Mendelian Inheritance

cont.

Inheritance patterns are often more complex than predicted by simple Mendelian genetics

- The relationship between genotype and phenotype is rarely as simple as in the pea plant characters Mendel studied.
- Many heritable characters are not determined by only one gene with two alleles.
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance

Mendelian Inheritance

In a random mating/crossing event, the ratios of genotypes and phenotypes are known and the ratios can be determined if they behave according to Mendel's Laws

P - F1 – F2 – F3 – F4 (Sexual reproduction)

Exceptions To Mendel's Original Principles

- Incomplete dominance
- Co-dominance
- Multiple alleles
- Polygenic traits
- Epistasis

- Pleiotropy
- Environmental effects on gene expression
- Linkage
- Sex linkage

Some inheritance patterns are exceptions to standard Mendelian inheritance

- There are **two** normal exceptions to Mendelian genetics, it involves:-
  - genes located in the **nucleus**, and
  - genes located outside the nucleus (**plastids**).

- In both cases, the **sex of the parent** contributing an allele is a factor in the pattern of inheritance

1) **Non Mendelian inheritance patterns due to genes located in the Organelles**

Inheritance of Organelle Genes

- **Extra nuclear** genes (*or* cytoplasmic genes) are found in **organelles** in the cytoplasm.
- Mitochondria, chloroplasts, and other plant plastids carry **small circular** DNA molecules.
- Extra nuclear genes are inherited **maternally** because **the zygote's cytoplasm** comes from the **egg**.

Inheritance of organelle genes

- Show non-Mendelian inheritance
- Meiosis-based segregation doesn't occur
- Mendelian ratios aren't observed

- Results of reciprocal crosses are different than those involving nuclear genes
- In humans, mtDNA markers can be tracked using molecular techniques

Mitochondria are essential for oxidative phosphorylation (make ATP)

- However they play important roles in most other metabolic functions of the cell:
- Pyrimidine synthesis
- Heme synthesis (red blood precursor cells)
- Amonia detoxification (liver)
- Cholesterol metabolism
- Sex hormone synthesis
- Free radical production and detoxification
- Apoptosis

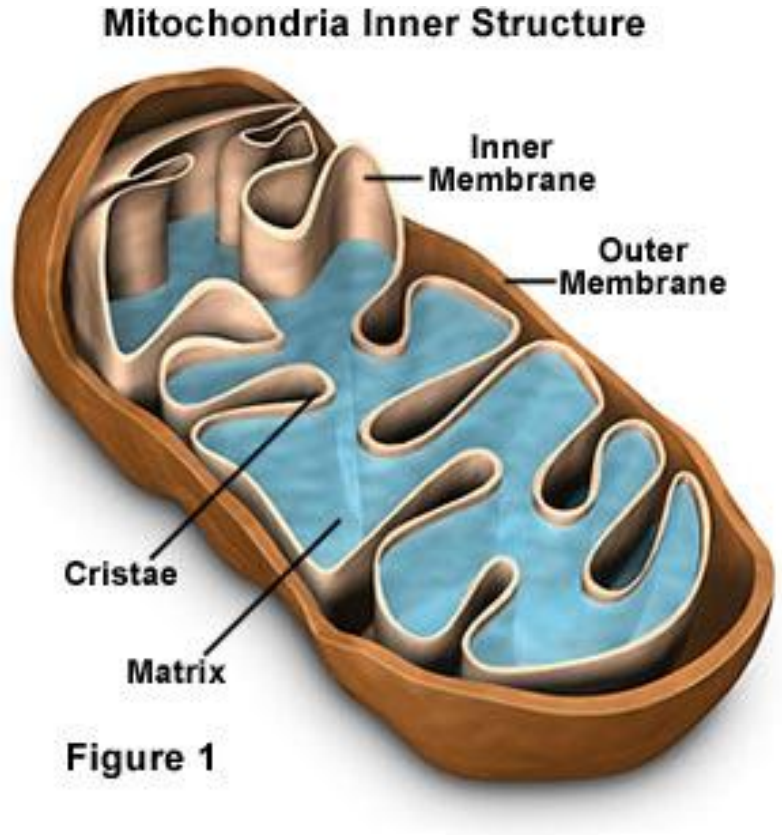
- Some defects in mitochondrial genes prevent cells from making **enough ATP** and result in diseases that **affect the muscular and nervous systems**

Example

1. **Mitochondrial myopathy** – a group of neuromuscular diseases caused by damage to the mitochondria

2. **Leber's hereditary optic neuropathy** (**LHON**)is a degeneration of **retinal ganglion cells** (RGCs) and their **axons** that leads to an acute or sub-acute loss of central vision.

Mitochondrial DNA



- Each cell contains **thousands** of mito, each containing copies of its **DNA**
- **Mito** DNA is in larger **quantities** in a cell
- Nuclear DNA is **larger** in **size**

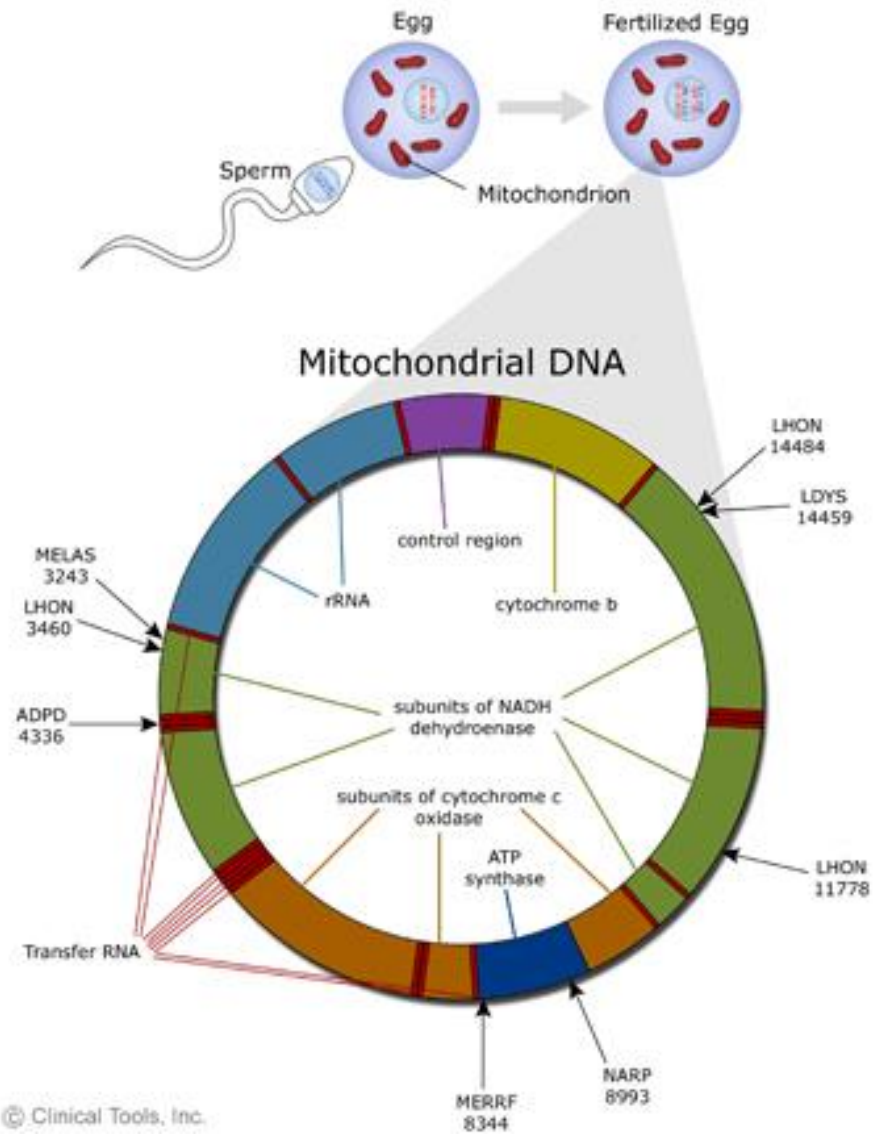
Mt DNA is inherited from mother

- Every sibling will get their mt DNA from their mother

- Why?

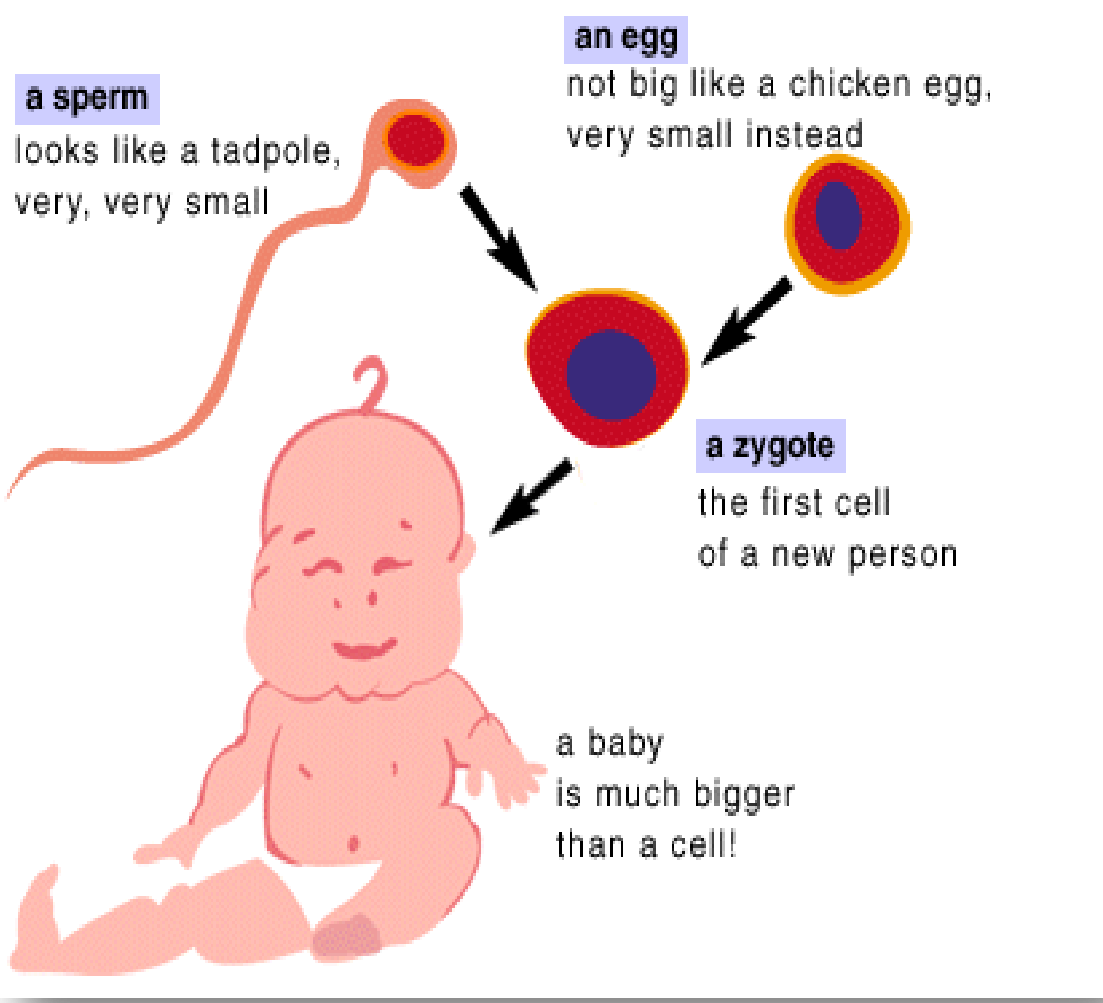
Why from mother?

- Egg contains 23 chromosomes and cell cytoplasm which contains thousands of maternal mitochondria.
- Sperm contains 23 chromosomes with very little cytoplasm

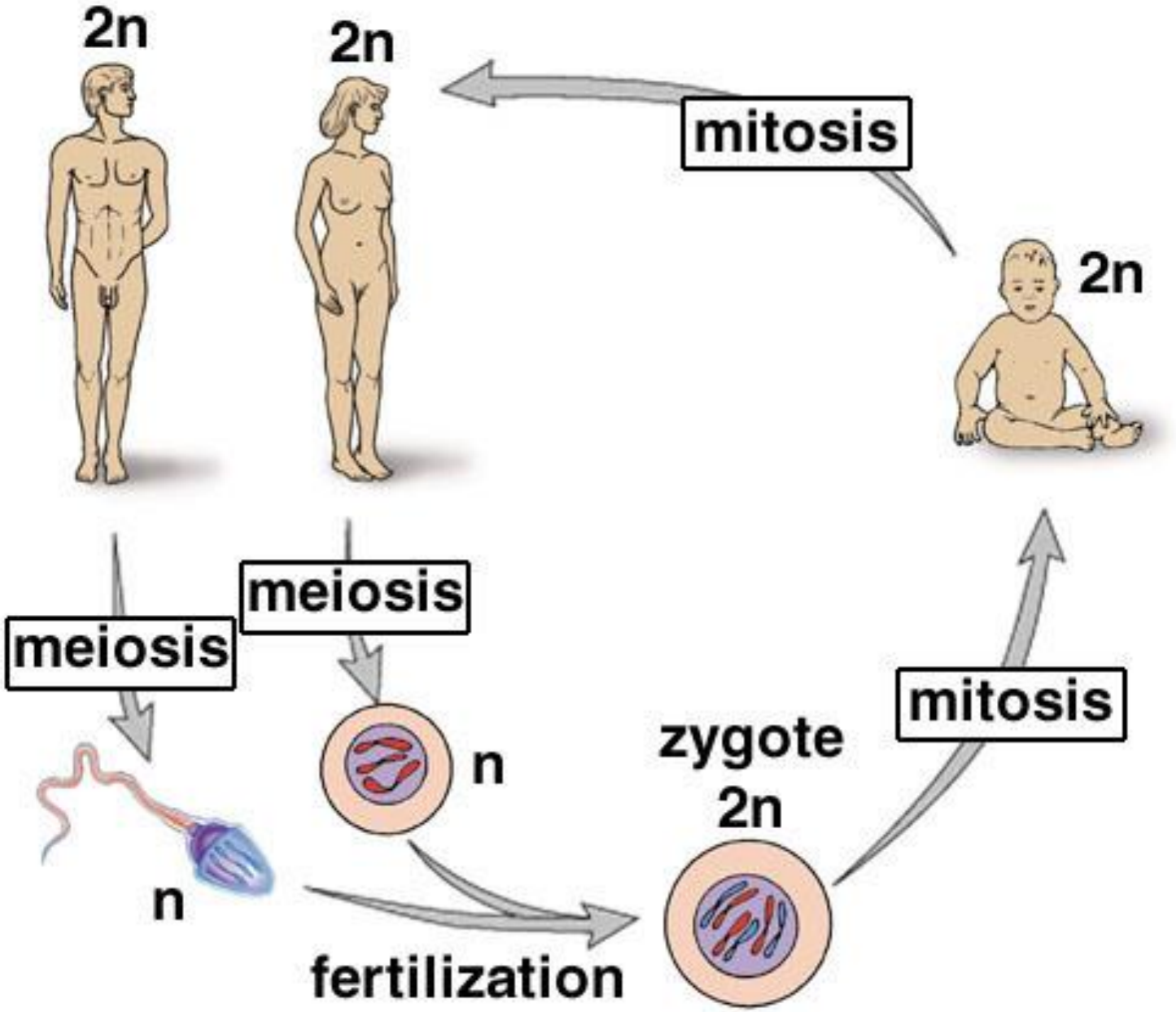


Zygote = Fertilized Egg

- Once the sperm enters the egg, those sperm mitochondria are usually **destroyed**.
- The zygote ends up filled with mitochondria from the **egg**, therefore the zygote **inherits** the **maternal** mitochondrial DNA.



Life Cycle of Humans



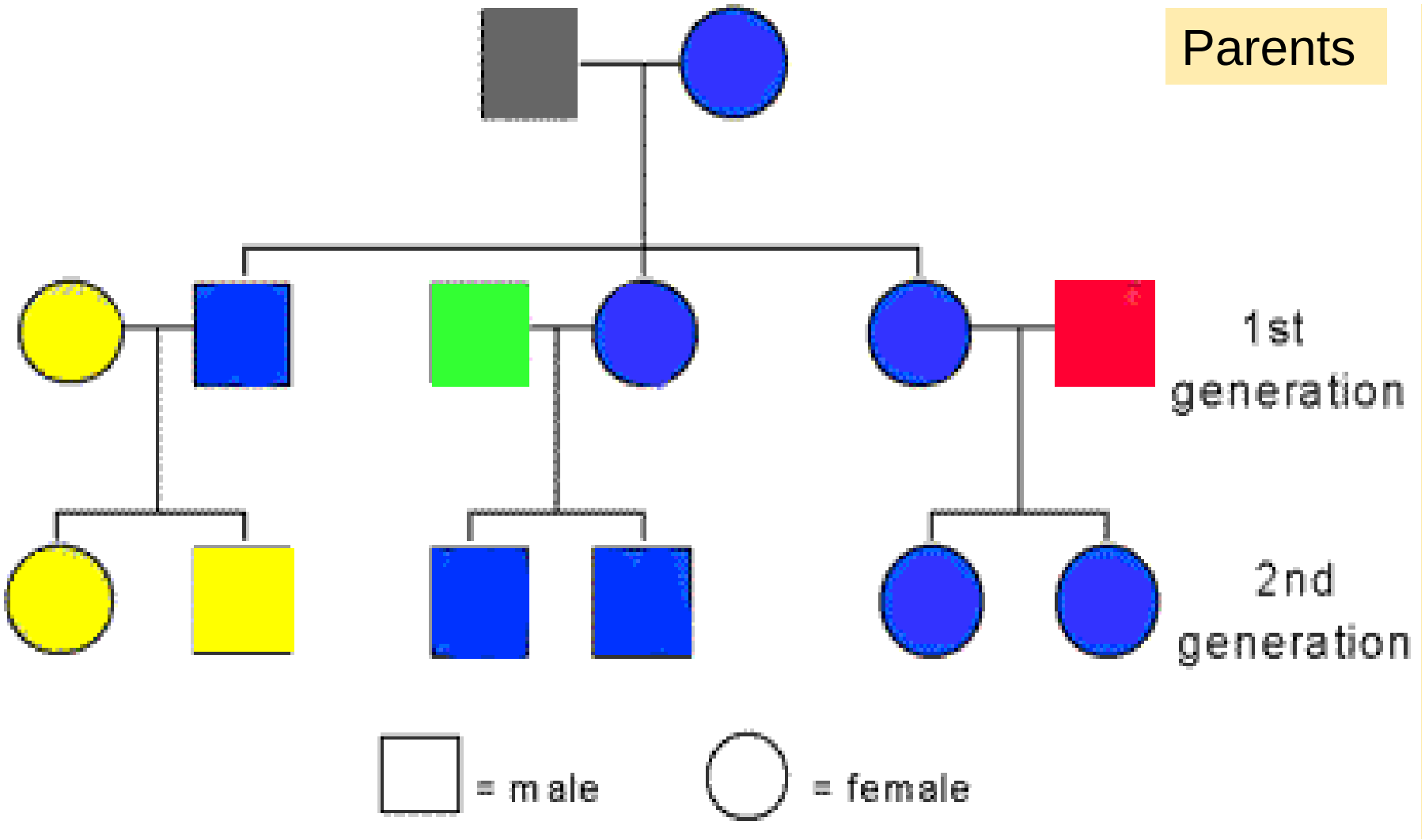
Mt DNA is 16,569 bases in length and consists of 2 different regions

- **Coding Region**:
  - Produces **13** proteins, **22**tRNAs, **2**rRNAs needed for **cell respiration**
  - This region has very little variability
  - So everyone's DNA in this region will be nearly the same sequence of TGCAs
- **Control Region:**

  This region is highly variable within the human population

Mutations occur in the **control region** of mito DNA at a regular rate and are **passed** onto children by the **mom**.

Maternal Inheritance Pattern with Mt DNA



How can we use this information?

- We can compare DNA from the **controlling** region to other living humans
  - See how **related** to you are **to each other**
- Compare to prehistoric remains of **human fossils**
  - Identify where your DNA **originated**
  - Identify **ancestral** relationships between modern populations
- Compare your highly variable regions to **other species**

- **Comparisons can be made by how many variations exist between her DNA and our DNA.**

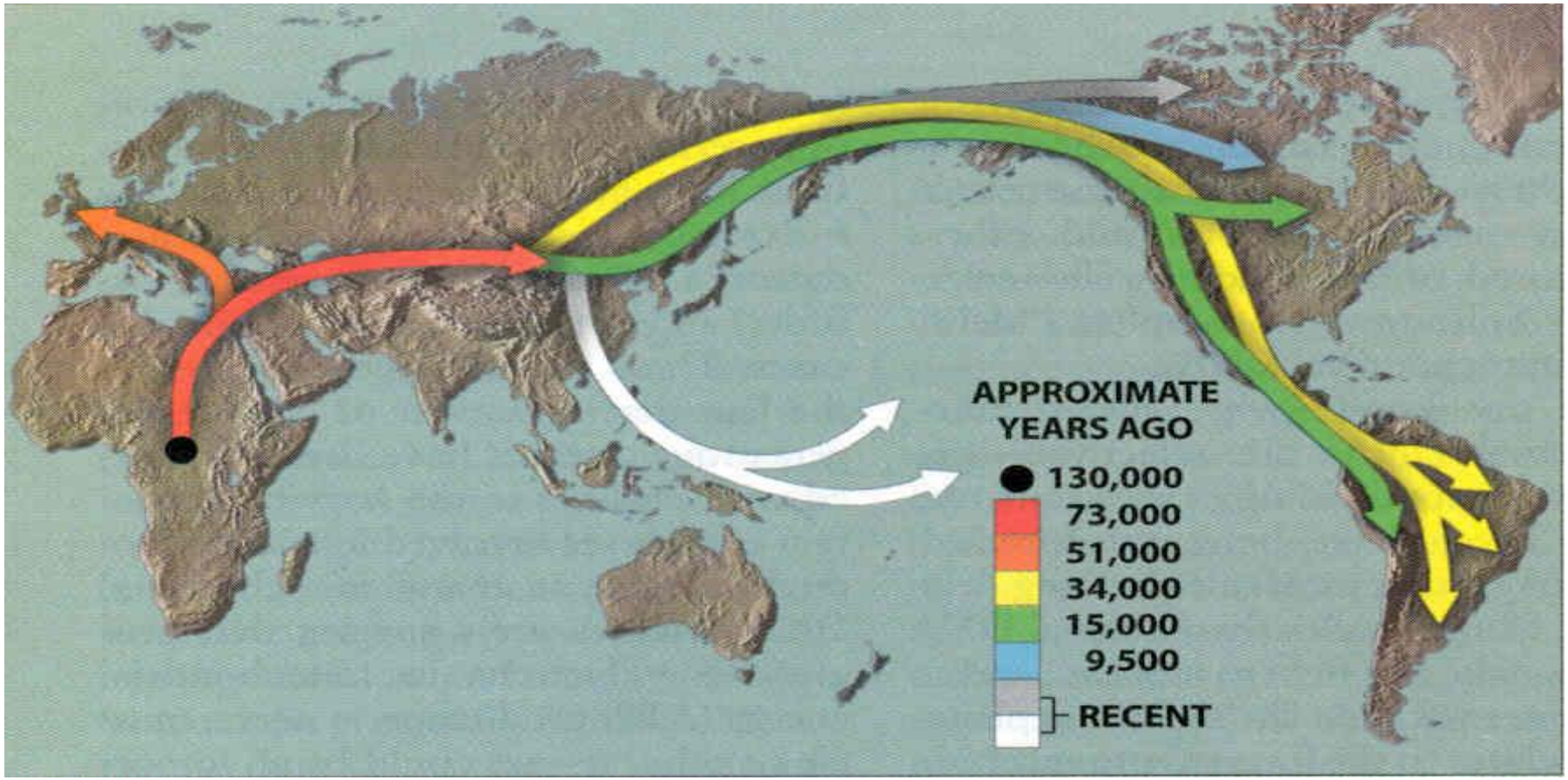
Mitochondrial DNA and Migrations

- Because mitochondrial chromosomes **don't recombine like nuclear chromosomes**, and because they are **maternally inherited**, mutations don't spread laterally through a population because of mating.

- Instead, they are passed, almost **clonally**, down through subsequent generations, often becoming "fixed" when small populations or individuals move to new areas.

- Mitochondrial genetic markers are therefore the best tools to follow human **migrations** that have taken place over the **millennia**.

Mitochondrial DNA polymorphisms track human migrations



- All humans descend from a small group of Africans
- This group originated in central Africa **~200,000** years ago
- The founding group was small (10^2-10^4 people)
- **Descendants of this group replaced all other hominids everywhere in the world**

But, **mussels** are unique among animals !!!

Biparental (maternal & paternal) inheritance is common in mussels of the genus ***Mytilus***

2) Non Mendelian inheritance patterns due to genes located in the nucleus

(a) Degrees of Dominance

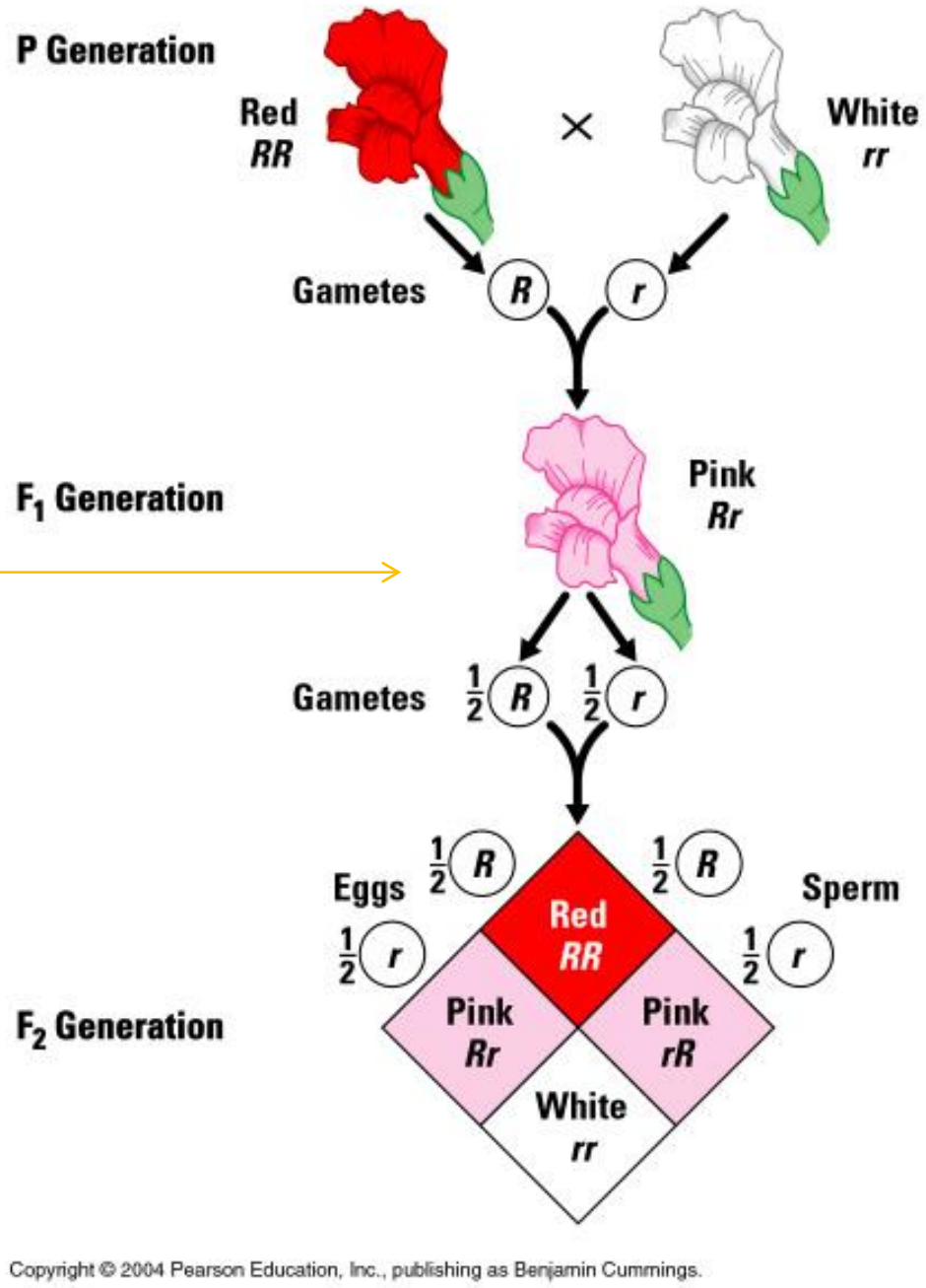
i. **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical.

ii. **Incomplete dominance:-** the phenotype of $\mathbf{F_1}$ hybrids is somewhere **between** the phenotypes of the two parental traits.

iii. **Codominance: -** two dominant alleles affect the phenotype in separate, distinguishable ways – i.e. **when two alleles are both expressed (neither masks the other)**

Incomplete dominance in snapdragon color

- In incomplete dominance F_1 hybrids have an appearance in between the phenotypes of the two parents



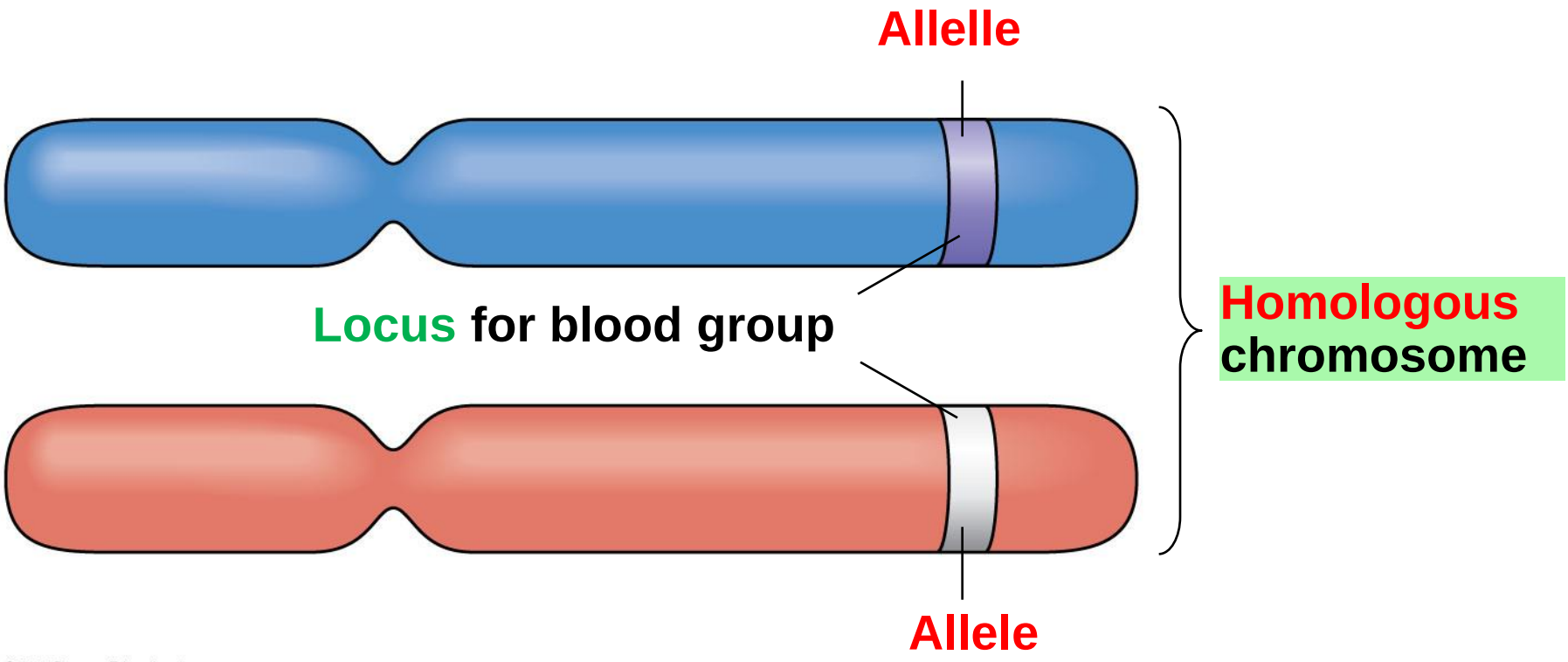
Frequency of Dominant Alleles

- Dominant alleles are **not** necessarily more common in populations than recessive alleles.
- For example, one baby out of 400 in the United States is born with extra fingers or toes – **Polydactyl**
- The **allele** for this **unusual trait** is **dominant** to the allele for the more common trait of **five digits** per appendage.
- In this example, the **recessive** allele **is far more prevalent** than the population's dominant allele

Linked genes tend to be inherited together because they are located near each other on the same chromosome

- **Each** chromosome has **hundreds** or **thousands** of genes (**except** the **Y** chromosome).
- Genes located on the **same chromosome** that **tend to be inherited together** are called **linked genes.**

Alleles, Locus



MULTIPLE ALLELISM

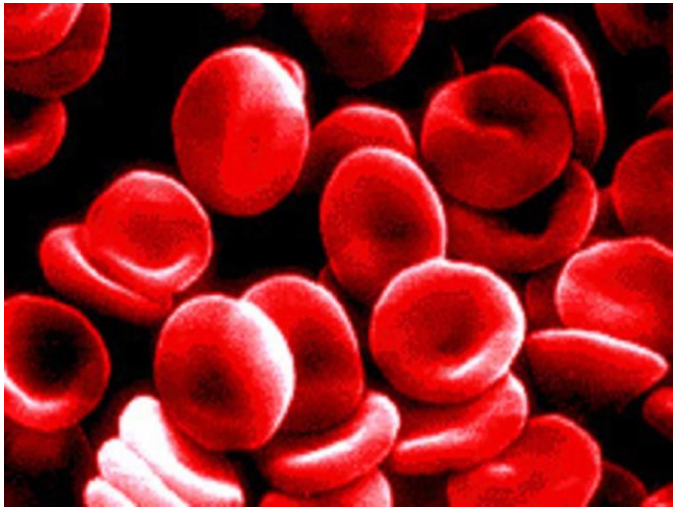
- When there is more than 2 alleles possible for a given gene.
- Allows for a larger number of genetic and phenotypic possibilities.
- Human blood type is an example of both **codominance** and a trait with **multiple alleles**.

- For example, the **four** phenotypes of the **ABO** blood group in humans are determined by three alleles for the enzyme (***I***) that attaches **A** or **B** carbohydrates to red blood cells: I^A, I^B, and *i*.

- The enzyme encoded by the I^A allele **adds** the **A** carbohydrate, whereas the enzyme encoded by the I^B allele adds the **B** carbohydrate; the enzyme encoded by the *i* allele adds neither

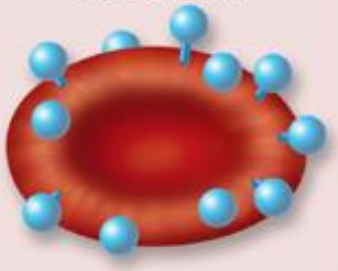
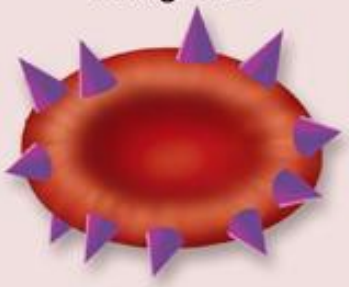
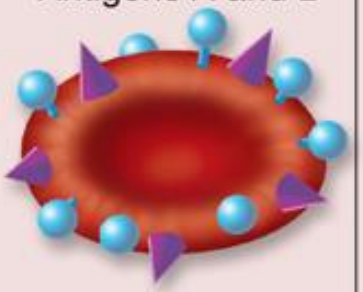
BLOOD TYPES

- 3 alleles of the I gene
  - I^A = A antigen on RBC
  - I^B = B antigen on RBC
  - **i** = neither A nor B antigen



Genotype	Blood type	Antibody
I^AI^A or I^Ai	A	Anti – B
I^BI^B or I^Bi	B	Anti – A
I^AI^B	AB	None
ii	O	Anti – A, Anti – B

ABO Blood Types

Erythrocytes	Antigen A 	Antigen B 	Antigens A and B 	Neither antigen A nor B 
Plasma	Anti-B antibodies 	Anti-A antibodies 	Neither anti-A nor anti-B antibodies	Both anti-A and anti-B antibodies 
Blood type	**Type A** Erythrocytes with type A surface antigens and plasma with anti-B antibodies	**Type B** Erythrocytes with type B surface antigens and plasma with anti-A antibodies	**Type AB** Erythrocytes with both type A and type B surface antigens, and plasma with neither anti-A nor anti-B antibodies	**Type O** Erythrocytes with neither type A nor type B surface antigens, but plasma with both anti-A and anti-B antibodies

BLOOD TYPE	GENOTYPE	ANTIBODY IN PLASMA	CAN RECIVE BLOOD FROM	
A	I^AI^A , I^Ai	**Anti-B**	**A, O**	
B	I^BI^B , I^Bi	**Anti-A**	**B, O**	
AB	I^AI^B	**None**	**A, B, AB, O**	**AB** = universal acceptor
O	ii	**Both Anti A & Anti-B**	**O**	**O** = universal donor

Hair Color is another examples of Multiple Alleles

Hair Color – Too many alleles exist to count

- There are over **20 different** shades of hair color.

Codominance - Situation in which both alleles of a gene contribute to the phenotype of the organism.

– Example – A solid white cow is crossed with a solid brown cow and the resulting offspring are spotted brown and white (called **roan**).

Homozygous white (**WW**)

x

Homozygous red (**RR**)

→

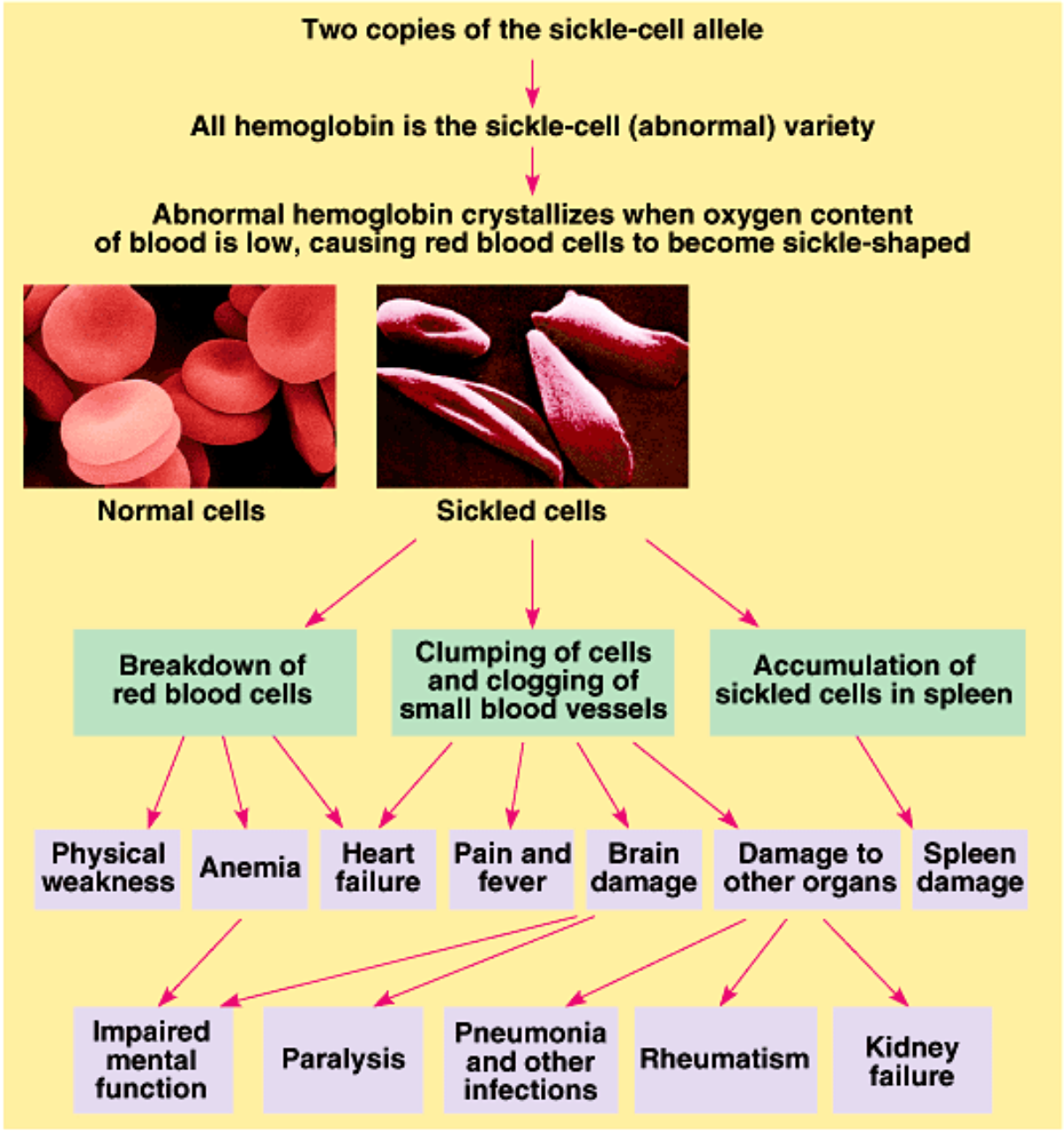
The offspring will have both red and white hairs (**RW**) **[HETEROZYGOUS]**

(c) Pleiotropy

- Most genes have multiple phenotypic effects, a property called **pleiotropy.**

- For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as cystic fibrosis and sickle-cell disease.

Pleiotropic effects of the sickle-cell allele in a homozygote



Pleiotropy:

A single gene may affect phenotype in many ways

Heterozygotes (said to have sickle-cell trait) are usually **healthy** but may suffer some symptoms

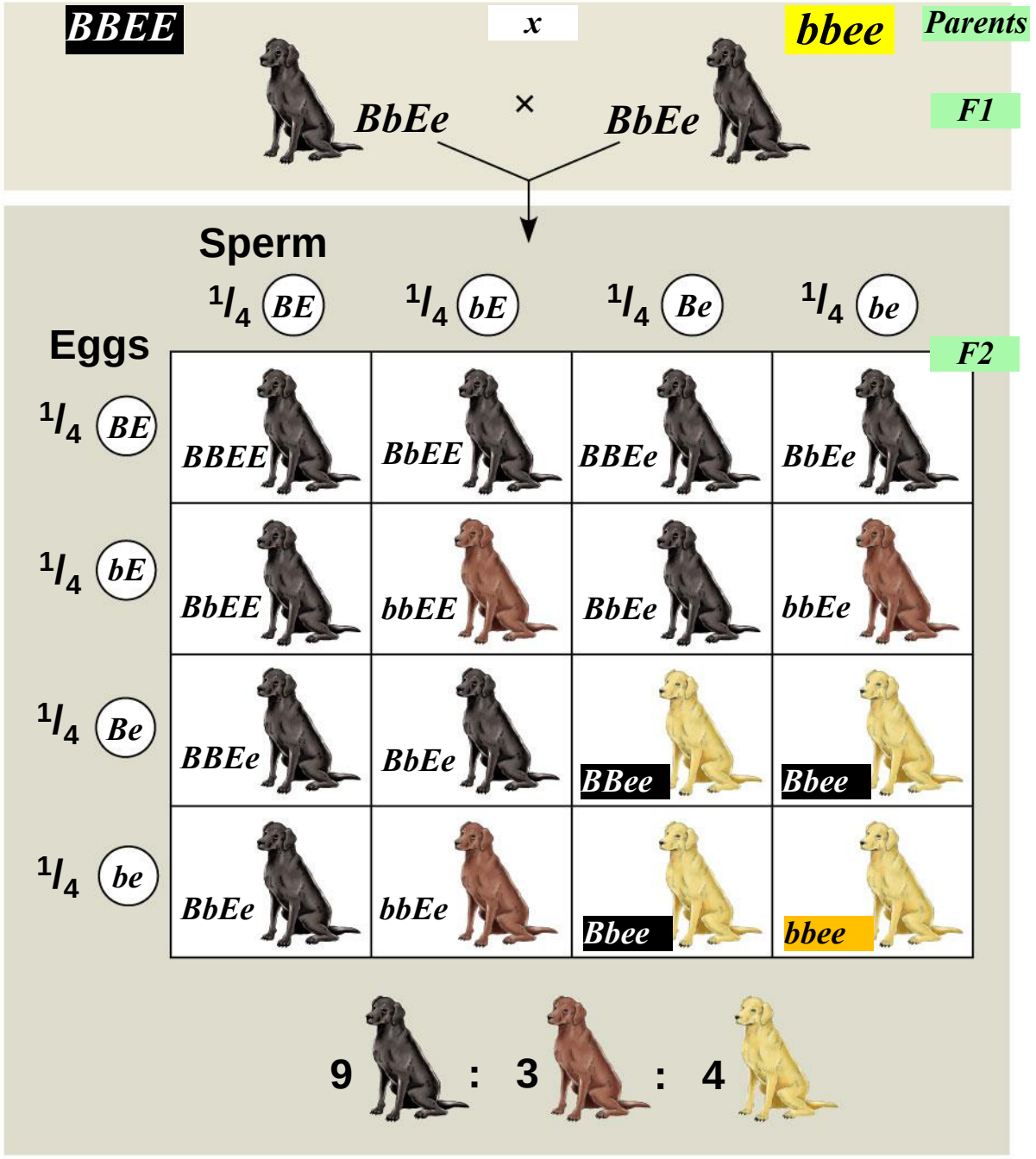
Heterozygotes are **less susceptible to the malaria parasite**, so there is an advantage to being heterozygous – **Selection.**

(d) Epistasis

- In **epistasis**, a gene at one locus **alters** the **phenotypic expression** of a gene at a second locus.
- For example, in Labrador retrievers and many other mammals, coat color depends on two genes.
- One gene determines the **pigment color** (with alleles ***B*** for **black** and ***b*** for **brown**).

- However, a second gene locus controls whether any **eumelanin** at all is **deposited** in the **fur**. Dogs that are homozygous recessive at this locus (***ee***) will have yellow fur no matter which alleles are at the first locus.

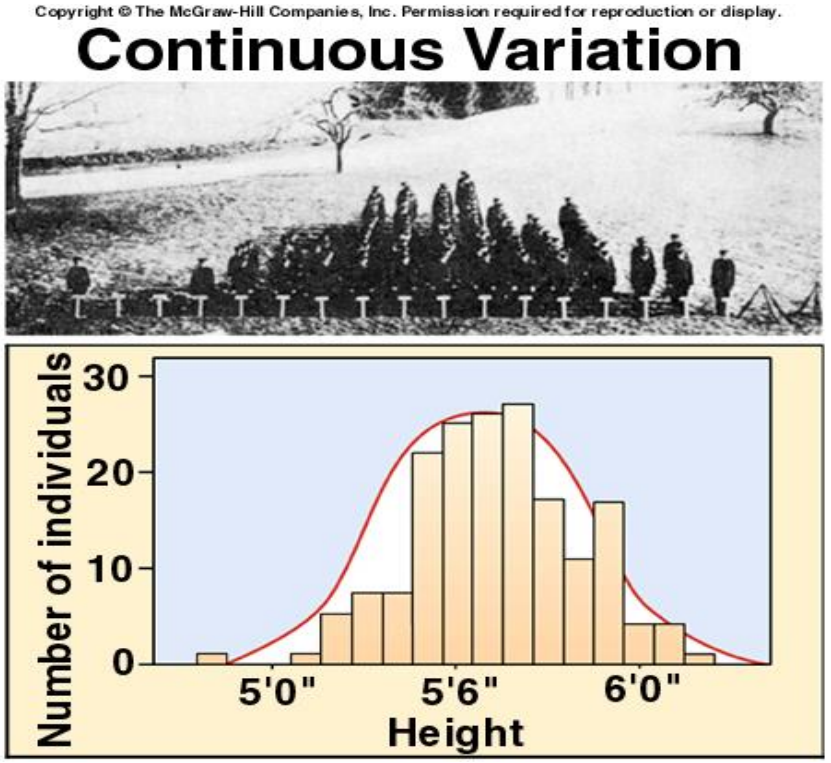
Epistasis



(e) Polygenic Inheritance

- **Quantitative characters** are those that vary in the population along a **continuum.**
- Quantitative variation usually indicates **polygenic inheritance**, an additive effect of two or more genes on a single phenotype.

- Most traits are not controlled by a single gene locus, but by the combined interaction of many gene loci. These are called polygenic traits.
- Polygenic traits often show continuous variation, rather then a few discrete forms:



Polygenic Trait

- **Polygenic Trait** - Trait controlled by two or more *genes*.

  - Polygenic traits often show a wide range of phenotypes.
  - Example: The wide range of skin color in humans comes about partly because more than four different genes probably control this trait.

Polygenic inheritance – 8 genes

Gene	contibution to phenotype	Determinant	1	2	3
A	20	Dominant	HD	HT	HR
B	33	Recessive	HD	HR	HT
C	12	Recessive	HD	HR	HD
D	5	Recessive	HR	HD	HT
E(3 allele)	10	Dominant	HR	HR	HR
F	7	Recessive	HR	HD	HT
G	4	Dominant	HD	HD	HD
H	9	Codominant			
	100%				

(f) Nature and Nurture:

The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character depends on **environment** as well as **genotype**.

- The **norm of reaction** is the phenotypic range of a genotype influenced by the environment. **They are broadest for polygenic characters**.

- For example, **hydrangea** flowers of the same genotype range from **blue-violet** to **pink**, depending on soil acidity

Environmentally-influenced

- Color of the Hydrangea flower determined by the pH of the soil
- **Acidic soil** → **blue** flower

- **Basic soil**→ **pink** flower

Such characters are called **multi-factorial** because **genetic** and **environmental** factors **collectively** influence phenotype

(*g*) Sex-Linked Characteristics are Determined by Genes on the Sex Chromosomes

Z - linked characteristics

X - Linked Characteristics

Y - linked characteristics

- Hairy ears

Insensitivity to certain colors

Light-sensitive opsin **proteins** made in the eye & needed for color vision are **encoded by** a cluster of **genes on the X** chromosome.

Mutations in these genes can lead to an insensitivity to certain colors (like **red** and **green**) when seen together ("**color vision deficiency**")

Sex-influenced Traits

- Aka, “Gender-influenced”
  - Usually influenced by **sex hormones** like estrogen, testosterone
  - Examples include **baldness** in humans, **plumage** in birds, **horns** on cattle

Z-linked characteristics

Indian blue Peacock is inherited as a Z-linked dominant trait.

ZW - blue Peacock

The cameo phenotype (plumage) **in** Results from a **Z-linked** allele that is **recessive** to the wild-type blue allele

ZZ - cameo

Y-linked characteristics (Hairy ears) -

- **Showing** variable expressivity and incomplete penetrance
- Could also be **autosomal dominant** characteristic expressed only in males

Penetrance: Refers to the proportion of people with a particular **genetic** change (such as a mutation in a specific **gene**) who exhibit signs and symptoms of a **genetic** disorder. If some people with the mutation do not develop features of the disorder, the condition is said to have reduced (or incomplete) **penetrance**

(Eg. 80% penetrance)

Expressivity: is the degree to which trait expression differs among individuals.

Unlike **penetrance**, **expressivity** describes individual variability, not statistical variability among a population of genotypes

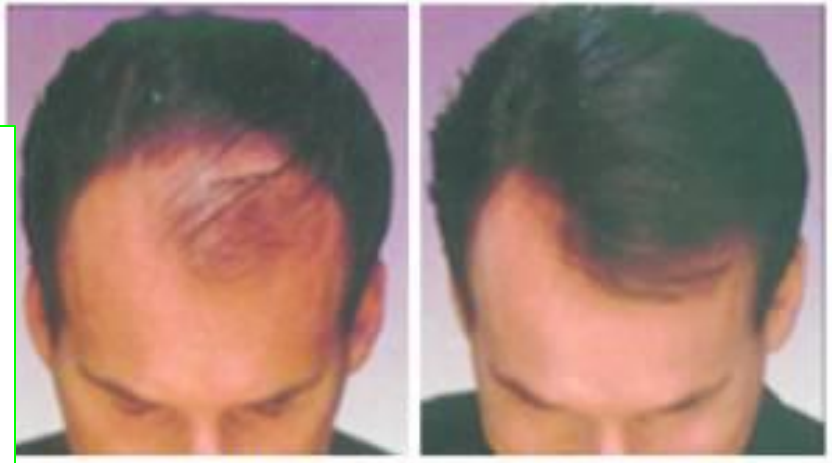
Sex influenced inheritance

- Inheritance can be affected by the sex of an individual, although the specific gene may not be carried on **X** chromosome.

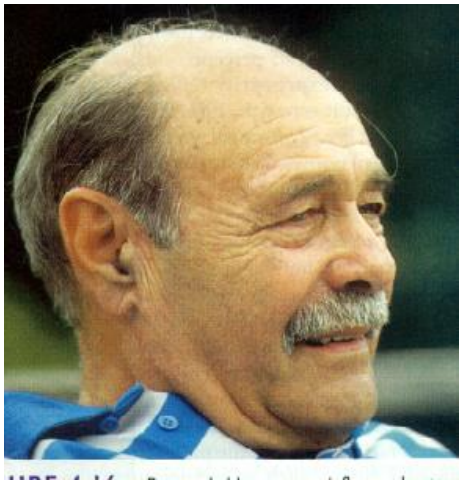
- Eg., the **feather phenotype** in chicken is controlled by a pair of alleles on **autosomes** but the expression of the alleles **is modified by sex Hormones**.

Sex-Influenced Traits

- Expressed in **males** and **females**
- Usually controlled by autosomal genes
- Generally phenotypic variations are due to hormonal differences between the sexes
- Some traits appear to be specific to one sex, but are not sex-linked: their genes are not on the **X** chromosome.
- Such a trait is called sex-influenced. More specifically, a trait that is dominant in one sex but recessive in the other is a sex-influenced trait.
- The best human example is male pattern baldness.
- Baldness is dominant in males: heterozygotes and homozygotes both become bald. In females, only homozygotes (which are relatively rare) become bald. Also, females tend to lose hair more evenly than men, giving a sparse hair pattern rather than completely baldness.



	BB	Bb	bb
male	bald	bald	hair
female	bald	hair	hair



(h) Genomic Imprinting *(example 1)*

- For a few mammalian traits, the phenotype depends on which parent passed along the alleles for those traits.
- Such variation in phenotype is called **genomic imprinting**.

Imprinting: The differential expression of the two alleles of a gene based on their parental origin, requires that the alleles be distinguished or marked. A candidate for the differentiating mark is DNA methylation.

- Genomic imprinting involves the **silencing** of certain genes that are "**stamped**" with an imprint during gamete production.

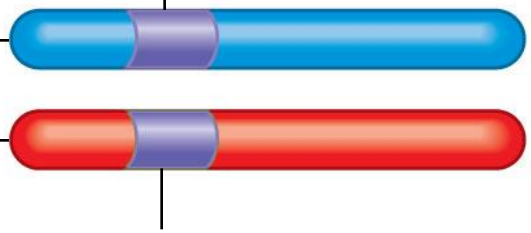
(a) Homozygote for *Igf2* allele

IGF2 is part of a cluster of genes on the short (***p***) arm of chromosome **11**, determines **size** of the mouse (*normal* / *dwarf*)

Normal *Igf2* allele is expressed.

Paternal chromosome

Maternal chromosome



Normal *Igf2* allele is not expressed.

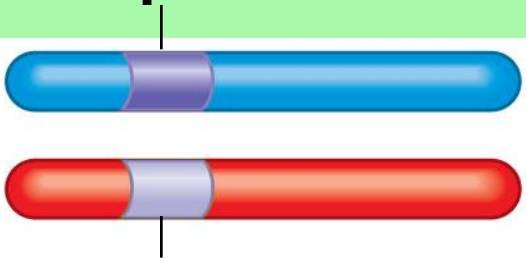
Normal-sized mouse (wild type)

Maternal allele is **silenced (imprinted)**

Mutant *Igf2* allele inherited from mother

Normal-sized mouse (wild type)

Normal *Igf2* allele is expressed.

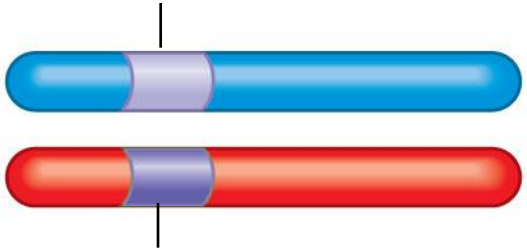


Mutant *Igf2* allele is NOT expressed. (imprinted)

Mutant *Igf2* allele inherited from father

Dwarf mouse (mutant)

Mutant *Igf2* allele is expressed.



Normal *Igf2* allele is Not expressed. (imprinted)

(b) Heterozygote for *Igf2* allele

- It appears that imprinting is the result of the **methylation** - (addition of - **CH_3**) of **cytosine** nucleotides.

- Genomic imprinting **affect only a small fraction** of mammalian genes.

- Most imprinted genes are critical for embryonic development.

Genomic Imprinting *(example 2)*

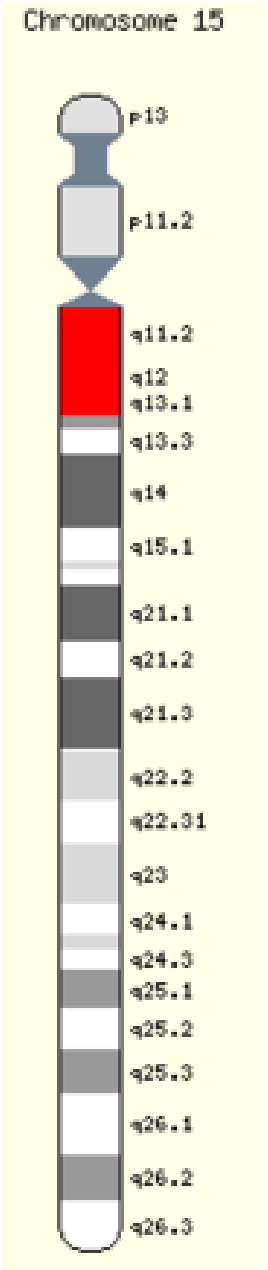
- It is apparent that the parental origin of genetic material does have an impact on gene expression and this effect has become known as **genomic imprinting**.

- Imprinted genes are either expressed *only* from the allele inherited from the ***mother*** or in other instances from the allele inherited from the ***father***.

Genomic Imprint of Chromosome #15

- Imprinted genes are either expressed *only* from the allele inherited from the ***mother*** or in other instances from the allele inherited from the ***father***.

  - Genetic deletion on the q arm of Chromosome #15 depends on whose chromosome you got it from.
  - If you got the deletion from Mom→ Angelman Syndrome
  - If you got it from Dad→ Prader-Willi Syndrome.



Angelman Syndrome

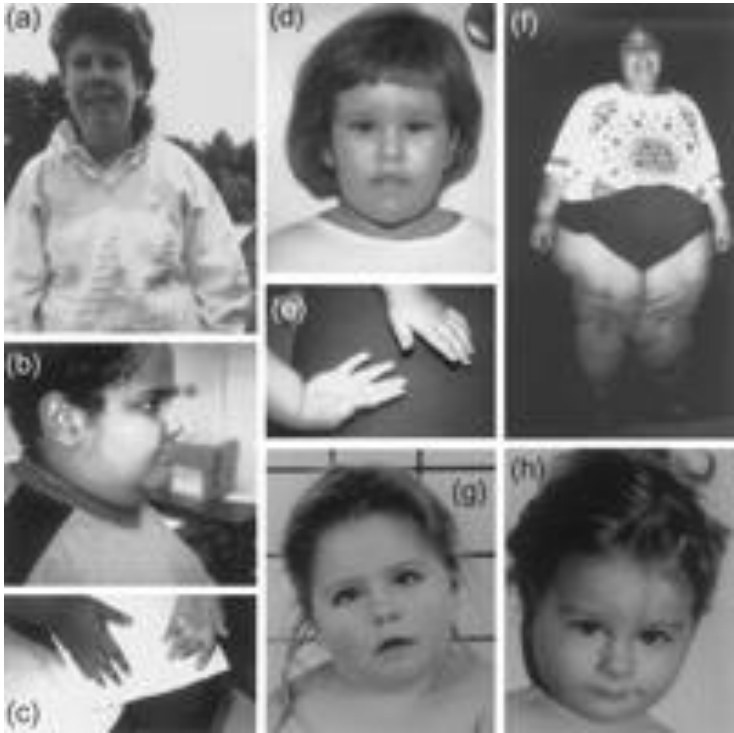
(Father imprinted, mum deleted gene expressed):

Absence of speech, mild to moderate mental retardation, small hands/feet, laugh a lot, dancing gait so called "Happy Puppet Syndrome."

Prader-WilliSyndrome

(Mother imprinted, dad deleted gene expressed):

Severe obesity, hyperactivity & severe mental retardation



(i) Multifactorial Disorders

- Many diseases, such as heart disease, diabetes, alcoholism, mental illnesses, and cancer have both **genetic** and **environmental** components.

- Little is understood about the genetic contribution to most multifactorial diseases

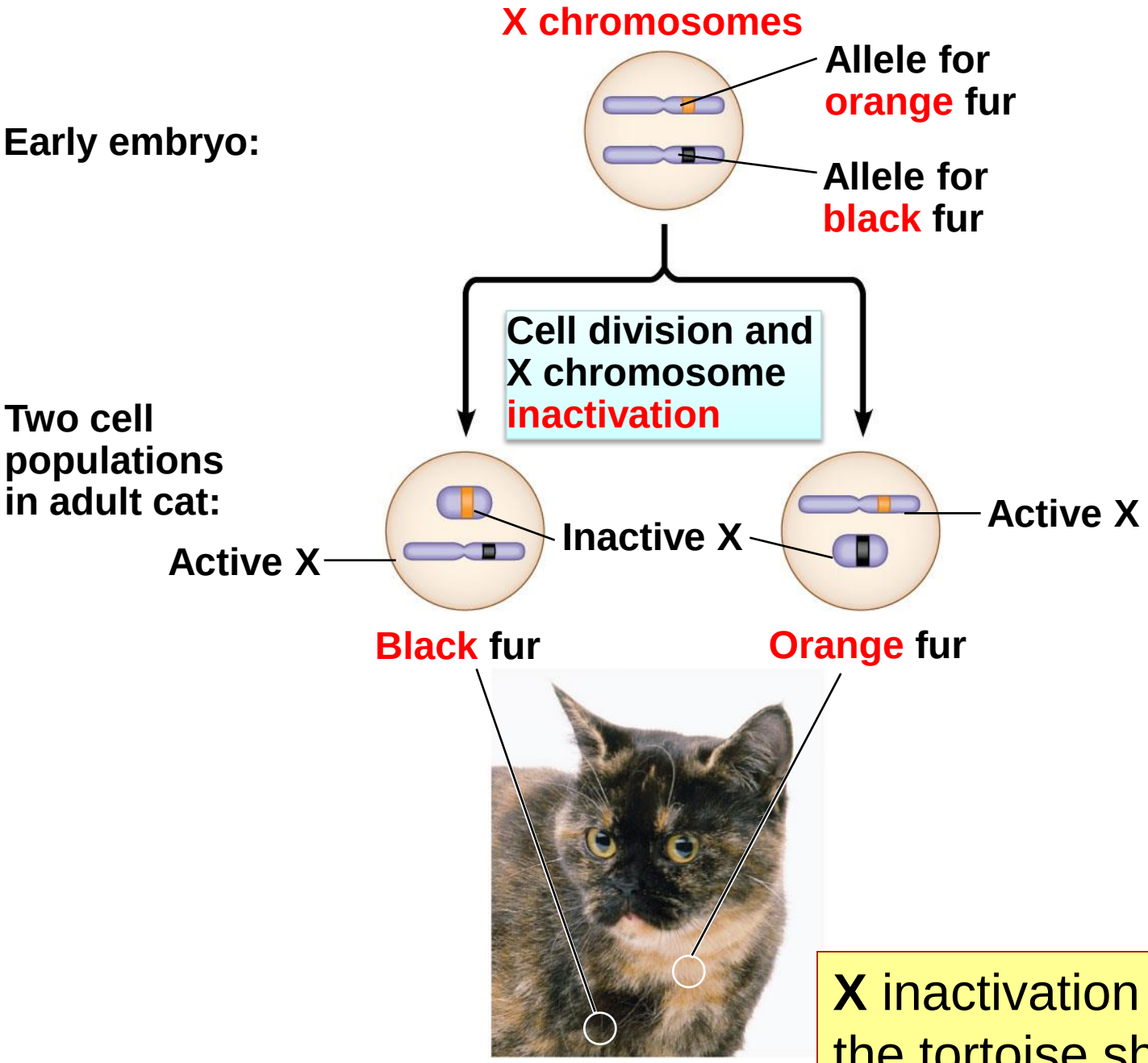
X Inactivation in Female Mammals & Mosaicism

- In mammalian females, **one** of the two **X** chromosomes in each cell is randomly inactivated during embryonic development.

  =**NOT PARMANENTLY**=

- The inactive **X** condenses into a **Barr body.**

If a **female** is **heterozygous** for a particular gene located on the **X chromosome**, she will be a **mosaic** for that character.

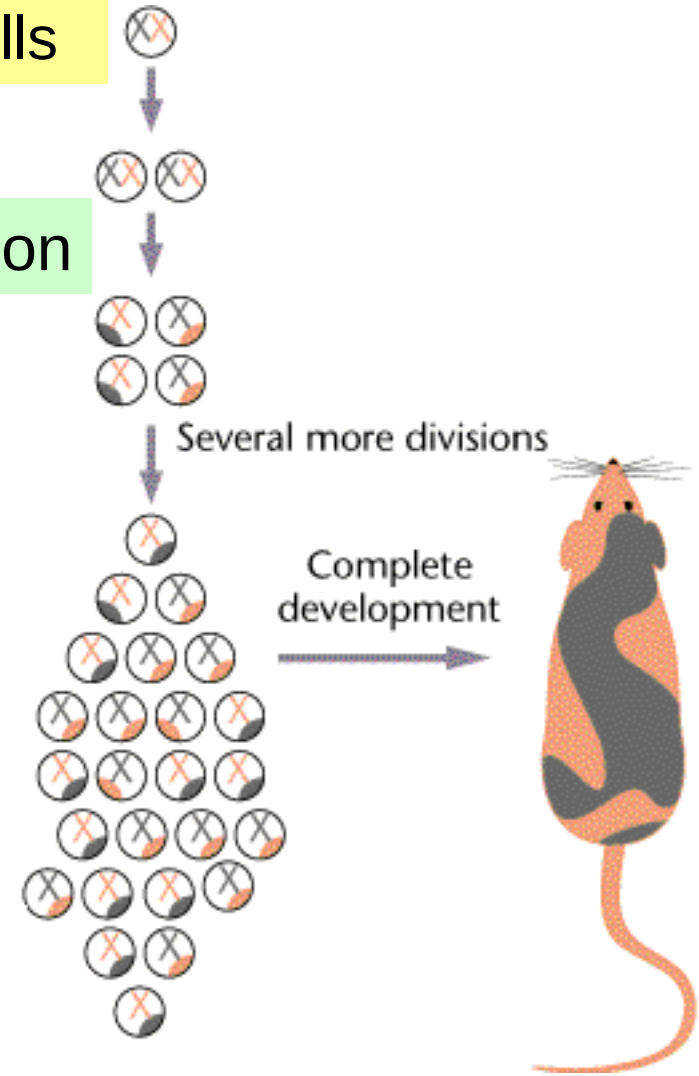


X inactivation and the tortoise shell cat

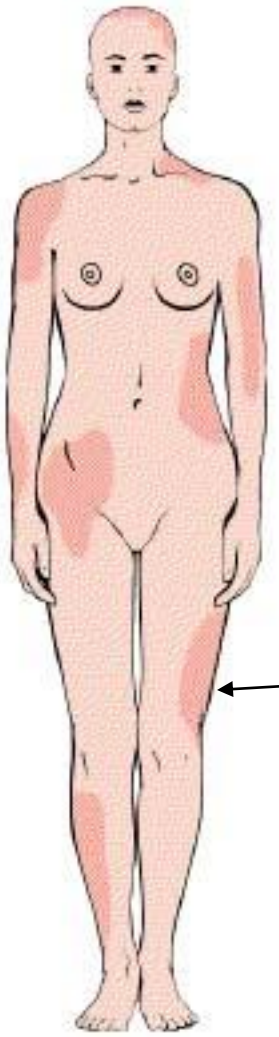
Lyon-Hypothesis: X-inactivation

ursor cell to all coat color cells

Random inactivation



Mosaicism Reveals the Random Inactivation of one X chromosome



Regions where sweat glands are absent.

Many human traits follow Mendelian patterns of inheritance

- Humans are not good subjects for genetic research
  - Generation time is too long
  - Parents produce relatively few offspring
  - Breeding experiments are unacceptable – **UNETHICAL**

- However, basic Mendelian genetics **endures** as the foundation of human genetics

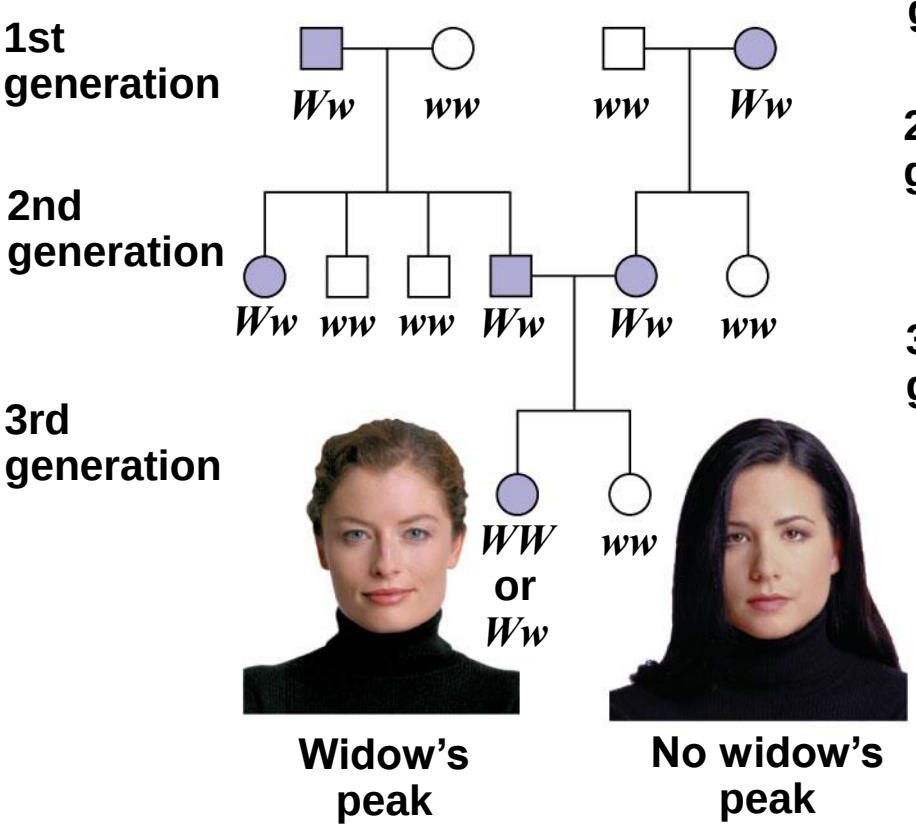
Pedigree Analysis

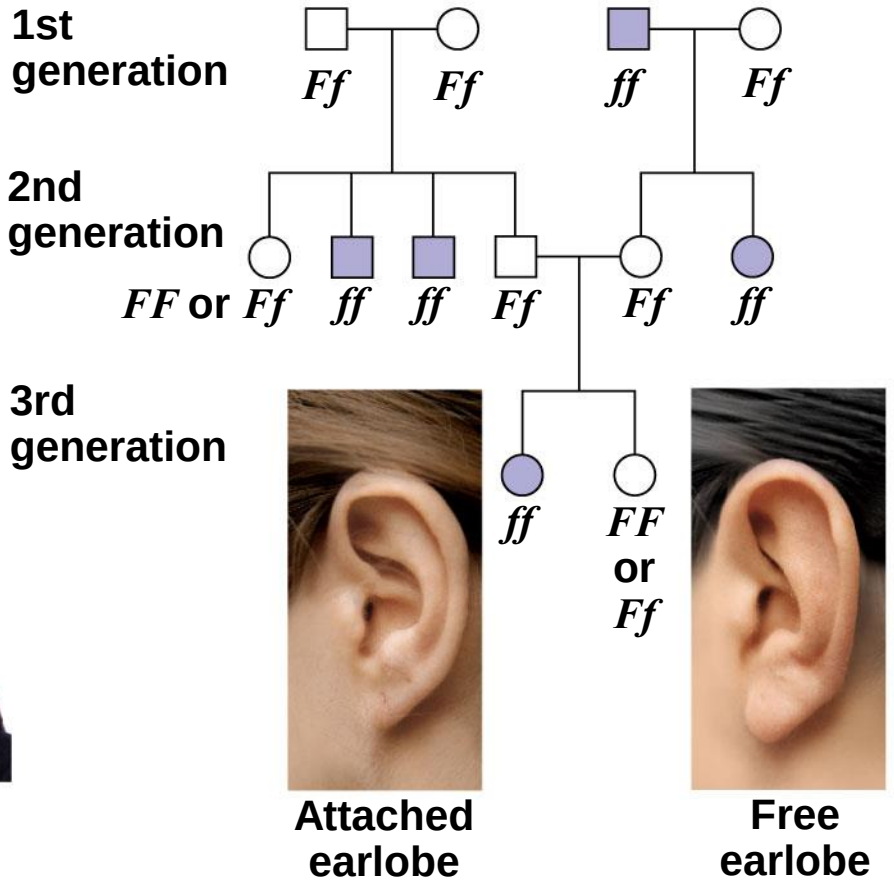
- A **pedigree** is a family tree that describes the interrelationships of parents and children across generations.
- Inheritance **patterns** of particular traits can be **traced** and described using pedigrees.
- Pedigrees can also be used to make **predictions** about future offspring.
- We can use the multiplication and addition rules to predict the **probability** of specific phenotypes

Pedigree Analysis

Key

Male | Female | Affected male | Affected female | Mating | Offspring





(a) Is a widow's peak a dominant or recessive trait?

b) Is an attached earlobe a dominant or recessive trait?

The Behavior of Recessive Alleles

- Many genetic disorders are inherited in a recessive manner, these range from relatively mild to life-threatening.
- Recessively inherited disorders show up only in individuals homozygous for the allele.

- **Carriers** are heterozygous individuals who carry the recessive allele but are phenotypically normal; most individuals with recessive disorders are born to carrier parents.

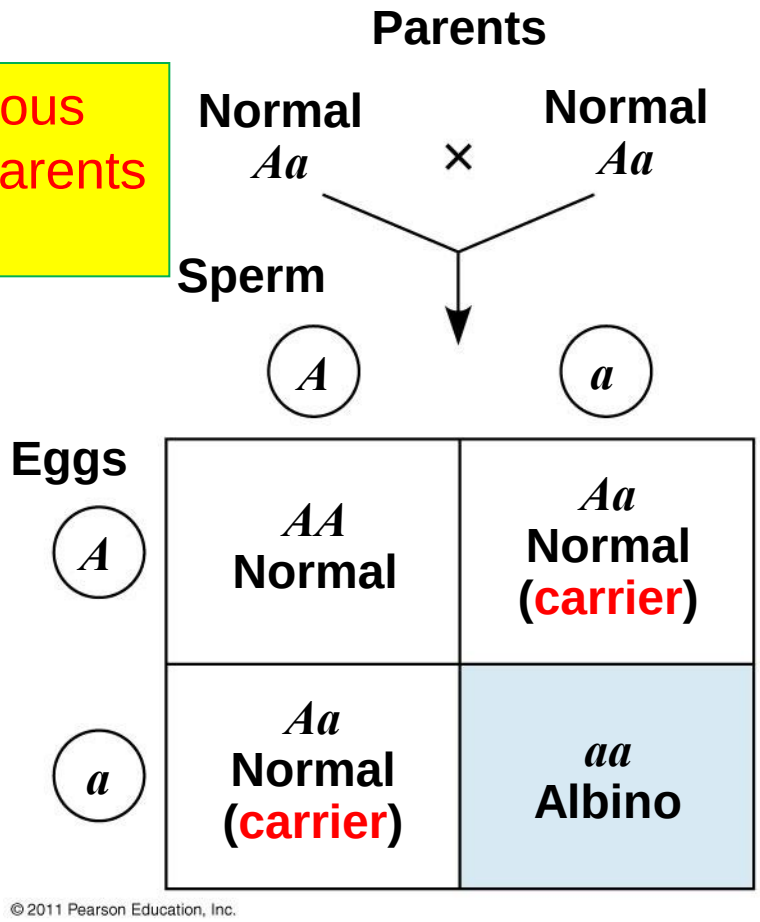
- Albinism is a recessive condition characterized by a lack of pigmentation in skin and hair

Albinism

Heterozygous (normal) parents **(carriers)**

Gametes

Offspring



- If a recessive allele that causes a disease is rare, then the **chance** of two carriers meeting and mating is **low**.

- **Consanguineous** mating (i.e., mating between close relatives) **increase** the chance of mating between **two carriers** of the same rare allele.

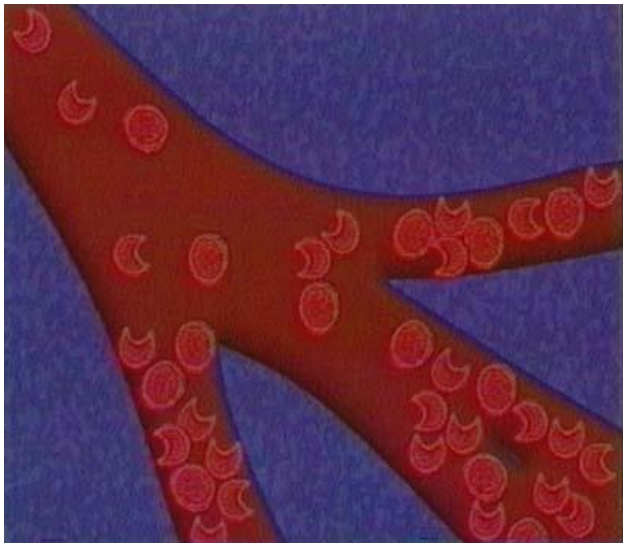
- Most societies and **cultures** have **laws** or **taboos** against marriages between close relatives.

Sickle-Cell Disease

- **Sickle-cell disease** affects one out of 400 African-Americans
- The disease is caused by the **substitution** of a single amino acid in the β-hemoglobin protein in red blood cells (**Glu** *with* **Val**)
- In **homozygous** individuals, **all** hemoglobin is abnormal (sickle-cell).
- Symptoms include physical weakness, pain, organ damage, and even paralysis.
- **Two alleles**
  1) Hb^A
  Encodes normal beta hemoglobin chain
  2) Hb^S
  Mutant allele encodes defective chain
- **Hb^S homozygotes produce only the defective hemoglobin; suffer from sickle-cell anemia**

- **Heterozygotes** (said to have sickle-cell trait) are usually **healthy** but may suffer some symptoms.

- Heterozygotes are **less susceptible to the malaria parasite**, so there is an advantage to being heterozygous – **Selection.**



Hemophilia

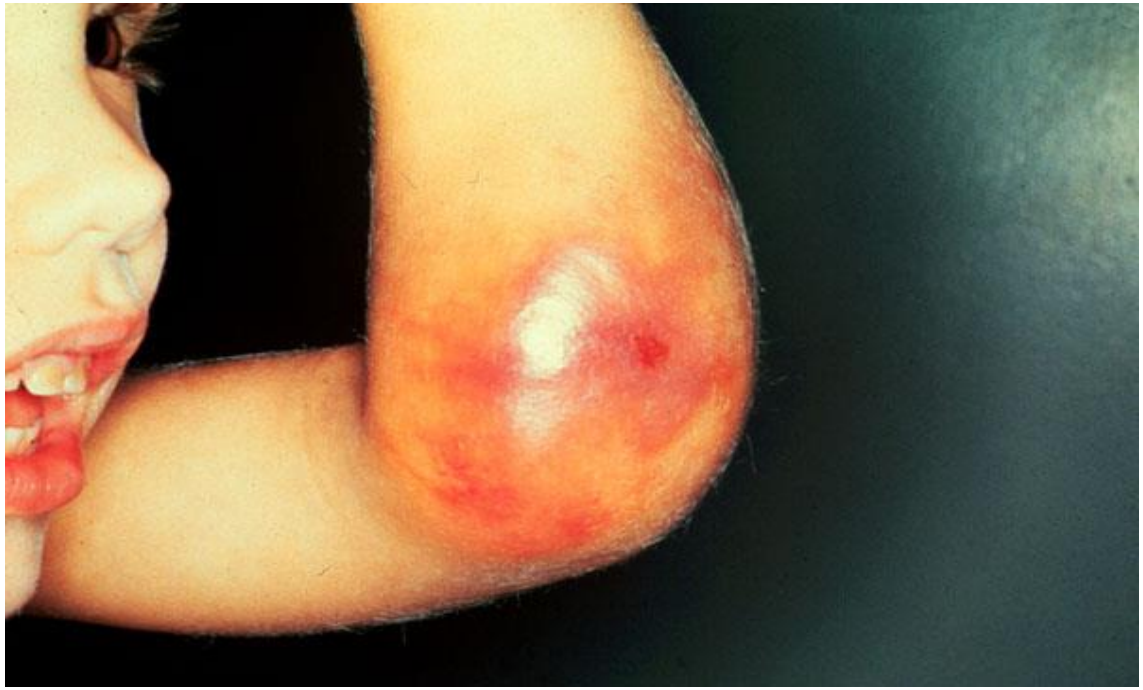
A blood disorder where the **blood does not clot properly**.

A **minor cut can cause serious injury** and demand medical attention.

Bleeding into the joints, internal bleeding and deep cuts can be fatal for hemophiliacs.

Genetic lack of one of the clotting factors produced by the liver.

There is **no cure** for hemophilia but treatment options with clotting factor transfusions are available.



Complications from hemophilia include:

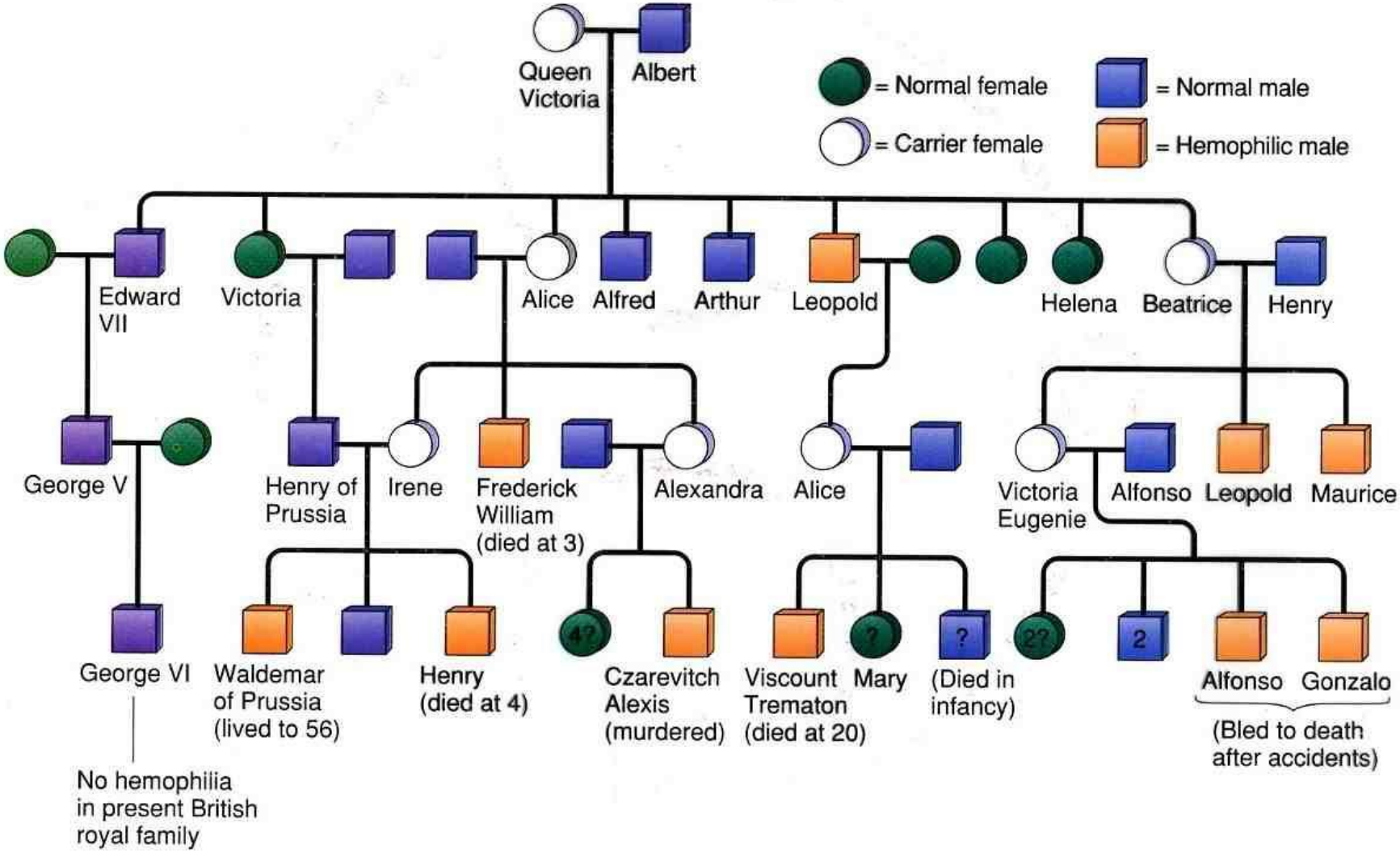
- bruising and bleeding into the muscles,
- bleeding into the joints, infection,
- adverse reaction to transfusions and serious bleeding.

Genetics of Hemophilia

- The gene for hemophilia is found on the **X chromosome**
- It is a **recessive** disorder.
- It is referred to as a sex-linked recessive disorder.
- **Males are more likely to get hemophilia.**
- Females have the possibility of being heterozygous for hemophilia (**Carrier**)

PEDIGREE OF QUEEN VICTORIA

Hemophilia is a <u>sex-linked</u> <u>recessive trait</u> defined by the absence of one or more of the proteins required for blood clotting.



Lactose intolerance in humans

Lactose=========>Glucose + Galactose

Lactase

- Human milk is **7%** lactose.
- Lactose is not absorbed through the wall of the digestive tract.
- In human infants, lactase is secreted in **intestine** which breaks the lactose into easily **absorbed** **Glucose** and **Galactose**.
- Production of the **lactase** enzyme **declines** in adults.
- The unabsorbed lactose creates cramps, diarrhea, and nausea.
- In some humans, lactase continues to be produced throughout adulthood.
- These individuals are called lactose absorbers.

Lactose intolerance in humans *cont*.

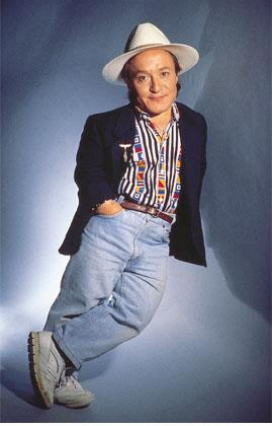
- Adult lactose absorption is inherited as an **autosomal dominant** trait.
- Lactose persistence and non-persistence reflect inheritence of different alleles of the lactase gene.
- Lactose intolerance is the result of being homozygous for the **recessive** lactase (WT) allele
- Being **homozygous** or **heterozygous** for the **mutant** allele allows lactase expression in adults when normally lactase expression is turned off.

Dominantly Inherited Disorders

- Some human disorders are caused by dominant alleles.

- Dominant alleles that cause a lethal disease are rare and arise by mutation.

- ***Achondroplasia*** is a form of **dwarfism** caused by a rare dominant allele, **is lethal when homozygous** for the **dominant** allele.



Achondroplasia

Parents

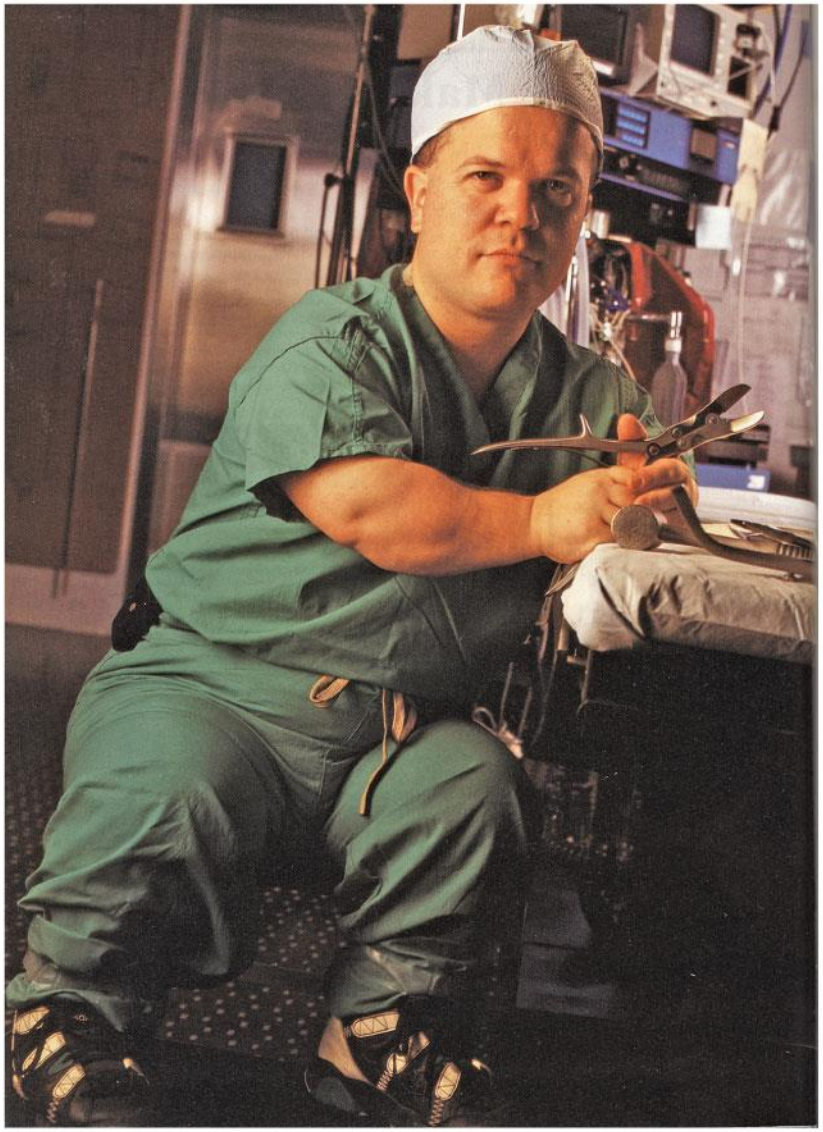
Dwarf *Dd* × Normal *dd*

Sperm

D *d*

Eggs

d	*Dd* Dwarf	*dd* Normal
d	*Dd* Dwarf	*dd* Normal



Huntington's Disease: A Late-Onset Lethal Disease

- The timing of onset of a disease significantly affects its inheritance.

- **Huntington's disease** is a degenerative disease of the nervous system [is due to $(CAG)_n$ repeats in the Huntingtin gene, beyond $n = 35$].

- The disease has no obvious phenotypic effects until the individual is about 35 to 40 years of age.

- Once the deterioration of the nervous system begins the condition is irreversible and fatal

Genetic Testing and Counseling

- *Counseling Based on Mendelian Genetics and Probability Rules.*

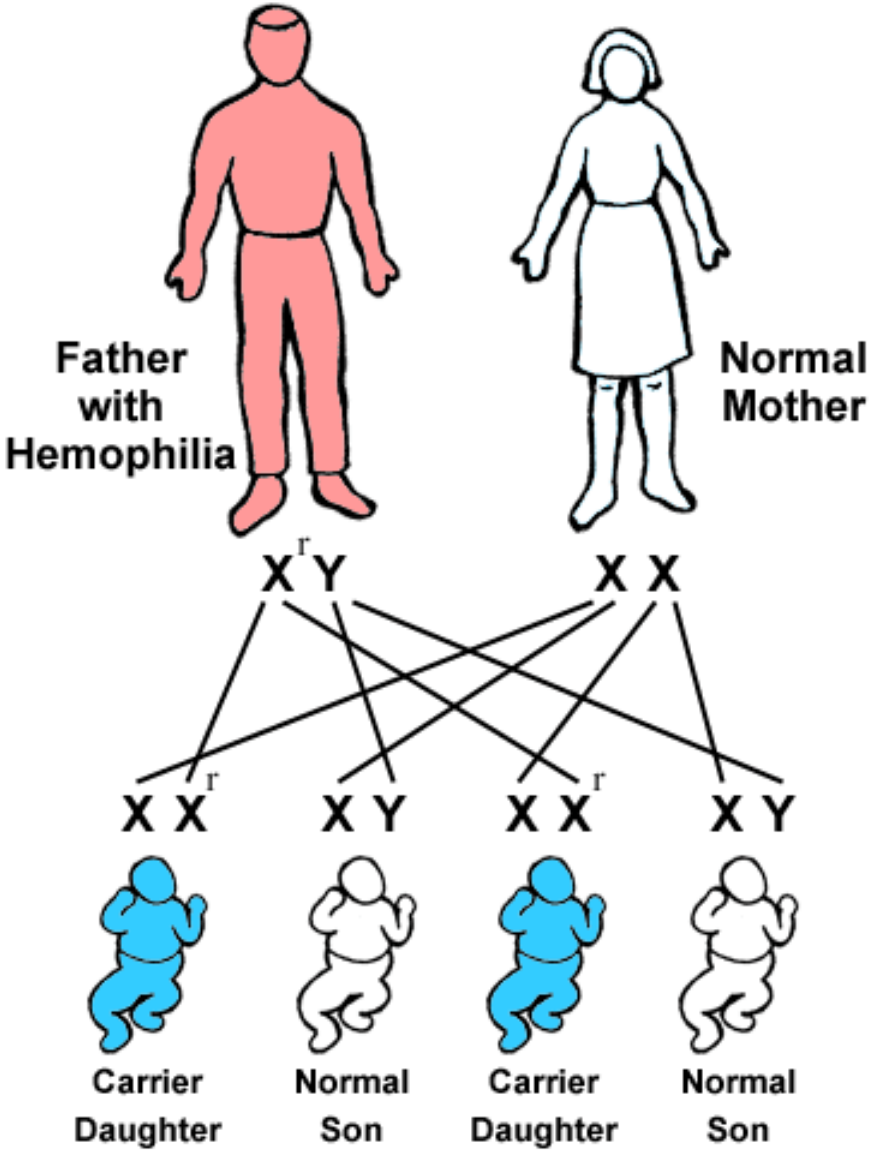
- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease.

- Using family histories, genetic counselors help couples determine the odds that their children will have genetic disorders.
- **For a growing number of diseases, tests are available that identify carriers and help define the odds more accurately**

Fetal Testing

- In **amniocentesis**, the liquid that bathes the fetus is removed and tested.
- In **chorionic villus sampling (CVS**), a sample of the **placenta** is removed and tested.
- Other techniques, such as *ultrasound* and ***fetoscopy***, allow fetal health to be assessed visually in utero.

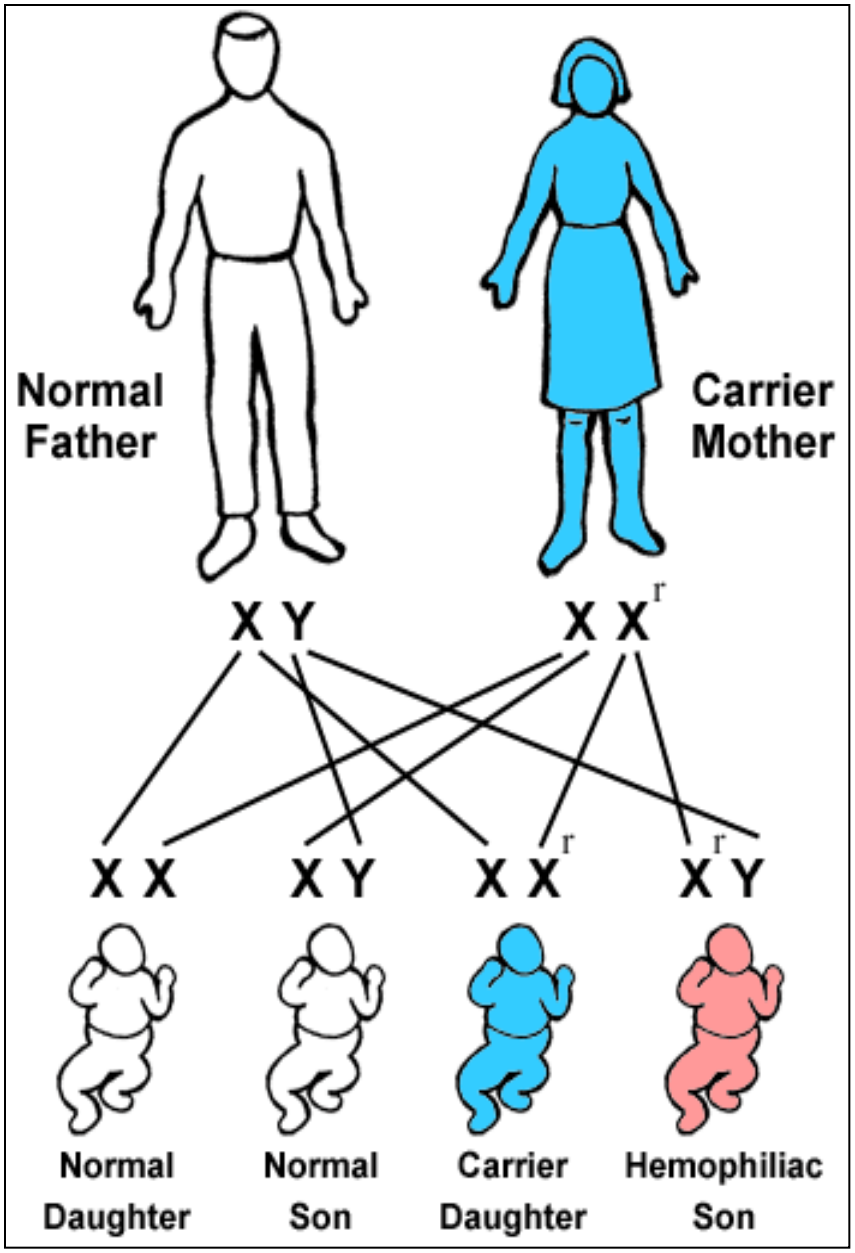
- ***Newborn Screening:*** Some genetic disorders can be detected at birth by simple tests that are now routinely performed in most hospitals in the United States



In this example:

The father has hemophilia. He cannot give his son hemophilia because he gives his son the **Y** chromosome.

He can give his daughter the recessive gene, but if her mother does not give her the recessive gene, she will not have hemophilia. She will be a **carrier**.



In this example:

The mother is a carrier of hemophilia.

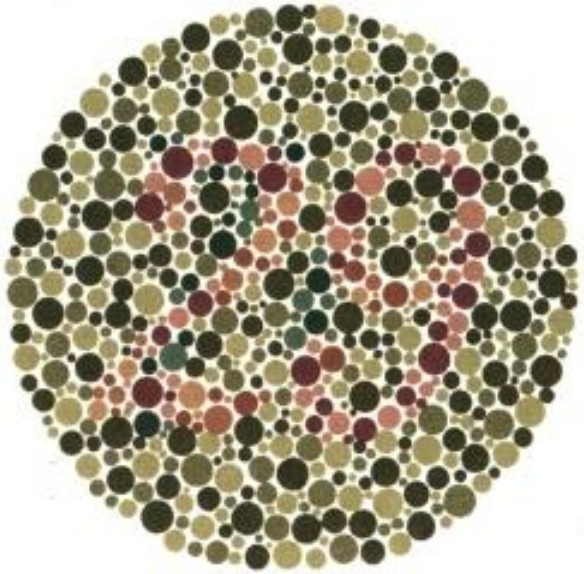
She does not have hemophilia but she is heterozygous for the trait = **Carrier**.

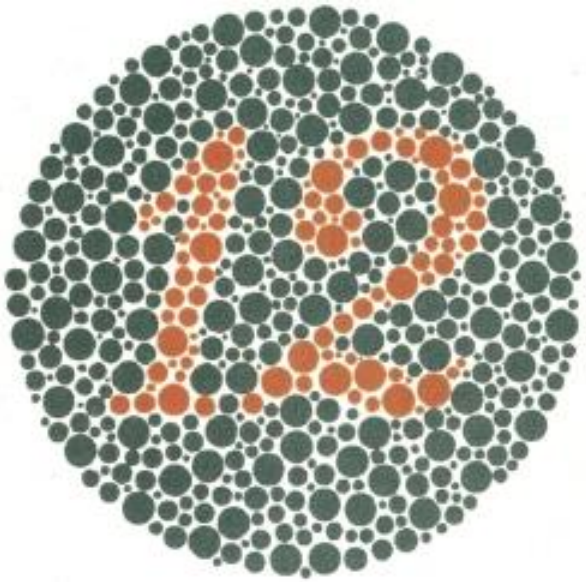
There is a **50%** chance her son will have hemophilia.

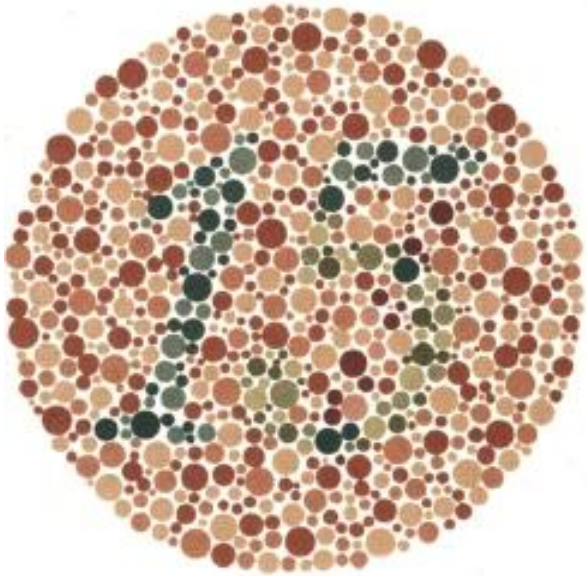
Color Blindness

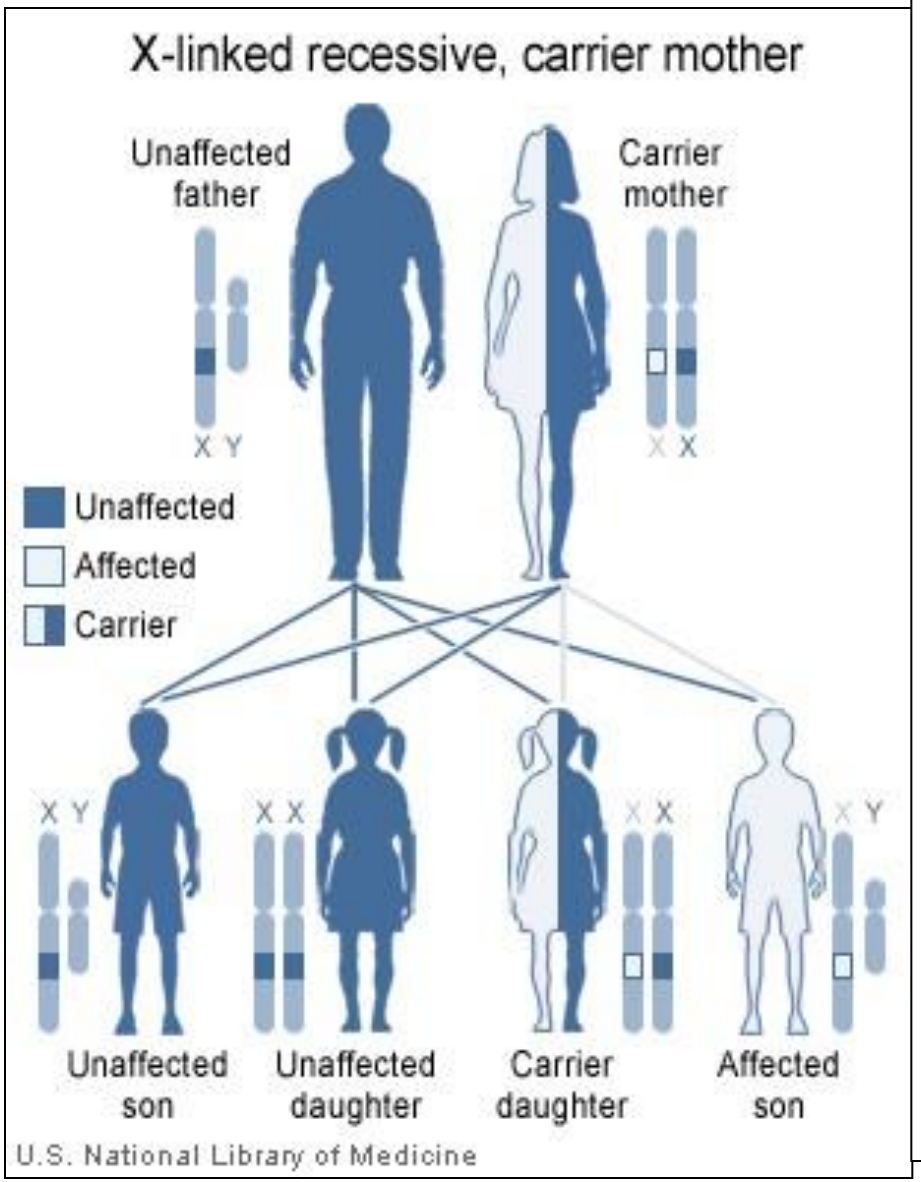
- Color Blindness is a sex-linked trait found on the **X** chromosome.

- **Males** are more likely to be color blind due to the fact they only have one **X** chromosome.









In this example:

The mother is a carrier of the colorblind gene.

There is a **50%** chance her son will be colorblind but unless the father is colorblind the daughter cannot end up colorblind.

Pedigree Chart: Inheritance Pattern for an X-linked Recessive Disease

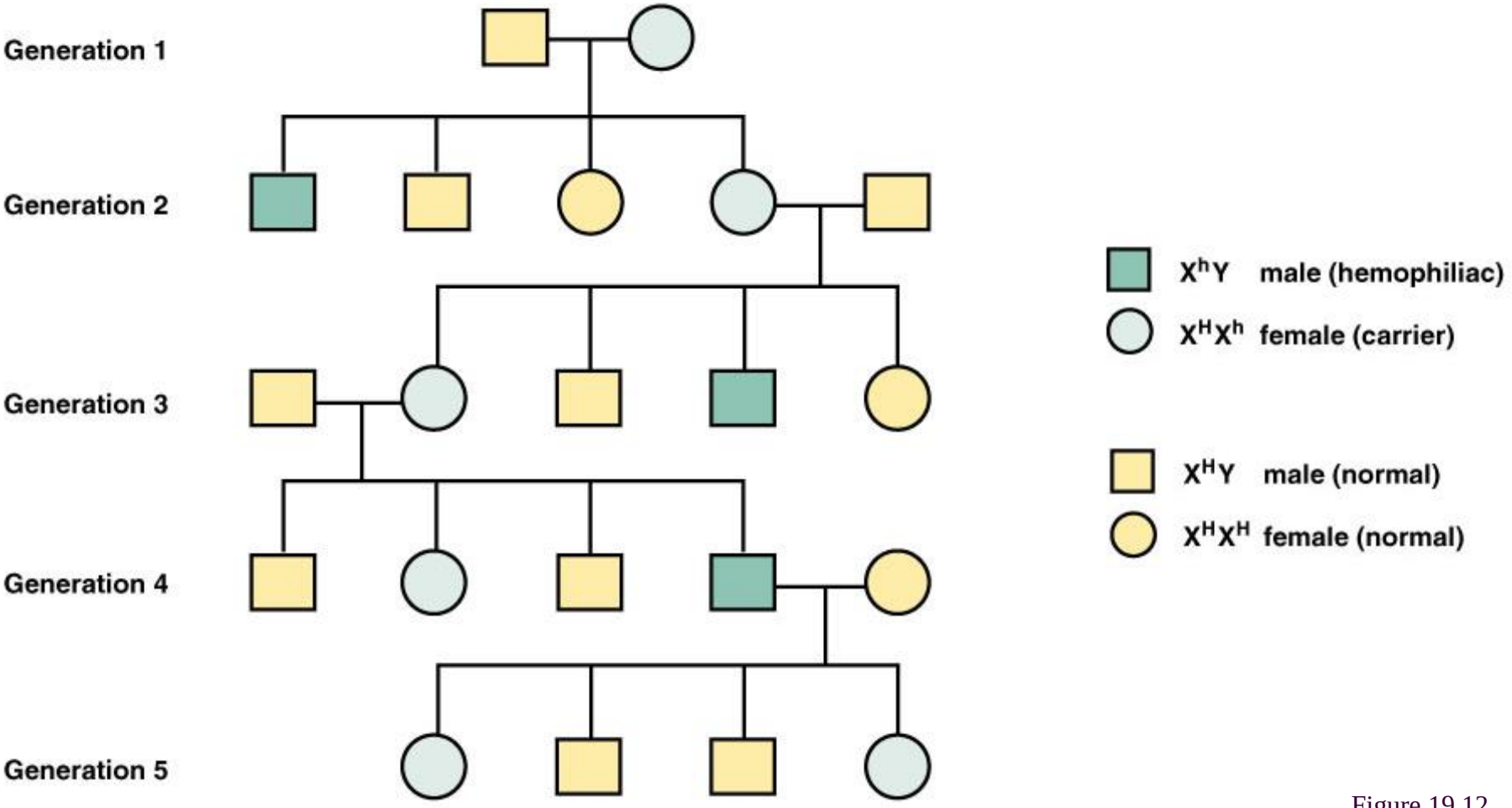


Figure 19.12