, sot he presence of the allele is "hidden" by the dominance of the normal allele. Albinos are unable to synthesize melanin, the pigment molecule responsible for most human skin coloring

3. The **environment** interacts with **genotype**

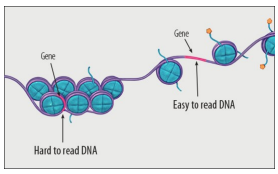
a. **Phenotype** is <u>more than sum</u> of **gene expression**

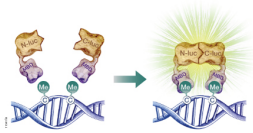
b. Sex of sea turtles depends on genes and **environment**. **Temperature** when eggs develop determine sex

c. **Human** traits also affected by **environment** (nutrition and health care)

d. **Epigenetics-** Epigenetics is the study of potentially heritable changes in gene expression (active versus inactive genes) that does not involve changes to the underlying DNA sequence

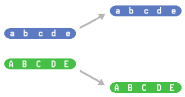
1. A change in the heritable phenotype without a change in genotype
2. This in turn affects how cells read the genes





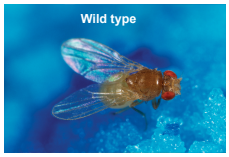
III. Gene Linkage and Mapping (7.3)

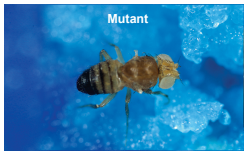
*A. Gene linkage was explained through **fruit flies***



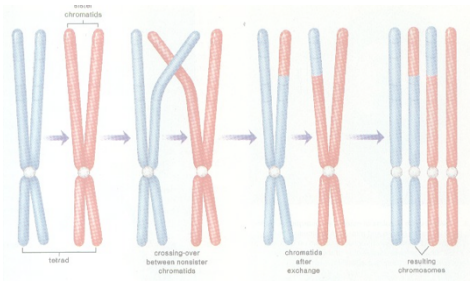
*1. **Thomas Hunt Morgan** worked with fruit flies (Drosophila melanogaster)*

*2. Some traits seemed to be inherited together. Morgan called them **linked traits. (found on same chromosome)***





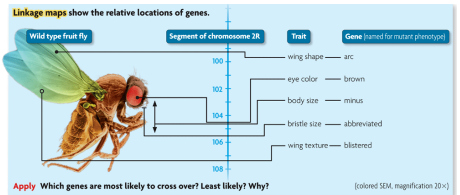
*3. Morgan concluded that because linked genes were not inherited together every time that chromosomes must exchange homologous genes during meiosis (**crossing over**)*



*B. Linkage maps estimate **distances** between genes*

*1. **Closer together**- more likely **inherited together***

*2. **Further apart**- more likely will be **separated** during meiosis.*



IV. Human Genetics and Pedigrees (7.4)

A. Human genetics follows the patterns seen in other organisms

1. Meiosis **independently assorts chromosomes** when gametes are made for sexual reproduction

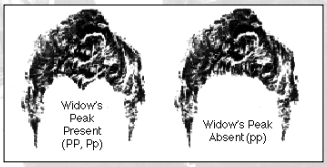
2. Human heredity involves same relationships between alleles (dominant/recessive, polygenic, sex-linked, etc)

B. Inheritance of some traits very complex

*1. **Multiple genes** and **alleles** can interact*

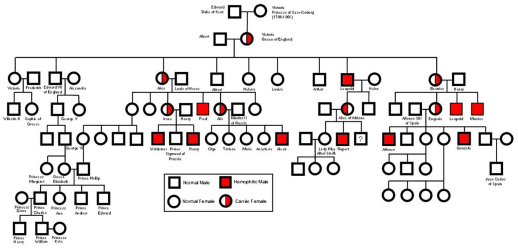
2. Single-gene traits can still be observed

a. Many examples of single-gene traits (hairline-widows peak)



b. Many genetic disorders also caused by single-gene traits (Huntington's disease, hemophilia, Duchenne's muscular dystrophy)

c. Much of what is known about human genetics comes from studying genetic disorders

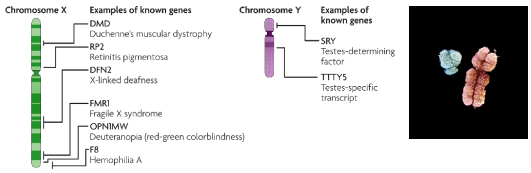


THE HISTORY OF HEMOPHILIA IN THE ROYAL FAMILIES OF EUROPE

C. Females can carry a sex-linked genetic disorder

*1. Both **male and females** can be carriers of **autosomal disorders***

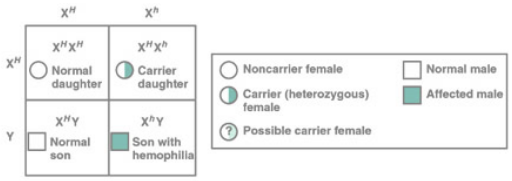
*2. **Only females** can be carriers of **sex-linked disorders***



3. Many genetic disorders carried on **X-chromosome**

a. **Male** who has gene for disorder on X-chromosome will have disorder

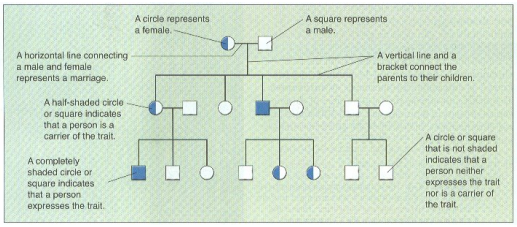
b. **Males** more likely to have this **disorder**



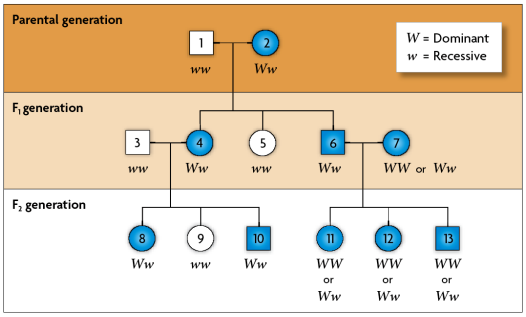
D. A pedigree is a chart for tracing genes in a family

1. **Phenotypes** are used to <u>infer</u> **genotypes** on a pedigree

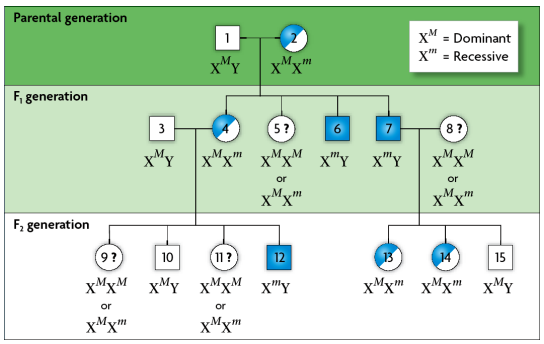
2. **Autosomal genes** show <u>different patterns</u> on a pedigree than **sex-linked genes.**



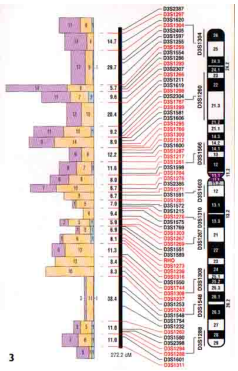
a. **Autosomal genes**



b. ***Sex-linked genes***



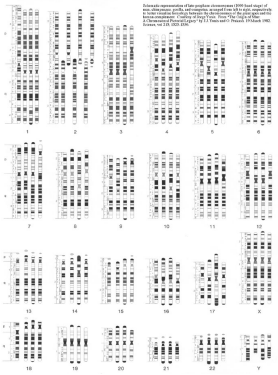
E. Several methods help map human chromosomes



*1. Human **genome so large** difficult to map*

2. Several methods used

*a. **Pedigrees** used for studying genetics in family*



F. **Human Genes-** human genome contains 20,000 to 22,000 genes. Much lower than earlier estimates of 80,000 to 140,000

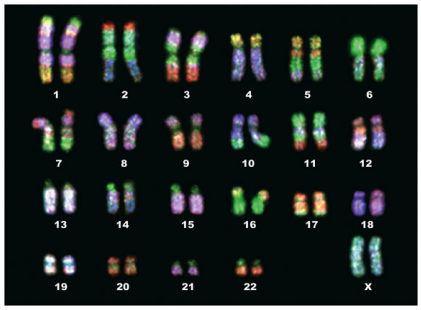
1. Contains 3164.7 million chemical nucleotide bases (A, C, T and G)

2. 99.9% of all nucleotide bases are exactly the same in all people

3. Less than 2% of genome actually codes for proteins

G. ***Karyotypes**- picture of all chromosomes in a cell*

1. *Stains used to produce patterns of bands*



*2. Used to identify certain genetic disorders in which there are **extra** or **too few chromosomes** (i.e. Down syndrome*