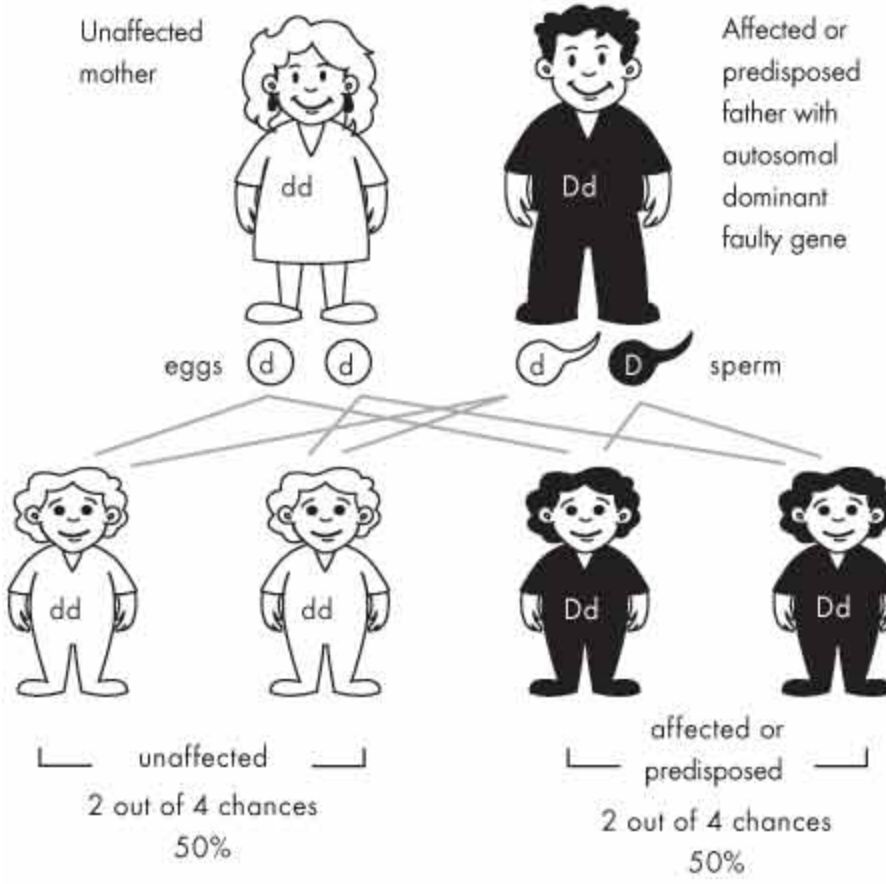
GENETICS
Chapter 7: Extending Mendelian Genetics

I. Chromosomes and Phenotype (7.1)



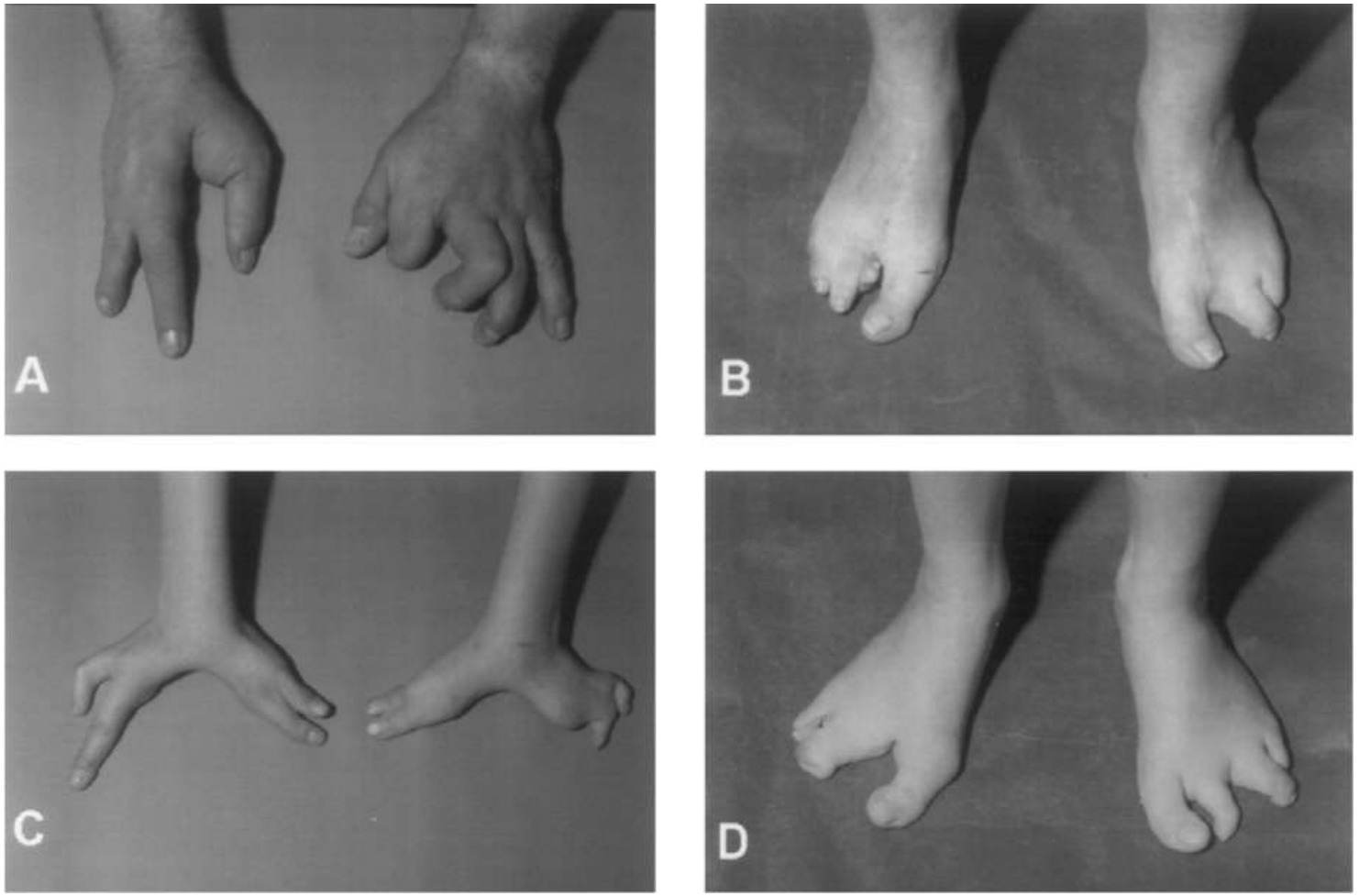
A. <u>Two</u> copies of each autosomal gene affect phenotype

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

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

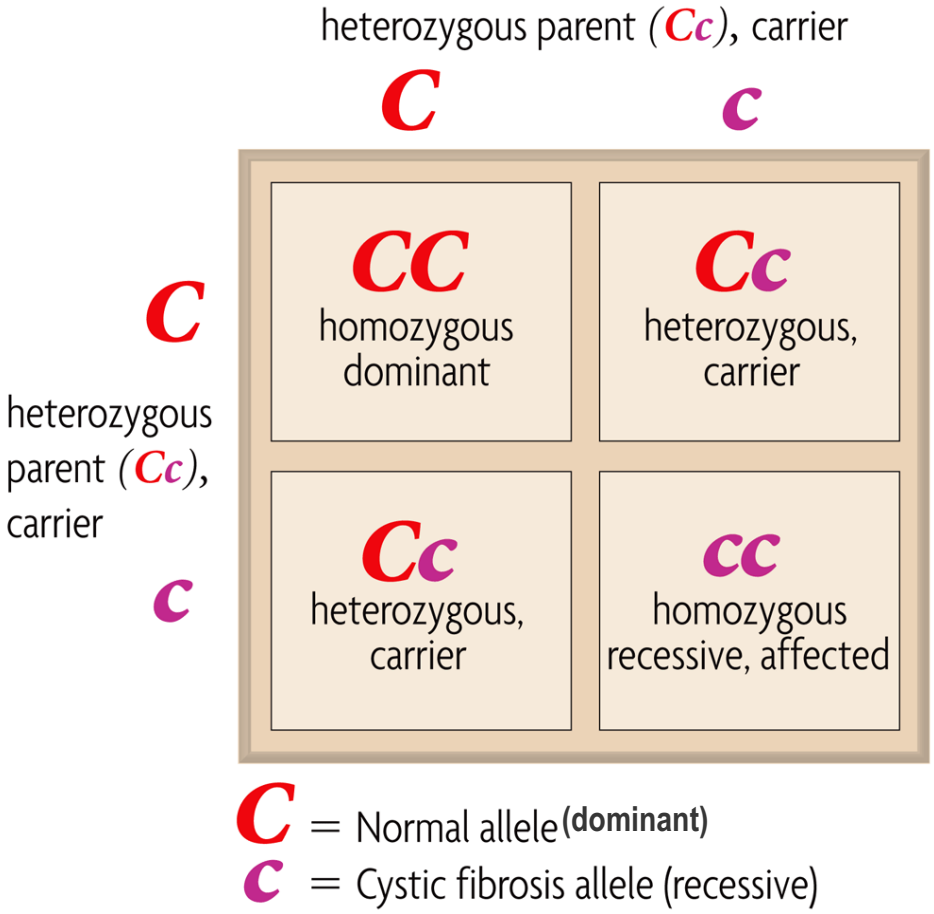
a. Chance of having disorder can be predicted

b. Use same principles as Mendel did



B. Disorders Caused by Recessive Alleles

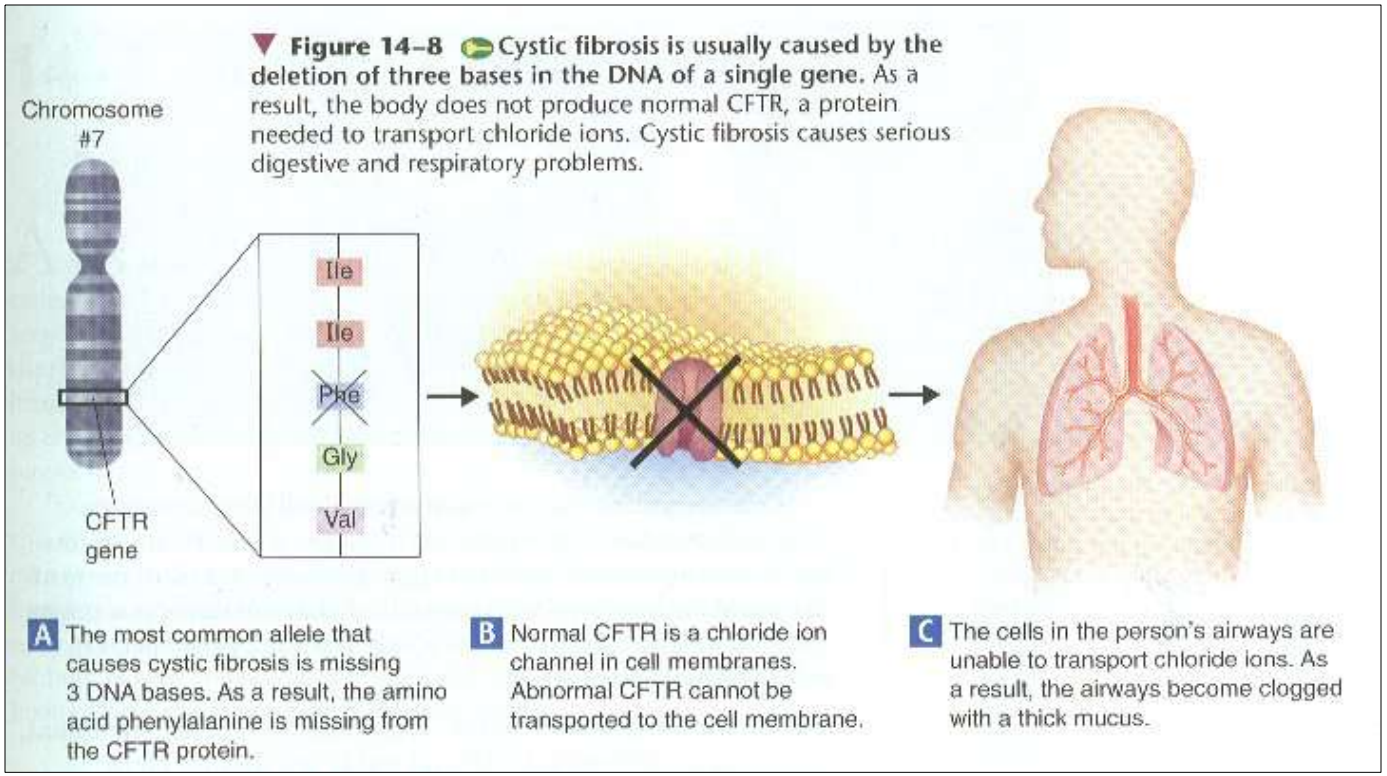
1. Some disorders are caused by recessive alleles on autosomes



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

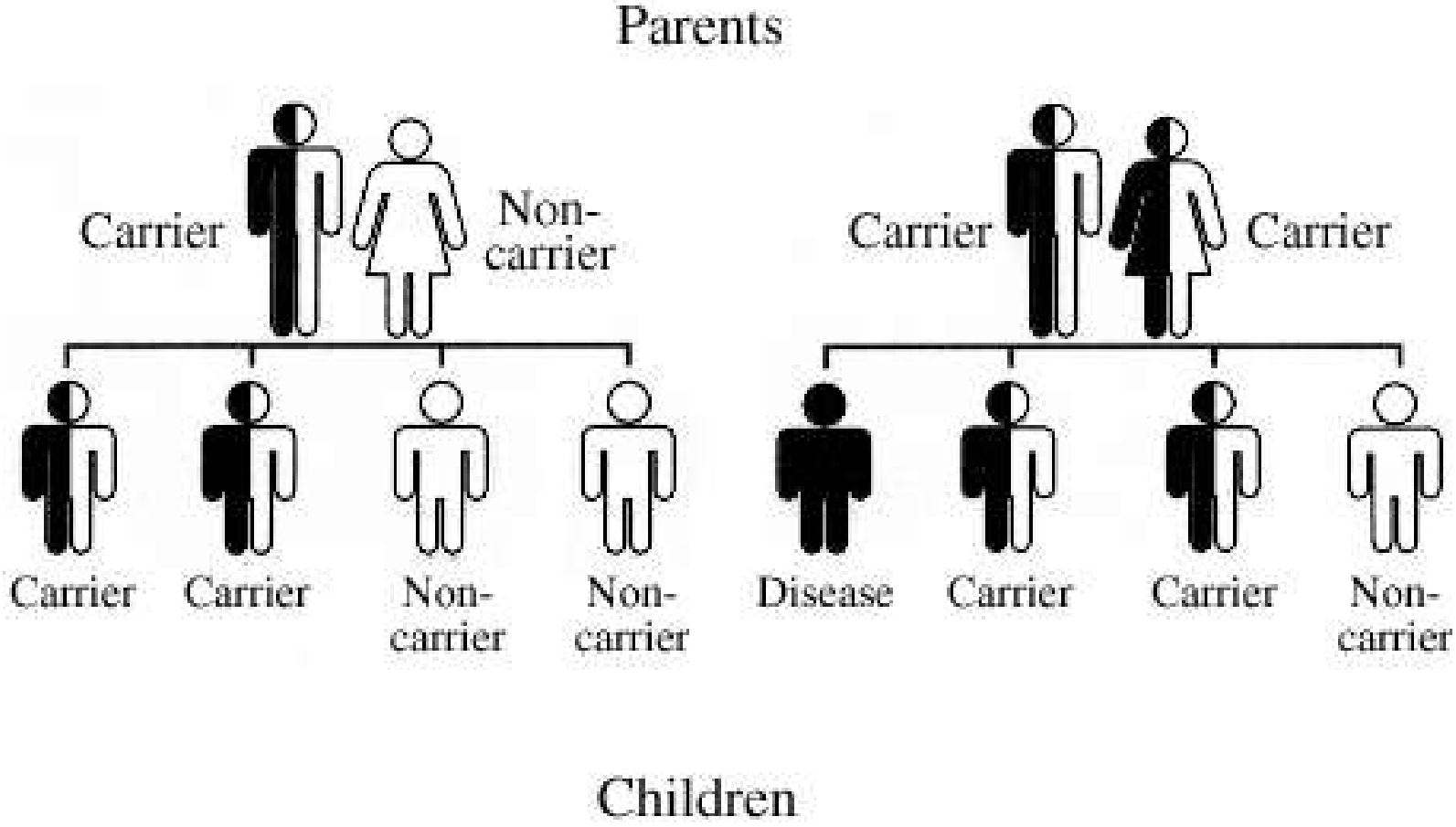
a. Disorders often appear in offspring of parents who are heterozygous

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



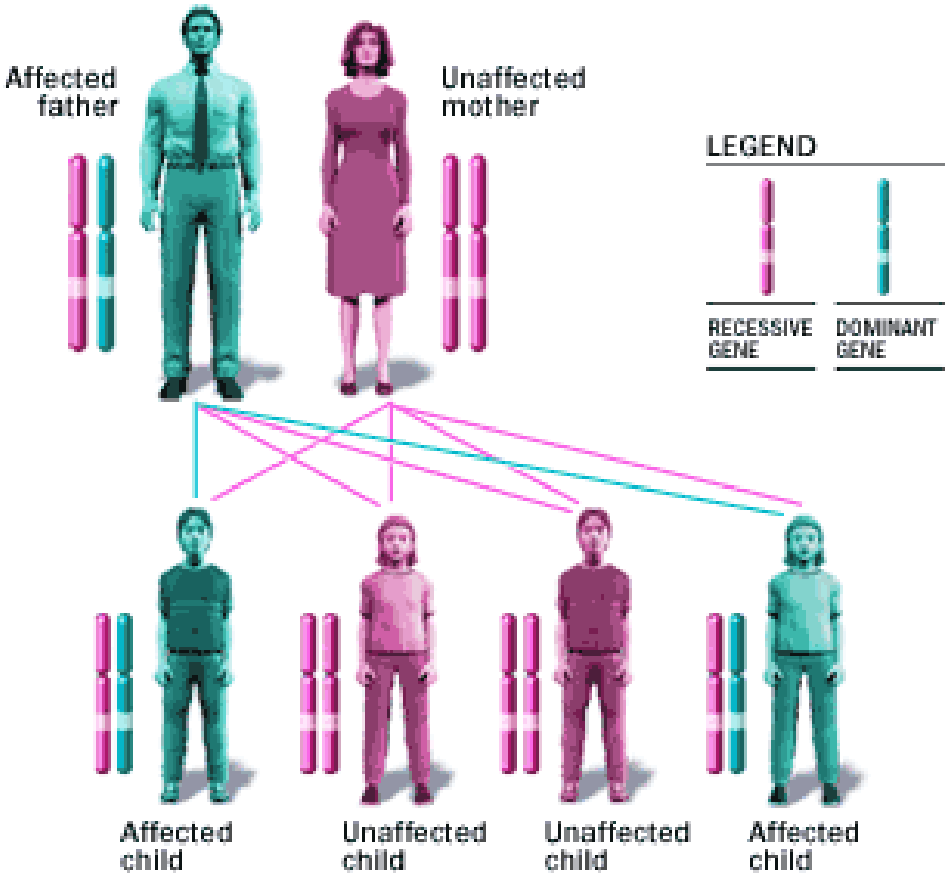
▼ **Figure 14–8** Cystic fibrosis is usually caused by the deletion of three bases in the DNA of a single gene. As a result, the body does not produce normal CFTR, a protein needed to transport chloride ions. Cystic fibrosis causes serious digestive and respiratory problems.

3. A person who is heterozygous for a disease is called a carrier - does not show disease symptoms



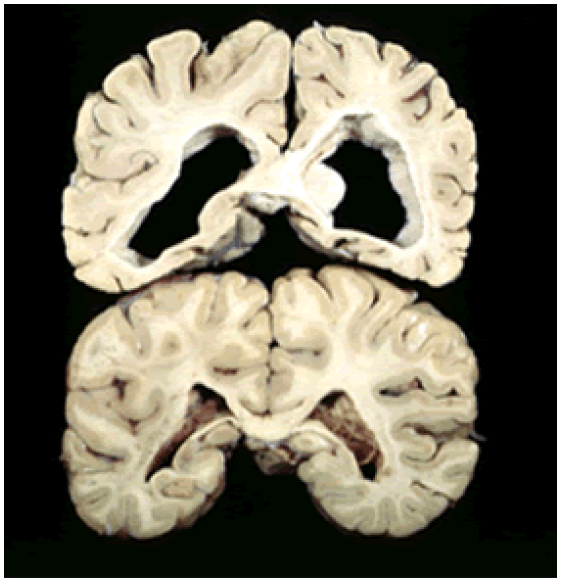
C. Disorders Caused by Dominant Alleles

1. Less common than recessive disorders

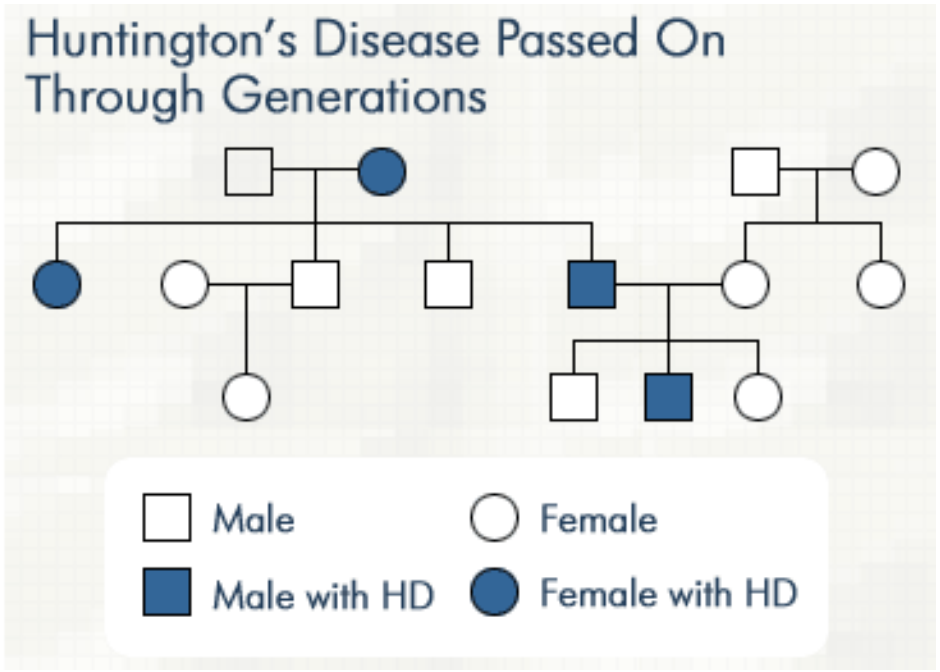


2. Huntington's Disease- damages nervous system and usually appears during adulthood.
a. 75% chance if both parents heterozygous

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

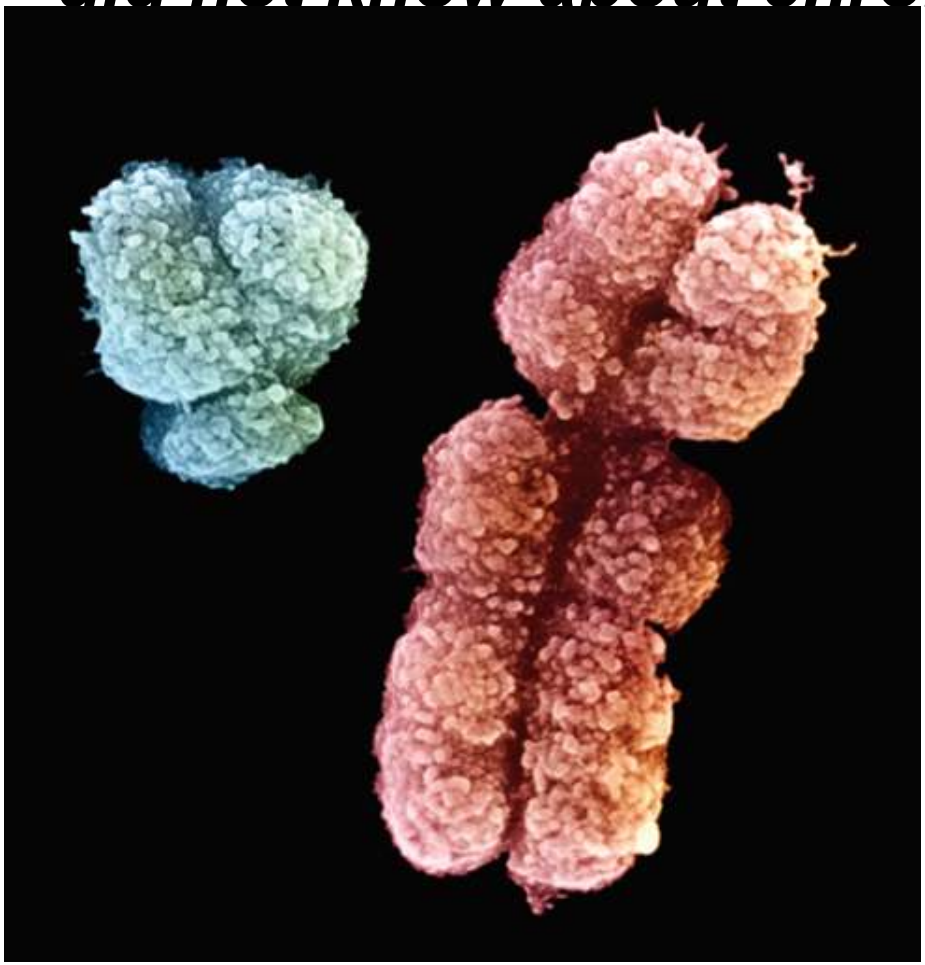


The human brain, showing the impact of HD on brain structure in the basal ganglia region of a person with HD (top) and a normal brain (bottom).



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

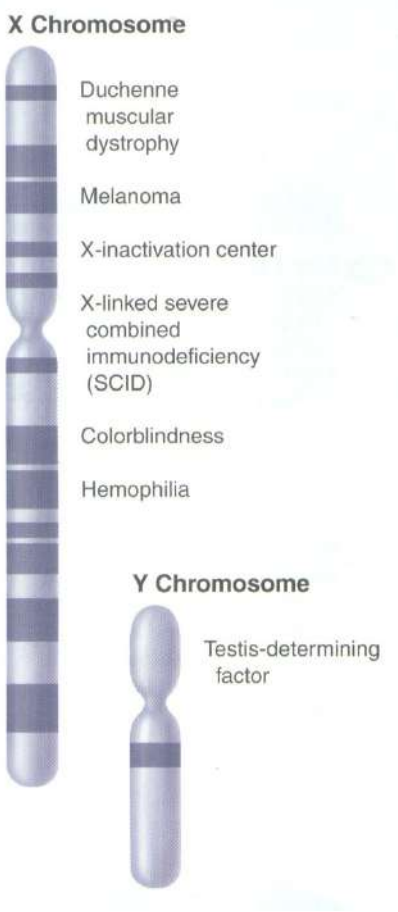
1. Mendel figured out much about heredity, but did not know about chromosomes



a. Mendel only studied autosomal traits

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

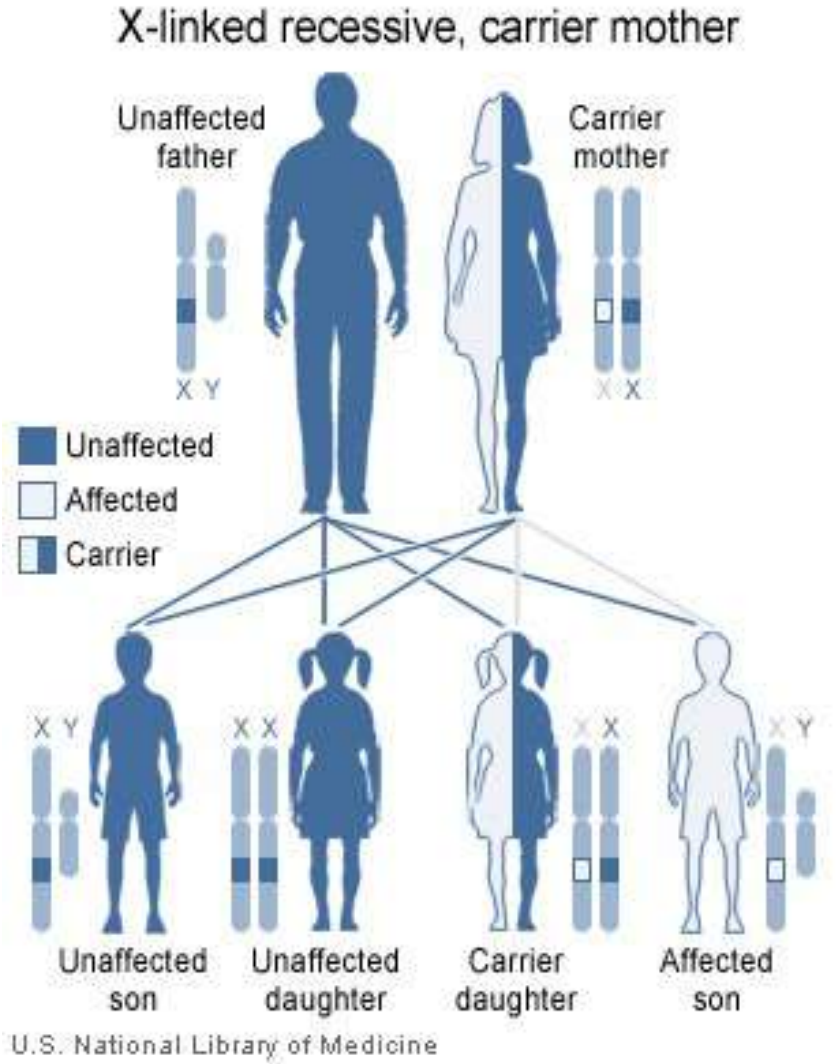
2. Sex-linked Genes

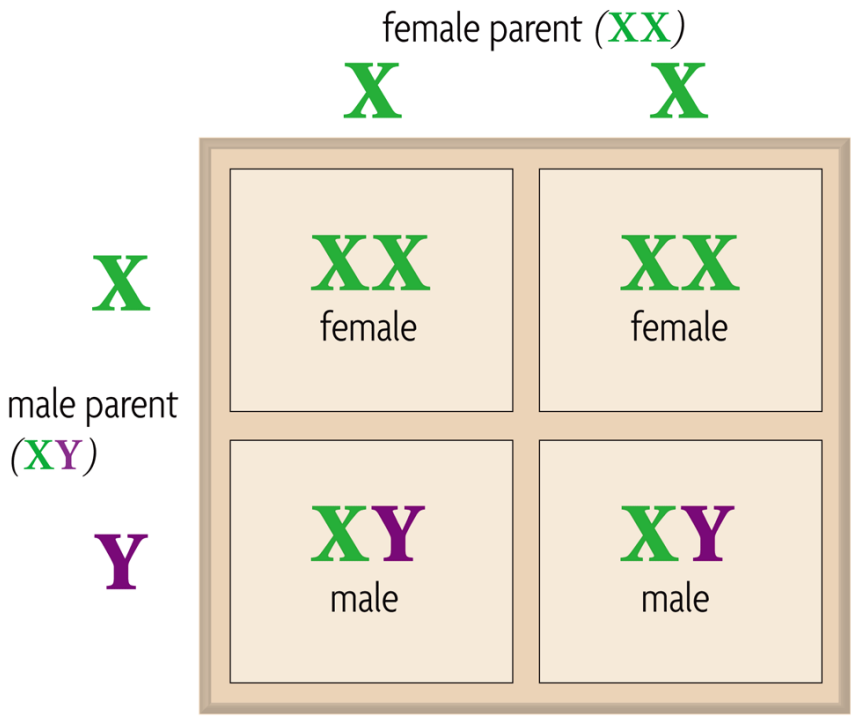


- a. Genes located on sex-chromosomes are called ___ genes
- b. ___ecies have specialized sex ___nes
- c. ___mals and some other animals, ___ with XX are female and XY are male
- d. ___osome <u>much larger</u> thanY

3. Expression of Sex-Linked Genes

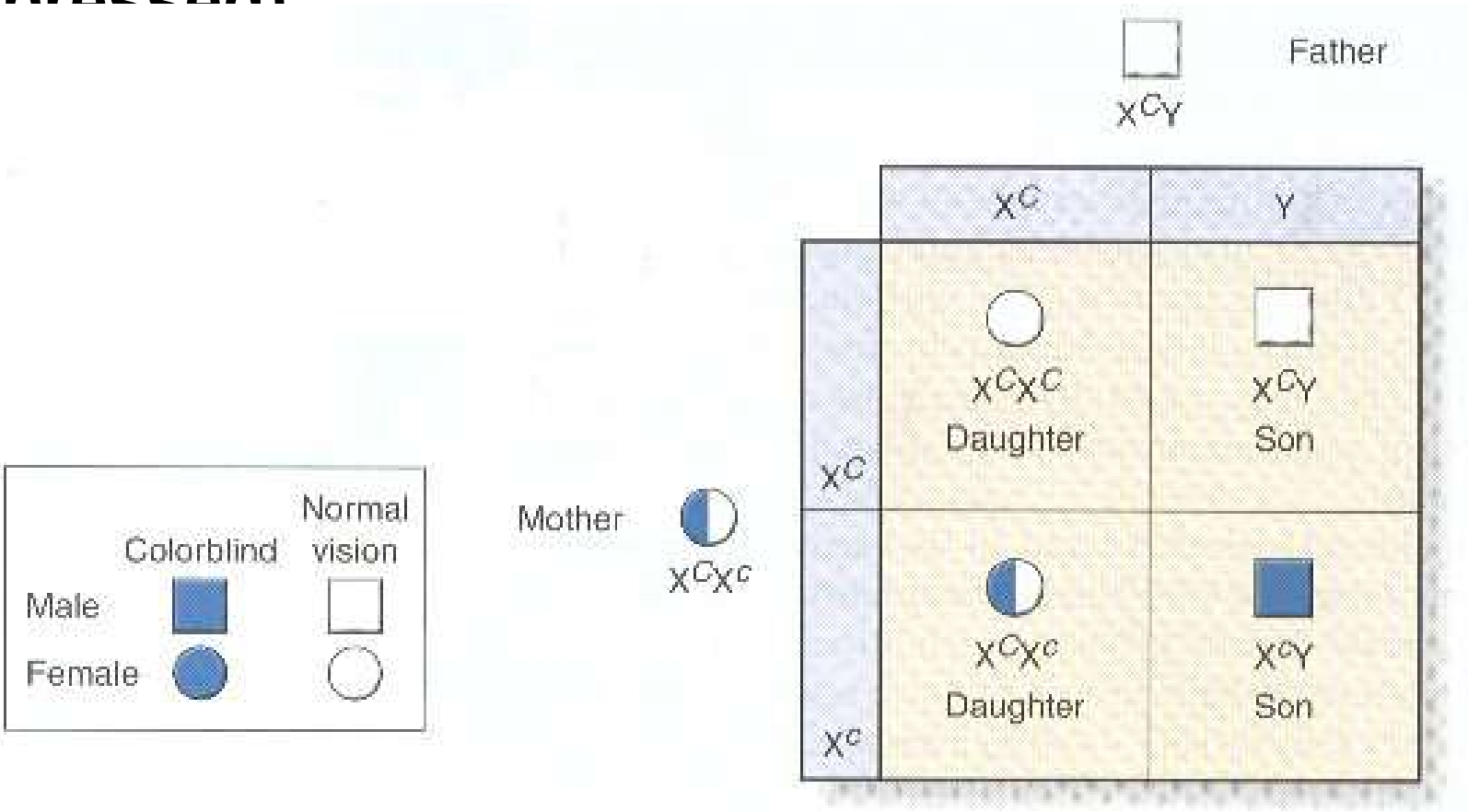
a. Males only have <u>one copy</u> of each chromosome (XY)



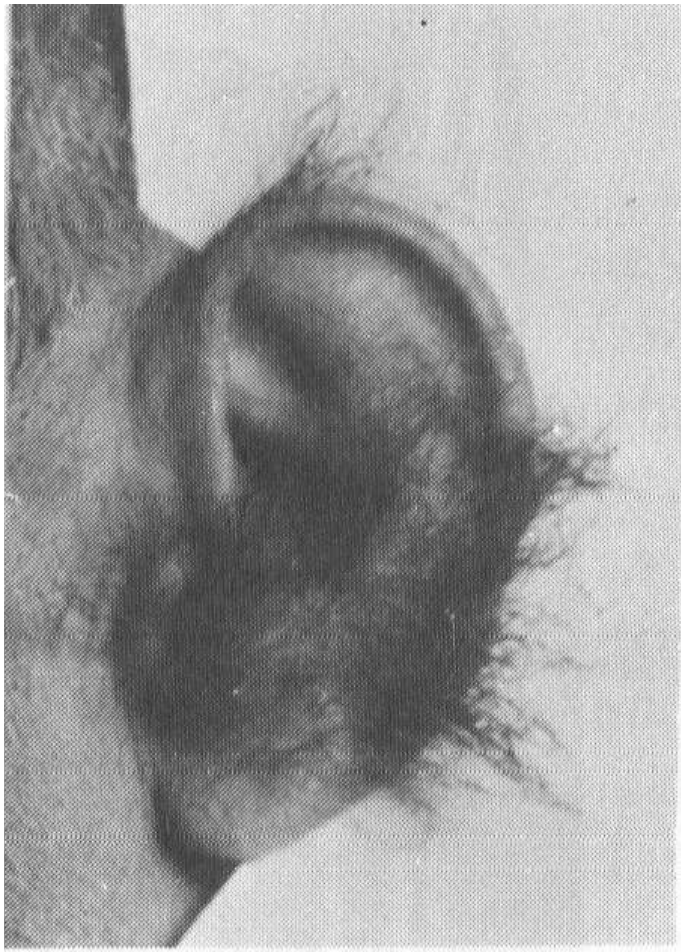


1). Express all alleles on each chromosome

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



***Y-linked Genes* –caused by gene found on Y chromosome that is not homologous with X chromosome (called *holandric gene*) Not many holandric genes**



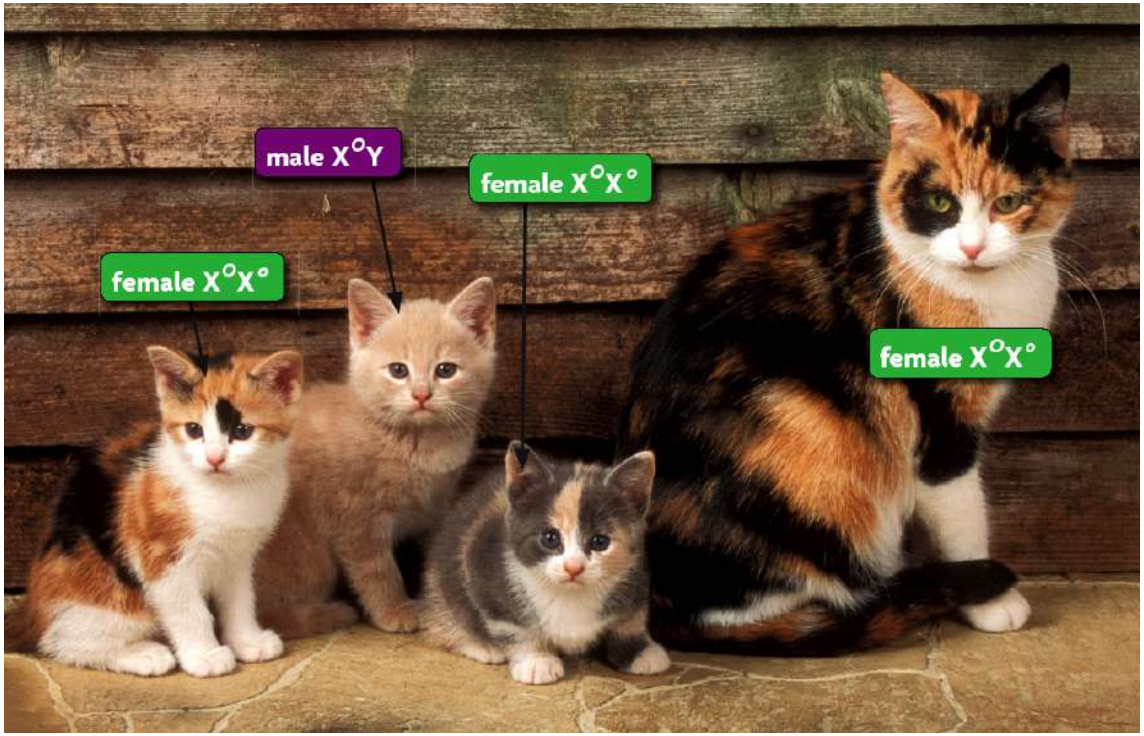
Is it possible for a female to inherit a holandric trait?

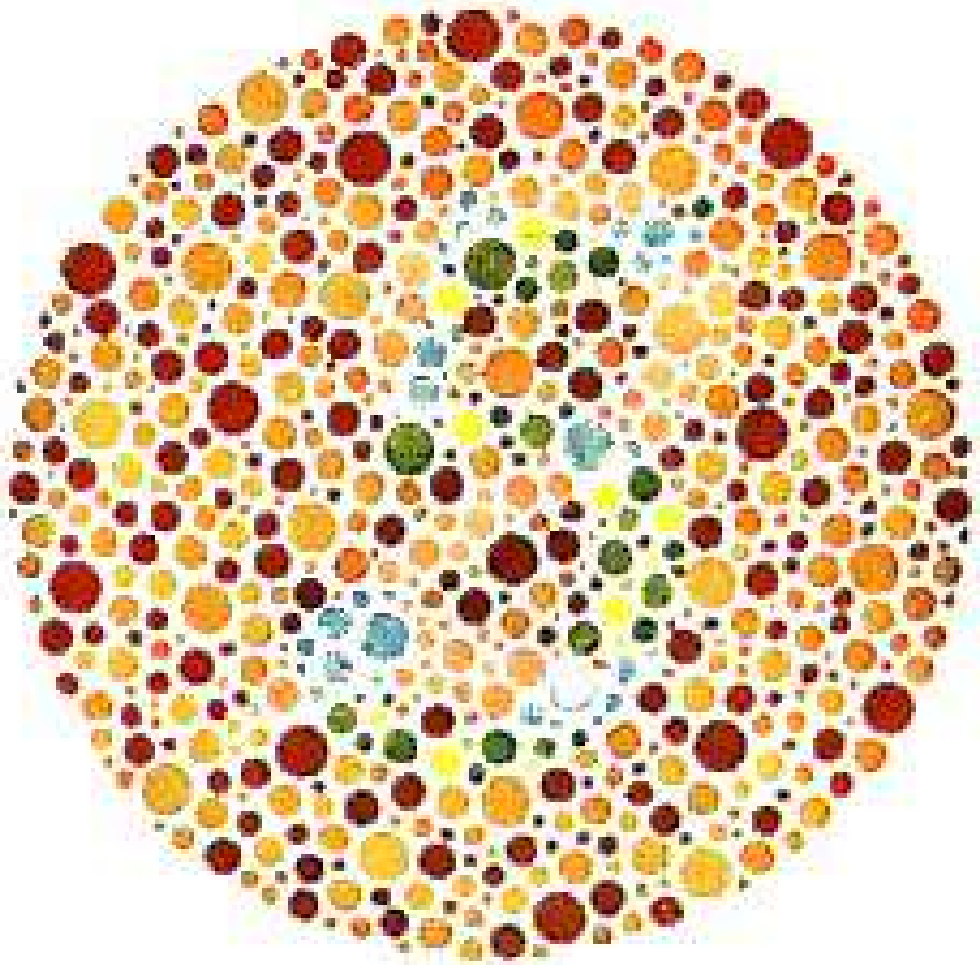
“Hairy Ears”- (hypertrichosis)

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

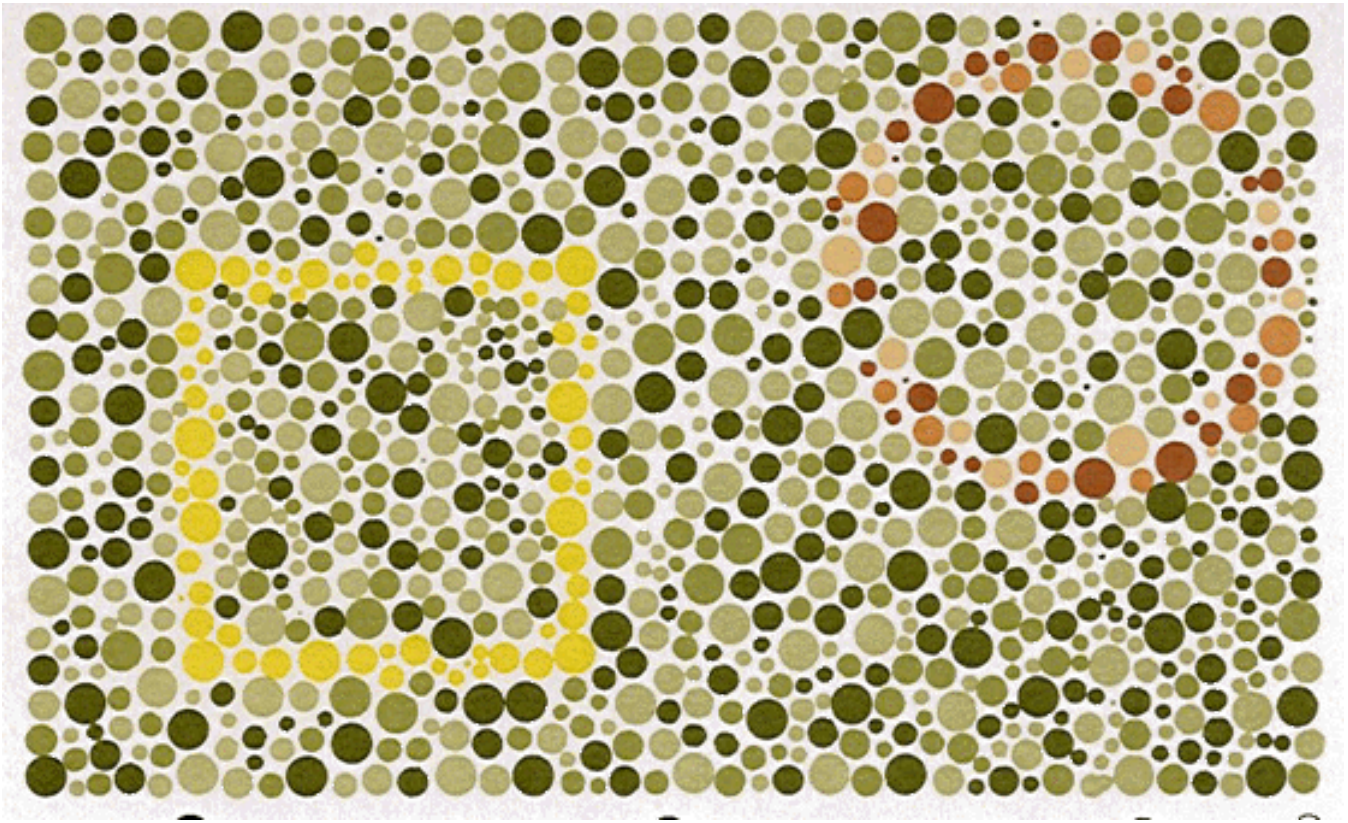




The individual with normal color vision will see a 5 revealed in the dot pattern.

WHAT NUMBER DO YOU SEE?

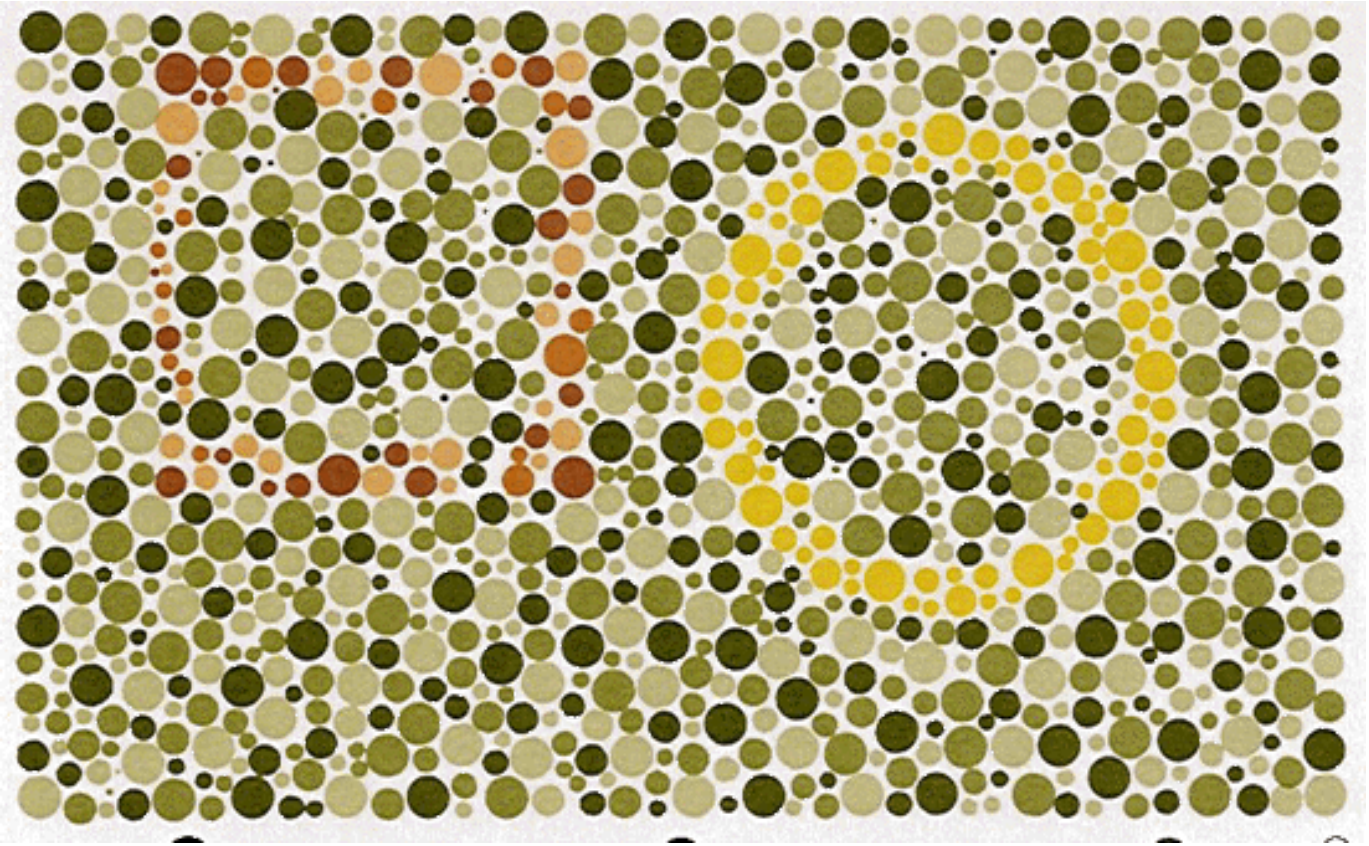
common) color blindness will see a 2 revealed in the dots.



Colorblind individuals should see the yellow square.

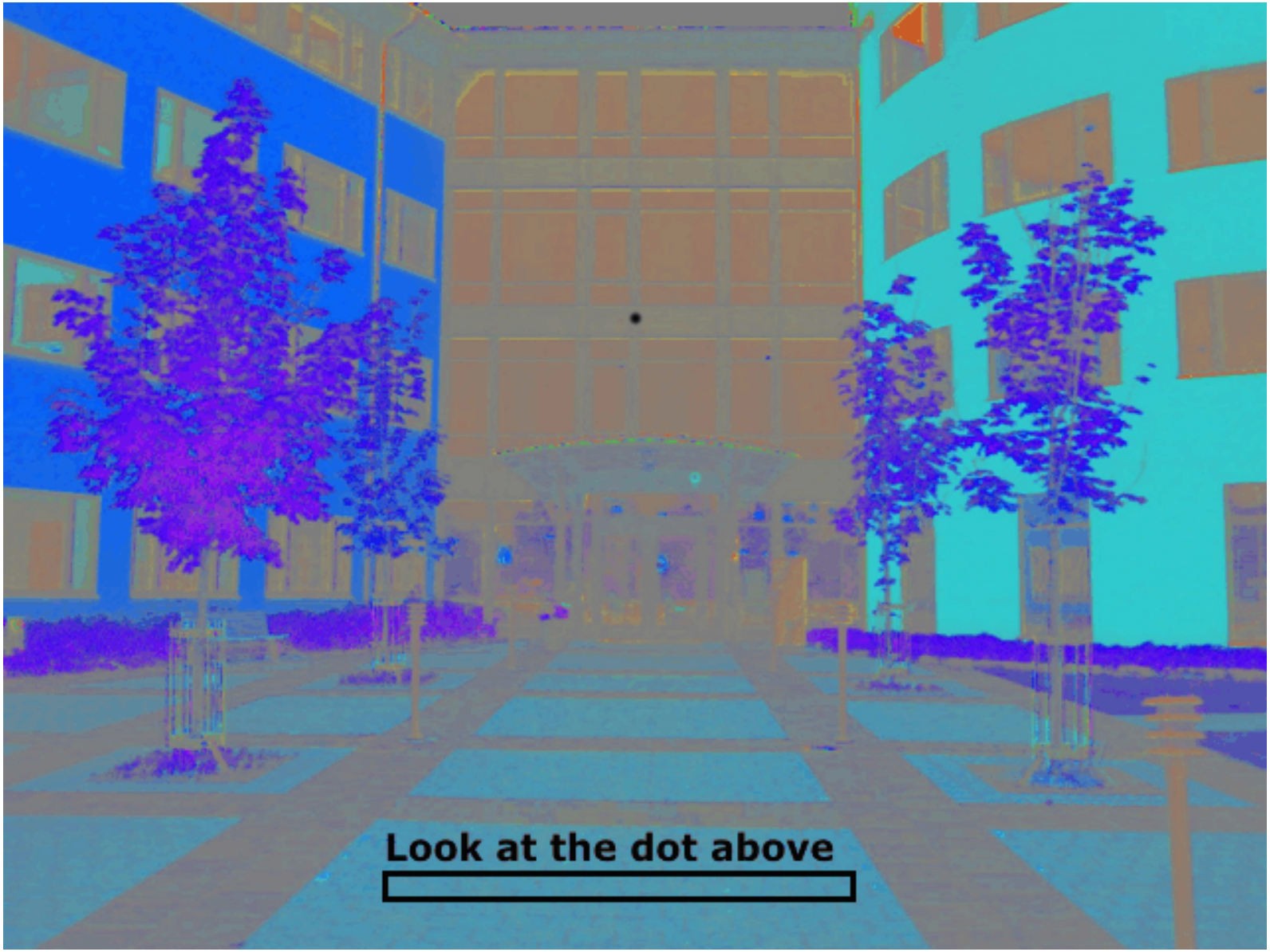
Color normal individuals should see the yellow square and a "faint" brown circle.

Try and find a circle, star, and/or square in 3 seconds.

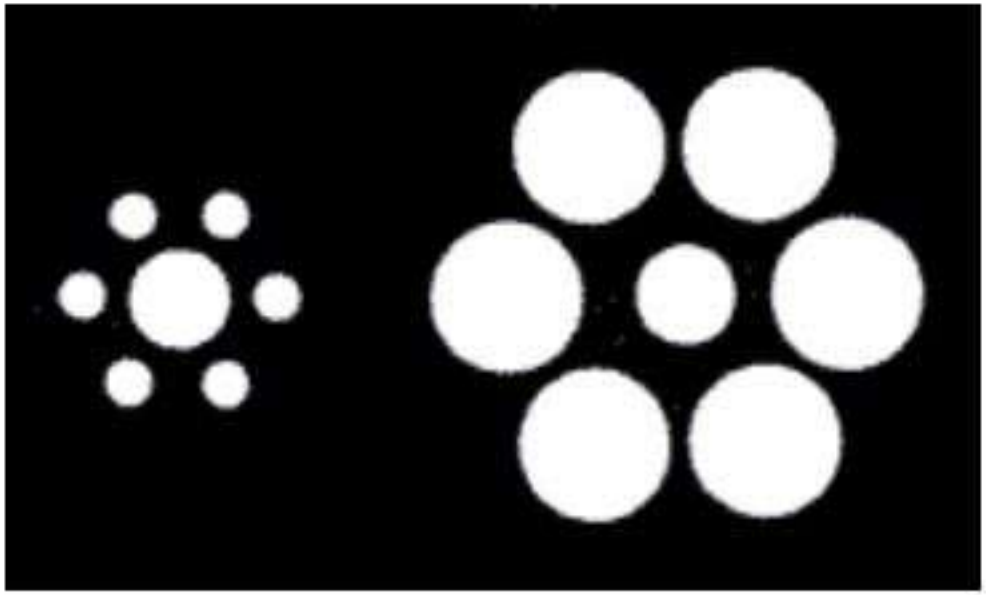


Try and find a circle, star, and/or square in 3 seconds.

Colorblind individuals should see the yellow circle. Color normal individuals should see the yellow circle and a "faint" brown square.


Look at the dot above

Is the left center circle bigger?



No, they're both the same size

YOUNG WOMAN OR OLD WOMAN?

Multiple Alleles- More than two possible alleles exist for a trait (e.g. blood type, eye color)

Figure 11–12 Coat color in rabbits is determined by a single gene that has at least four different alleles. Different combinations of alleles result in the four colors you see here. **Interpreting Graphics** *What allele combinations can a chinchilla rabbit have?*

Full color: CC, Cc^{ch}, Cc^{h}, or Cc

Chinchilla: $c^{ch}c^{h}$, $c^{ch}c^{ch}$, or $c^{ch}c$

Himalayan: $c^{h}c$ or $c^{h}c^{h}$

Albino: cc

Key

- C = full color; dominant to all other alleles
- c^{ch} = chinchilla; partial defect in pigmentation; dominant to c^{h} and c alleles
- c^{h} = Himalayan; color in certain parts of body; dominant to c allele
- c = albino; no color; recessive to all other alleles

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
green	B_1B_1

Phenotype	Genotype
Steel blue	B_2B_2

Phenotype	Genotype
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

=

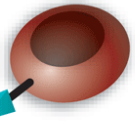
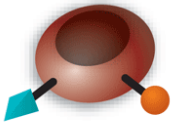
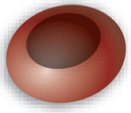
roan

Co-Dominance

In certain varities of chickens, black and white feather colors are caused by codominant alleles. Thus the heterozygous phenotype, speckled black and white, is a result of the expression of both alleles

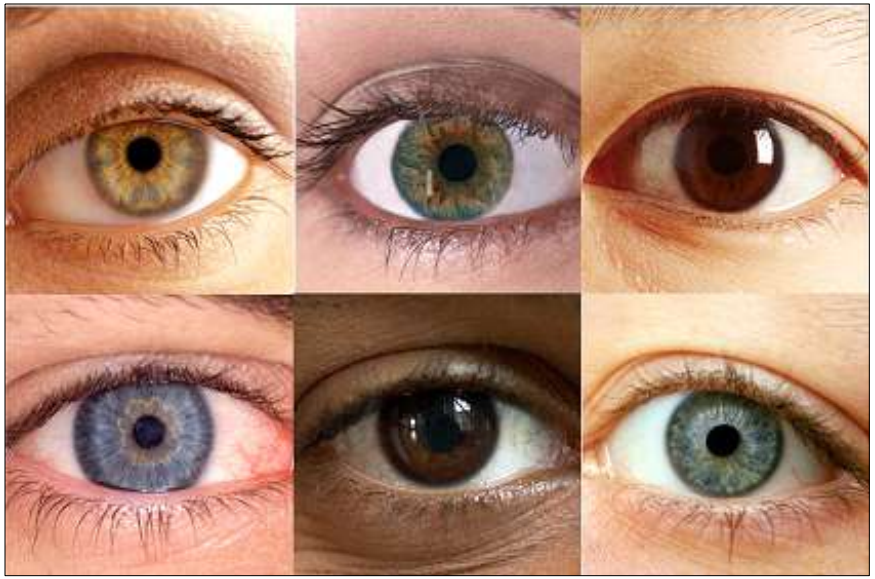
3. Human blood type is example of codominance

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)

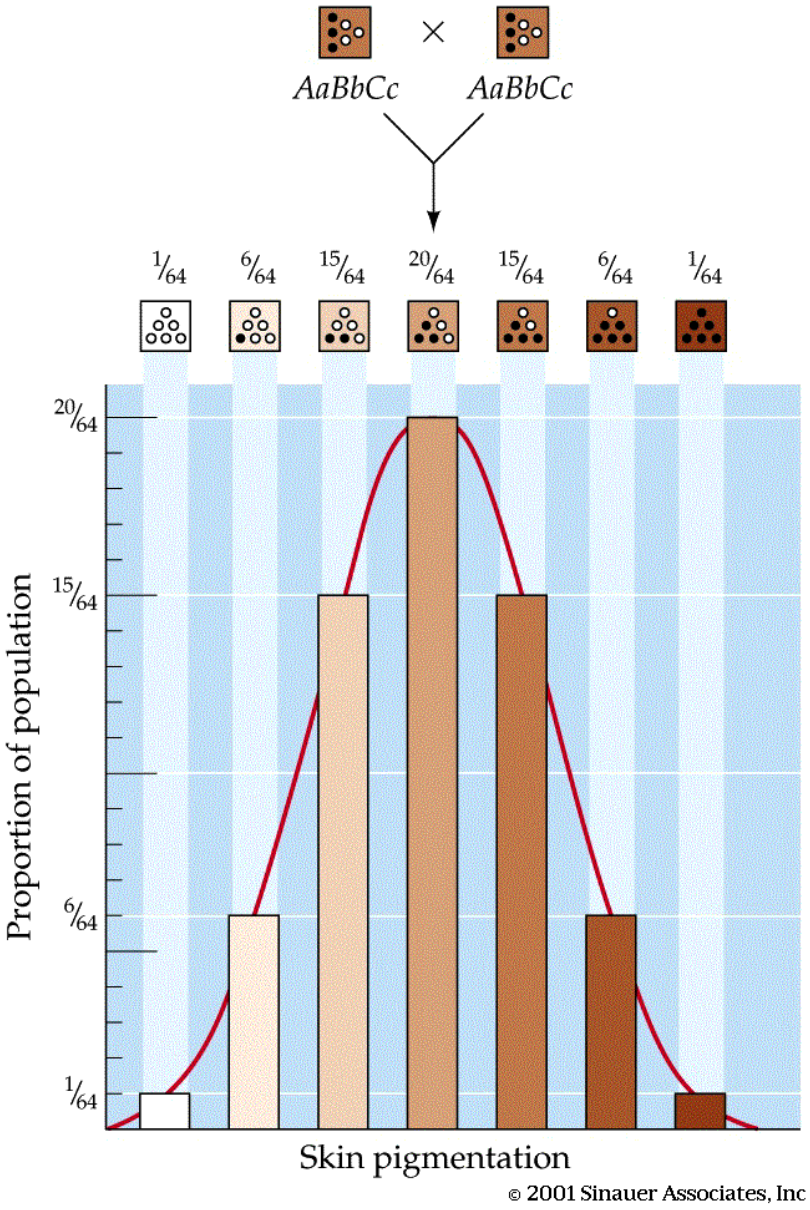
b. Human eye color shows at least 3 genes (hypothesize that are still genes undiscovered as well) Order of dominance: brown > green > blue.



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

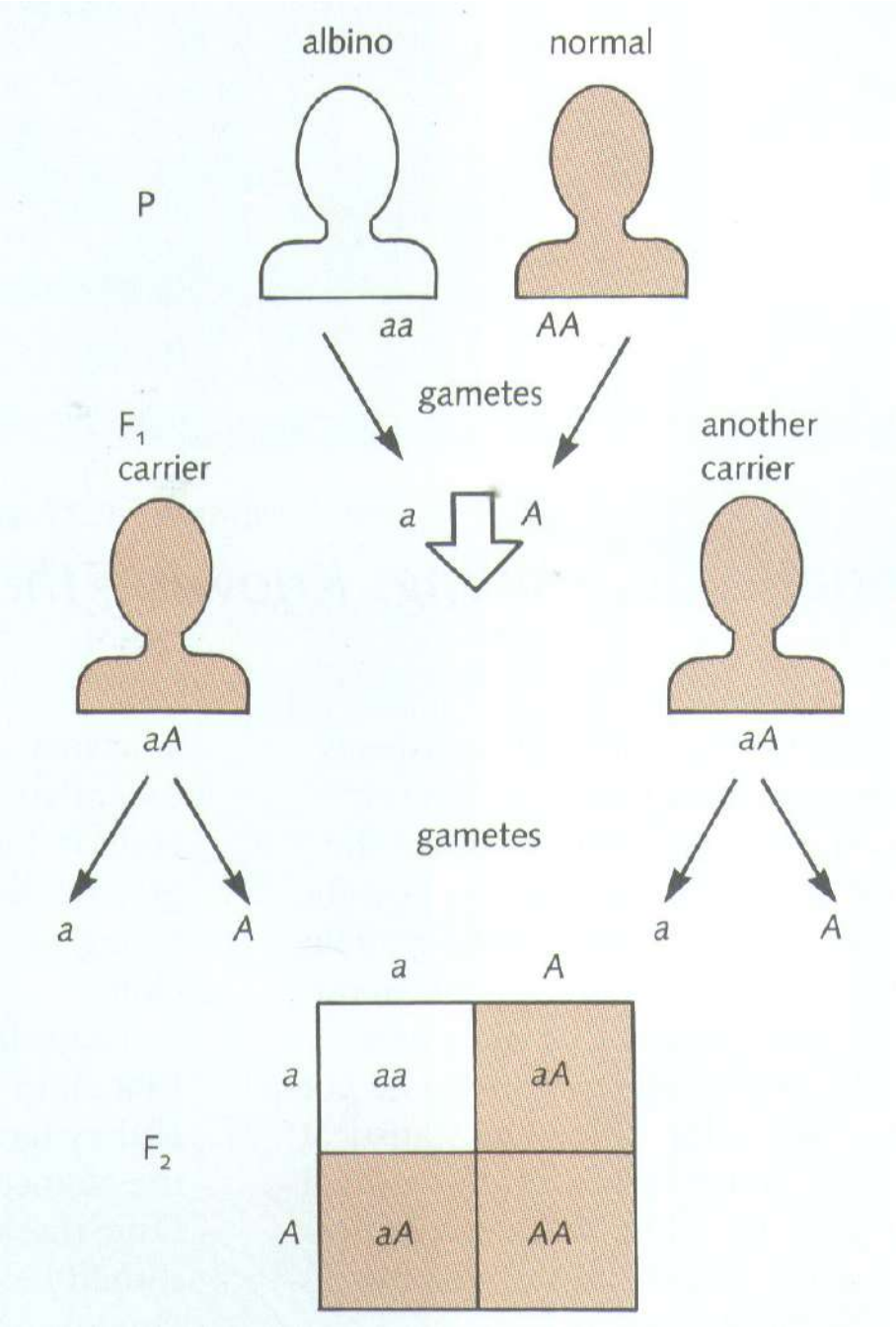
2. Epistasis- when one gene overshadows all of the others. Albinism is caused by this type of gene

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



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

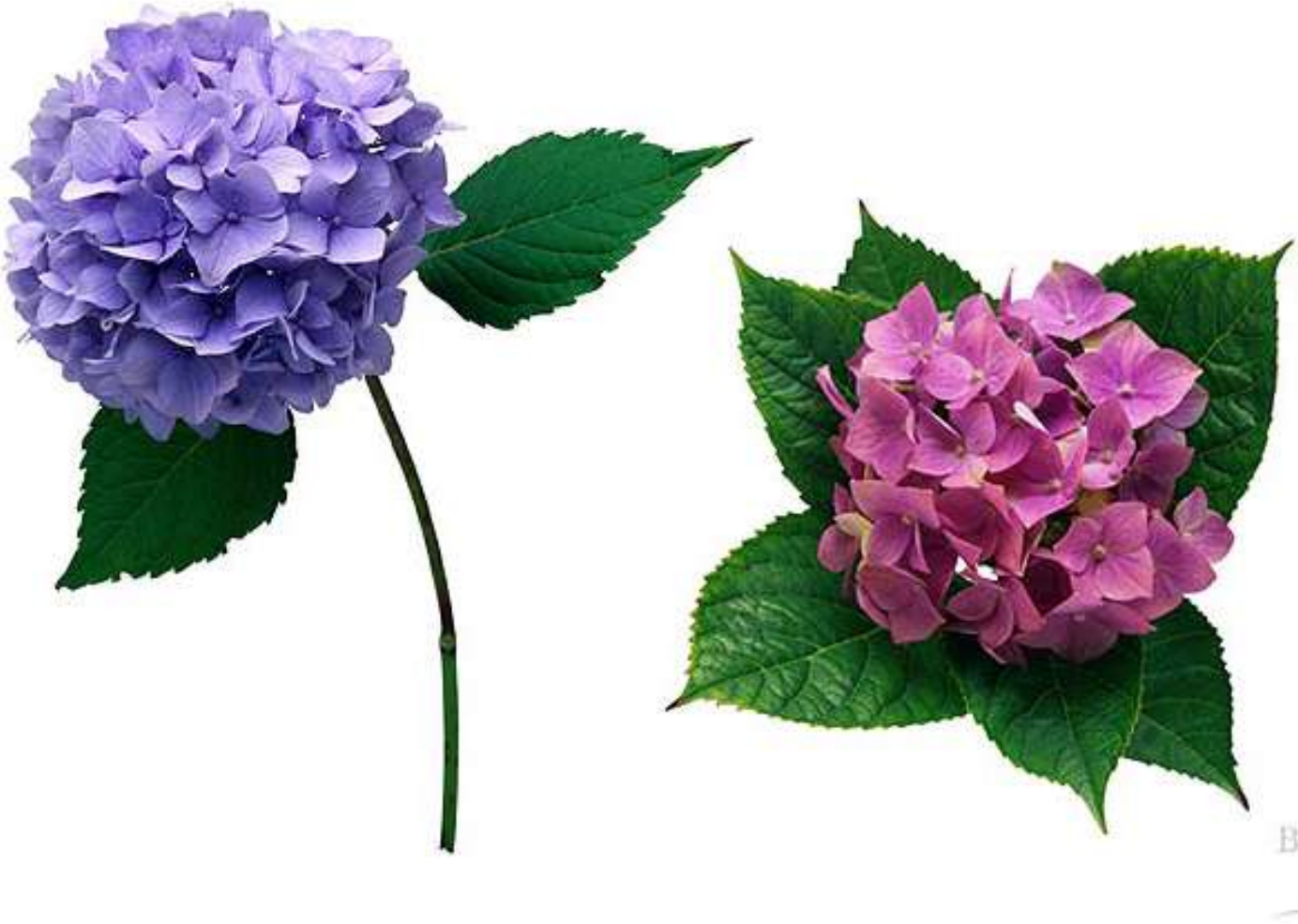
a. Phenotype is more than sum of gene expression

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)

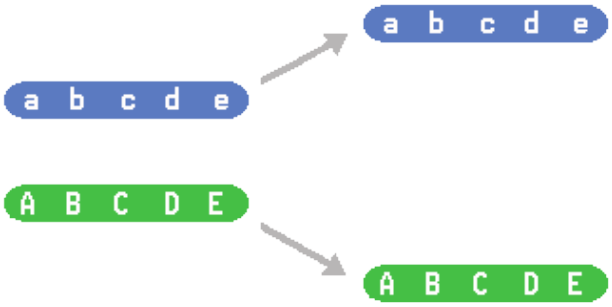
The expression of coat color genes in Siamese cats varies with temperatures. Black pigment is produced only in those areas of the skin which are lowest in temperature, such as the ears and tail

Color of Hydrangea flowers is influenced by soil pH

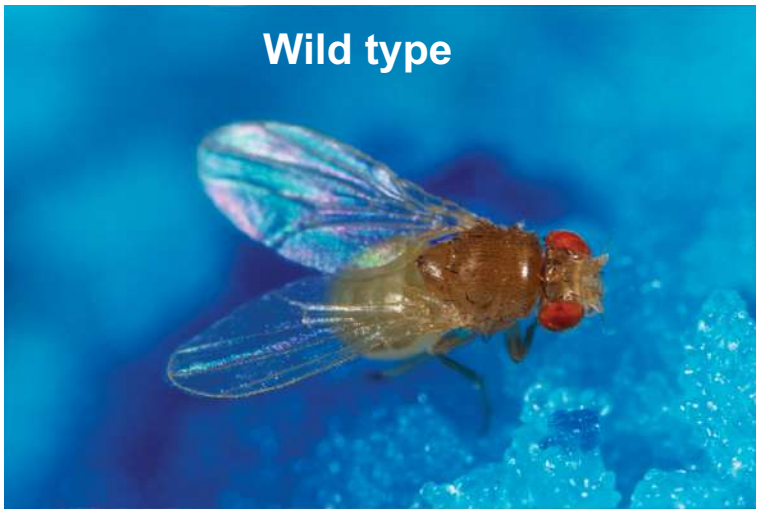


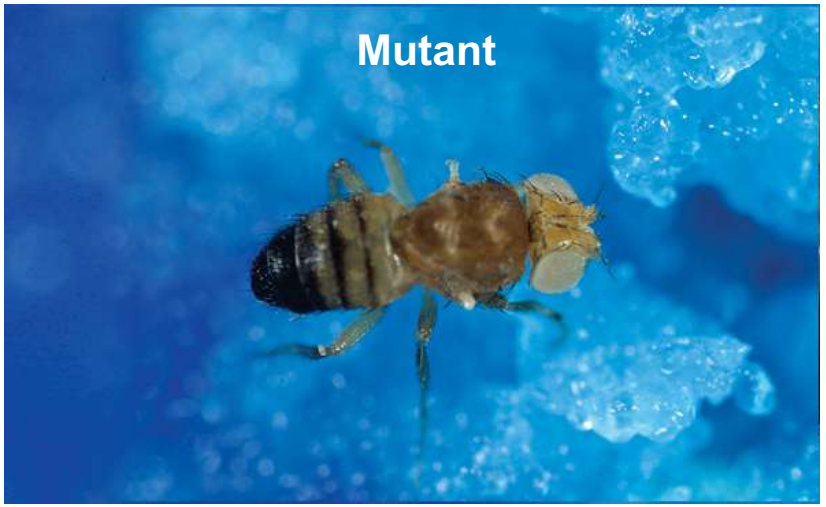
III. Gene Linkage and Mapping (7.3)

A. Gene linkage was explained through fruit flies

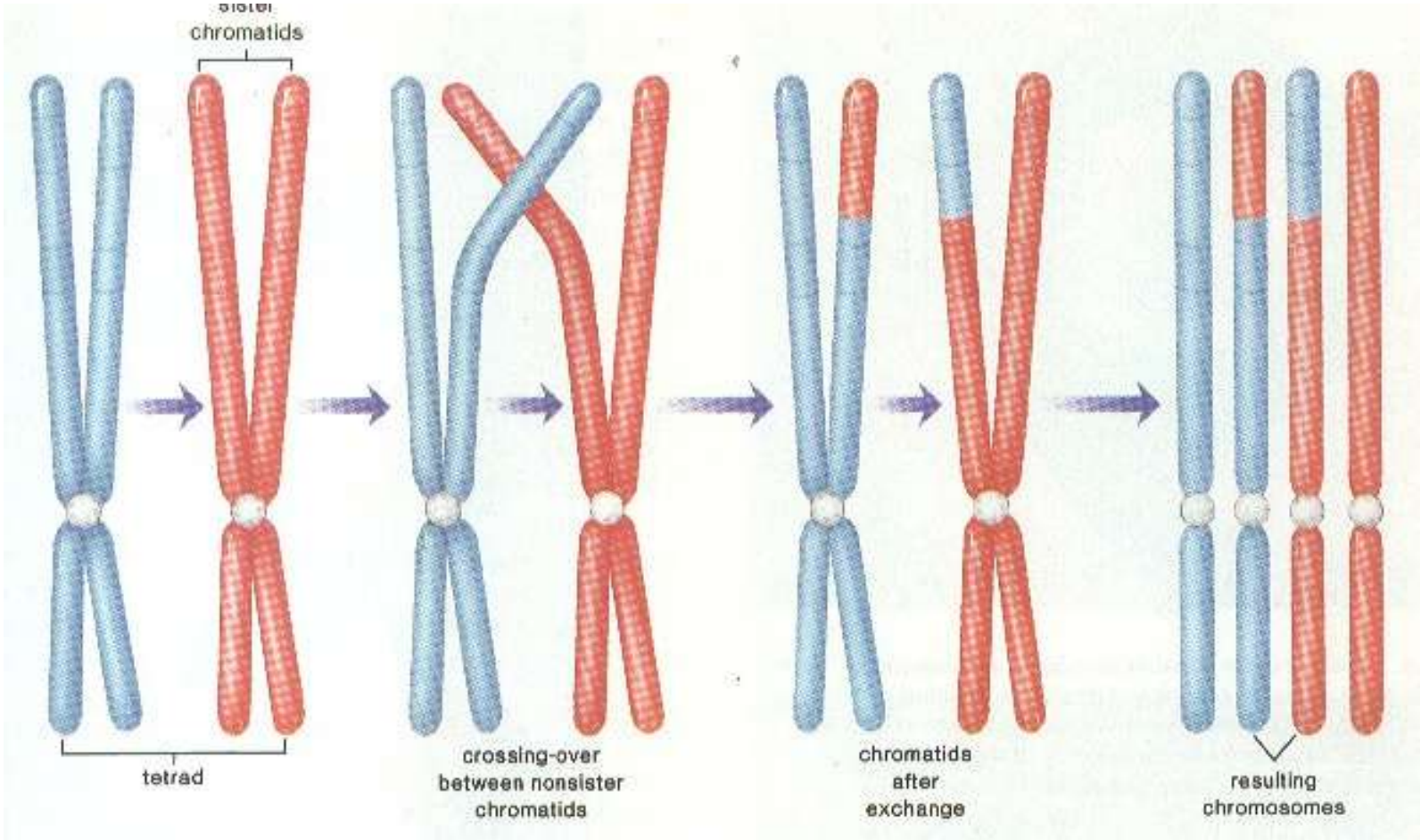


1. Thomas Hunt Morgan worked with fruit flies (Drosophila melanogaster)
2. Some traits seemed to be inherited together. Morgan called them linked traits. (found on same chromosome)





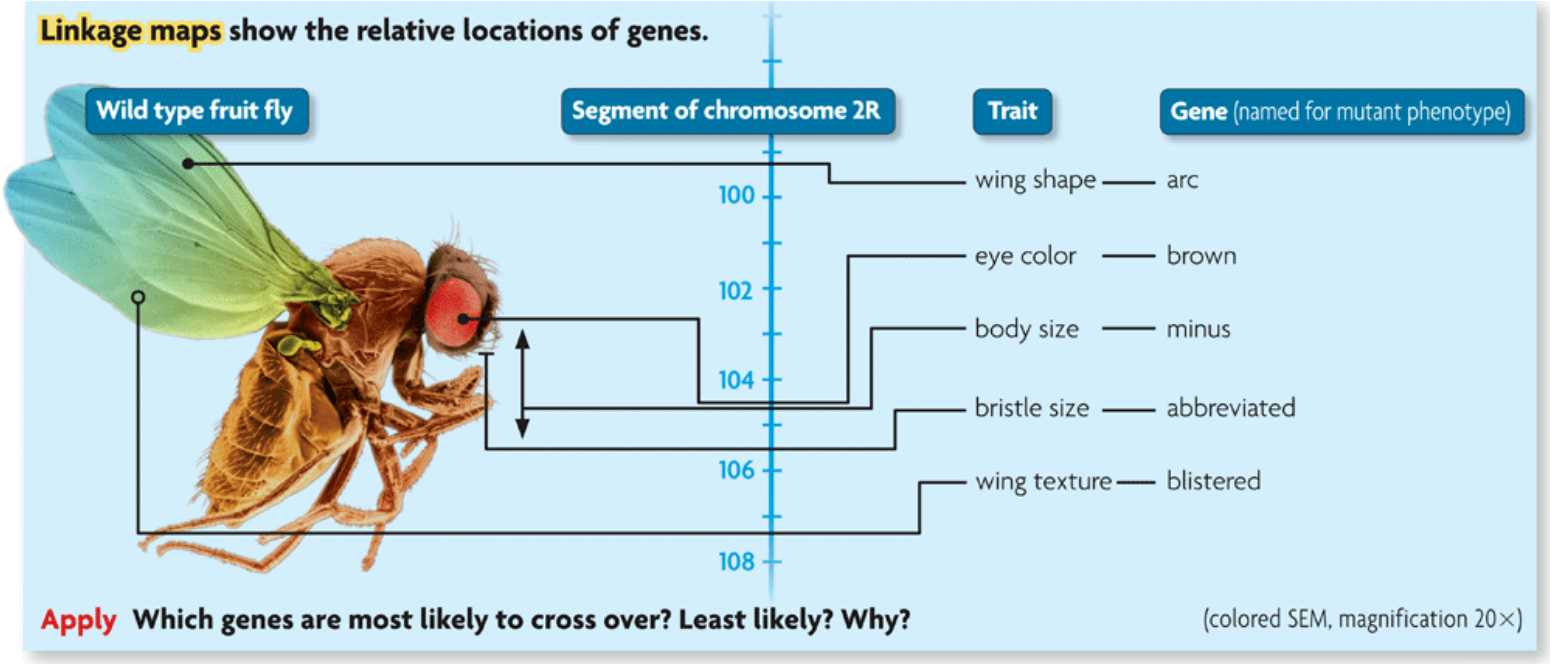
3. Morgan concluded that because linked genes were not inherited together every time that chromosomes must exchange homologous genes during meiosis (crossing over)

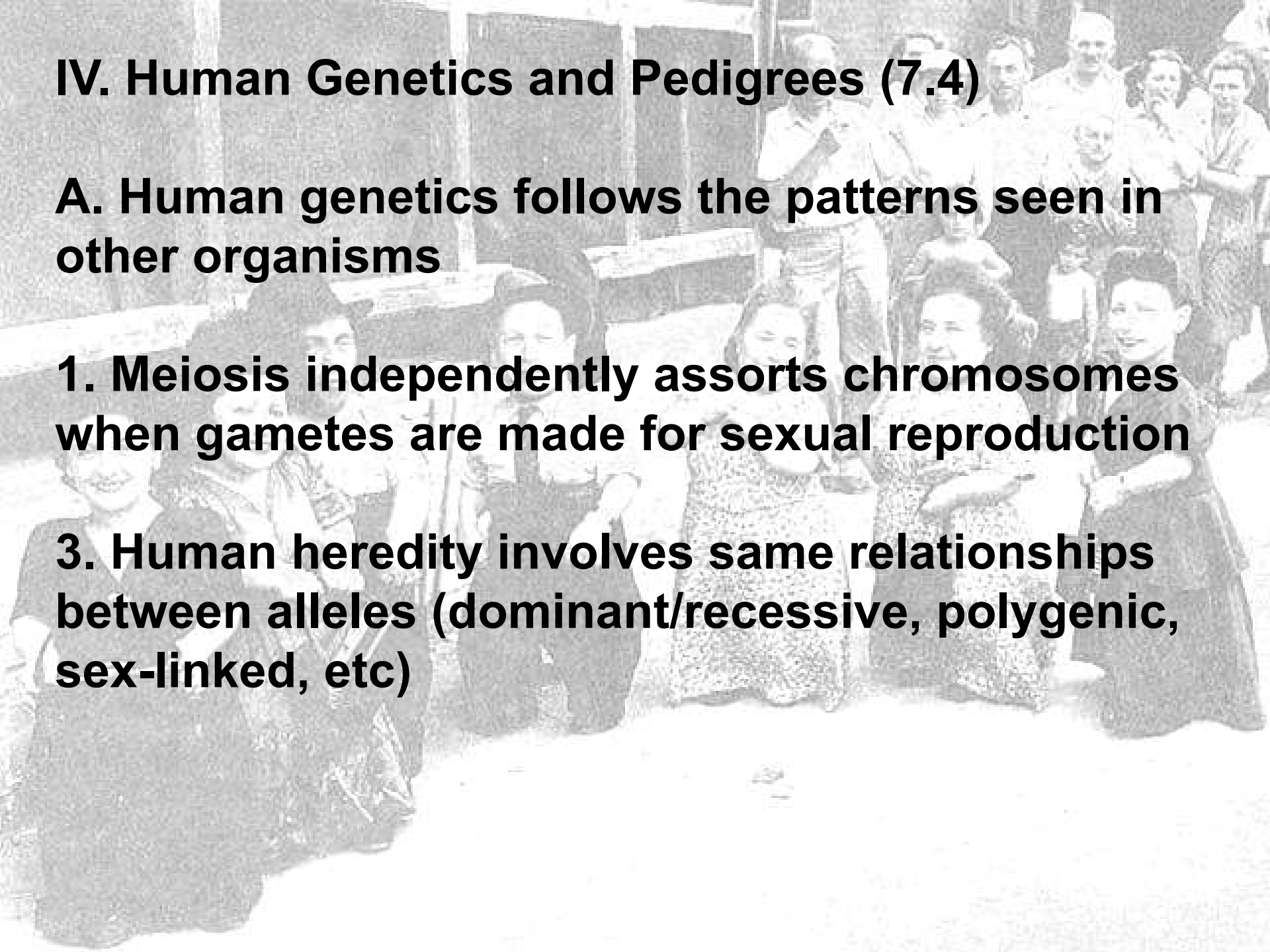


B. Linkage maps estimate distances between genes

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

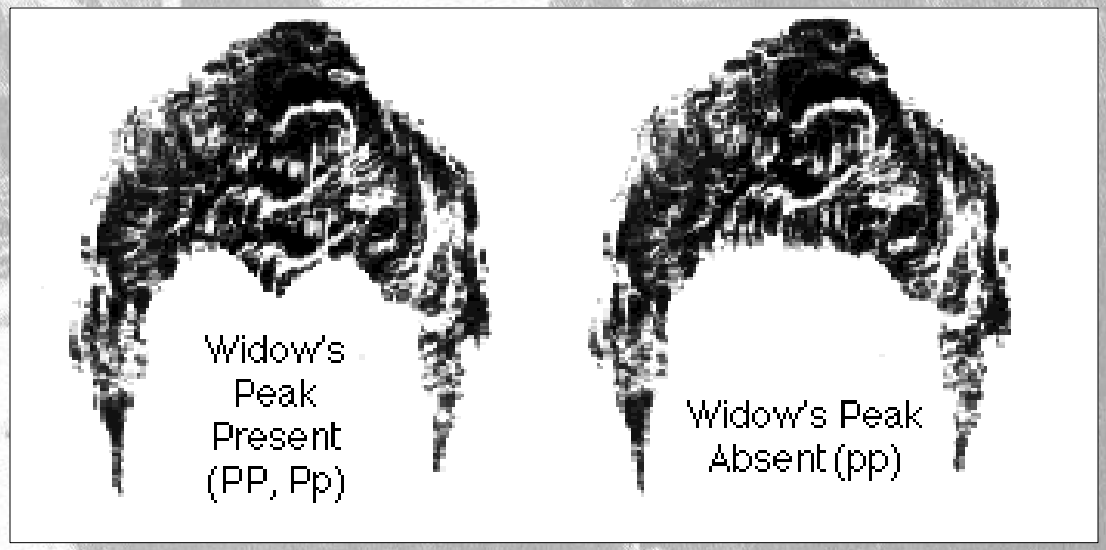
3. Human heredity involves same relationships between alleles (dominant/recessive, polygenic, sex-linked, etc)

B. Inheritance of some traits very complex

1. Multiple genes and alleles can interact

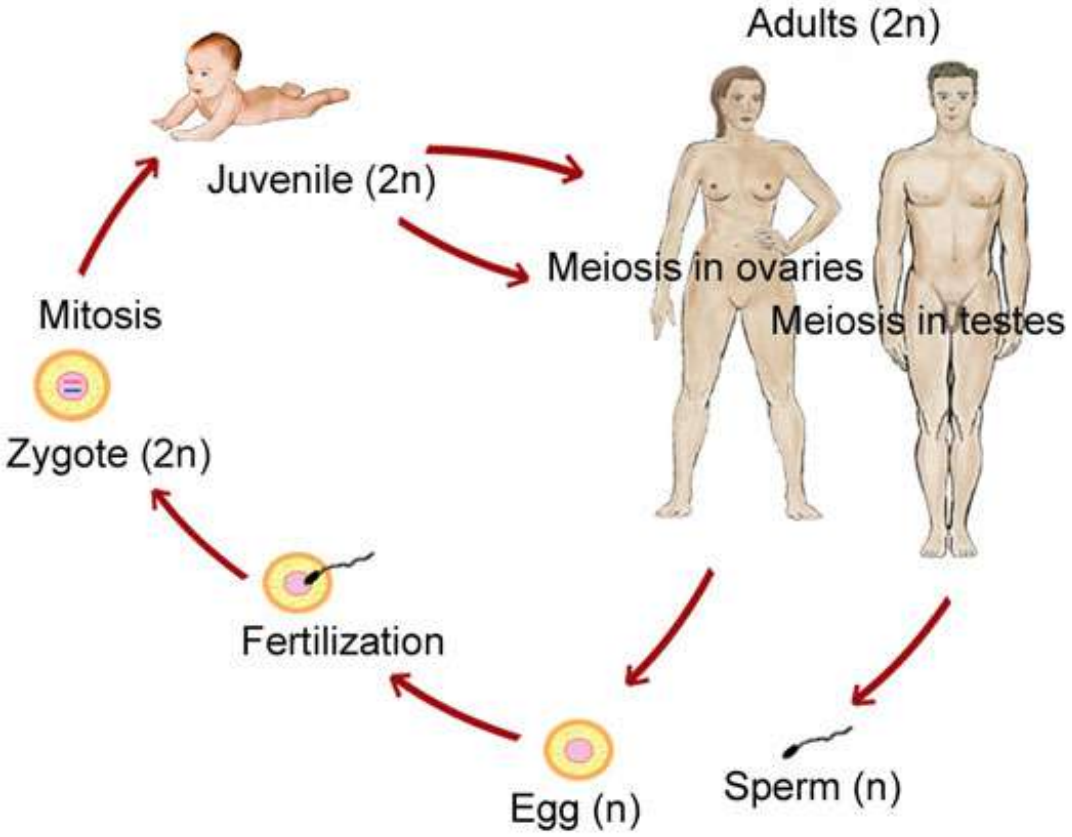
2. Single-gene traits can still be observed

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)

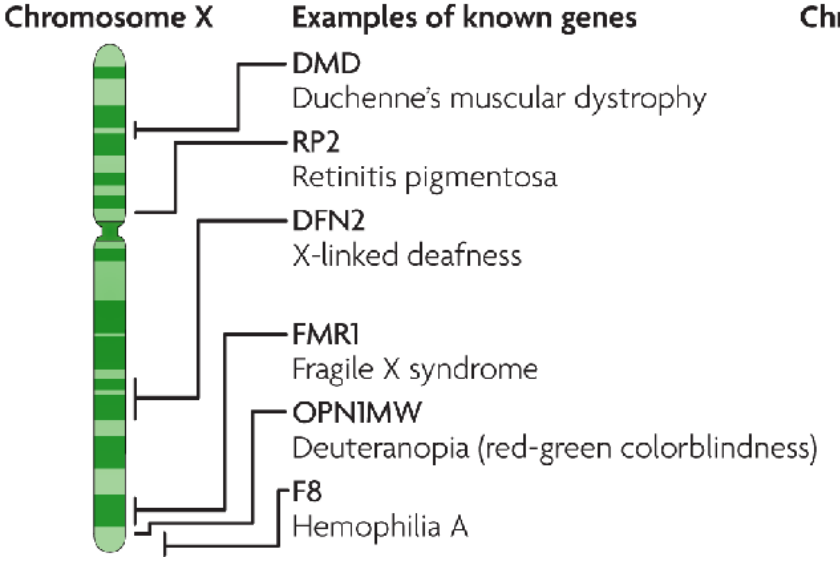
c. Much of what is known about human genetics comes from studying genetic disorders

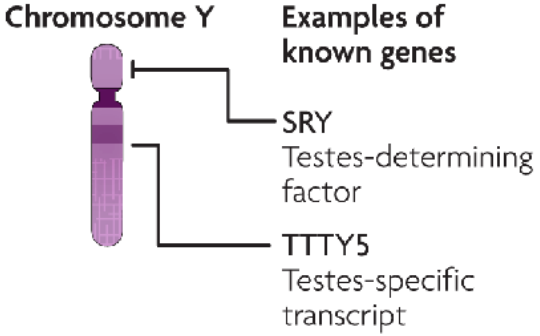


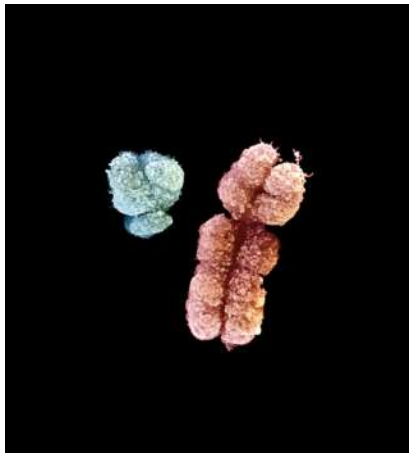
C. Females can carry a sex-linked genetic disorder

1. Both male and females can be <u>carriers</u> of autosomal disorders

2. Only females can be <u>carriers</u> of sex-linked disorders



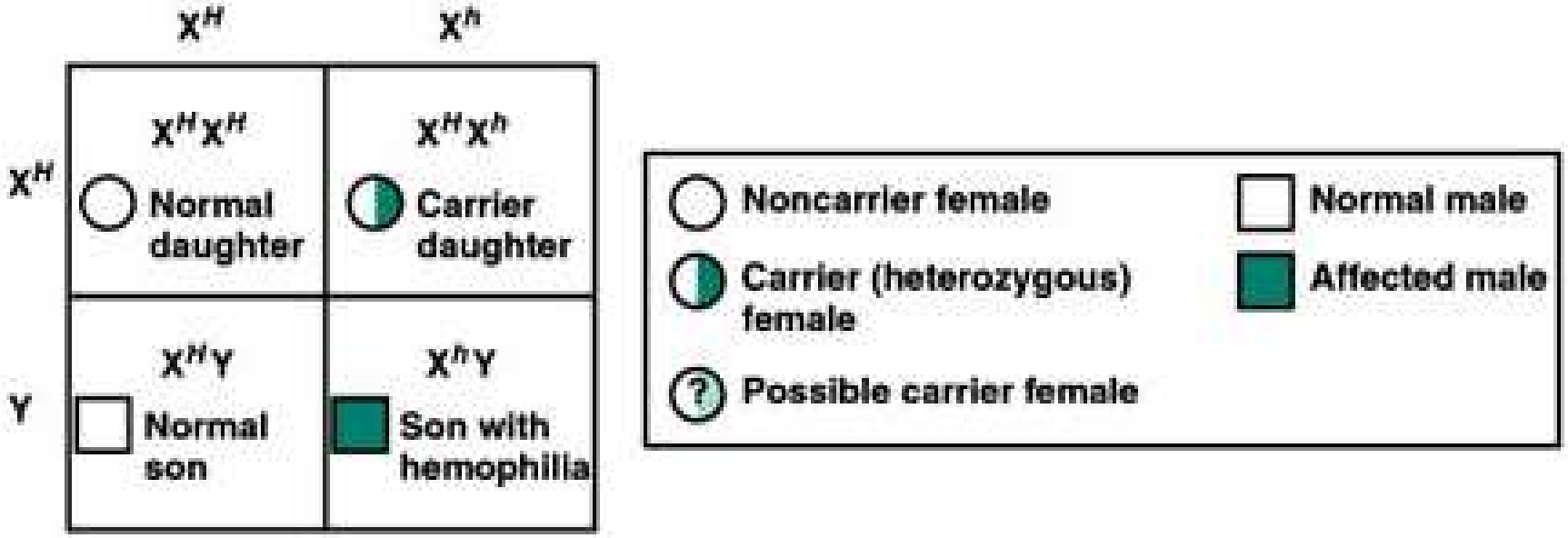




3. Many genetic disorders carried on X-chromosome

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

b. Males <u>more likely</u> to have this disorder



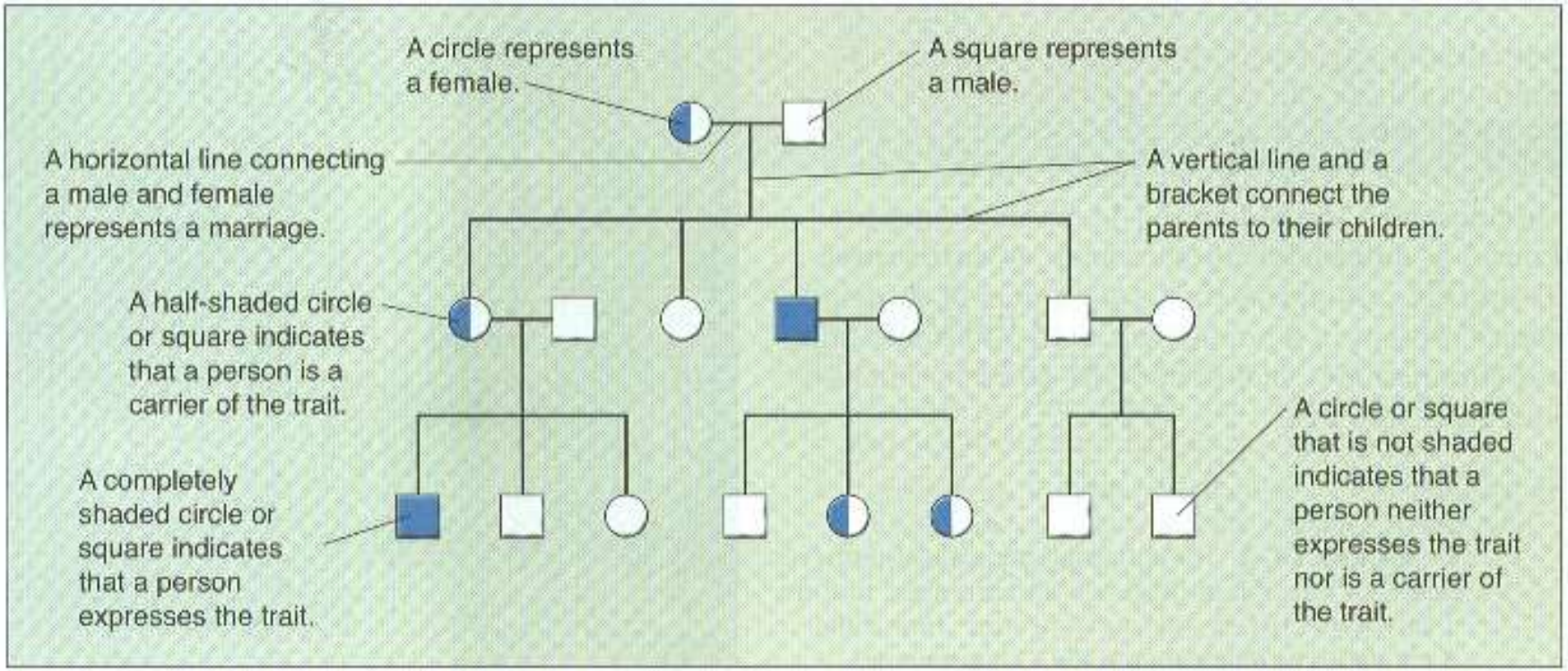
Some X-linked Recessive Genetic Traits

TRAITS	PHENOTYPE
Adrenoleukodystrophy	Atrophy of adrenal glands, mental deterioration; death 1–5 years after onset
Color-blindness	
Deutan	Insensitive to green light; 60%–75% of color blindness
Protan	Insensitivity to red light; 25%–40% of color blindness
Fabry's disease	Metabolic defect caused by lack of enzyme alpha-galactosidase A; progressive cardiac, renal problems, early death
Glucose-6-phosphate dehydrogenase deficiency	Benign condition that can produce severe, even fatal anemia in presence of certain foods, drugs
Hemophilia A	Inability to form blood clots; caused by lack of clotting factor VIII
Hemophilia B	Christmas disease; clotting defect caused by lack of factor IX
Icthyosis	Skin disorder causing large, dark scales on extremities, trunk
Lesch-Nyhan syndrome	Metabolic defect caused by lack of enzyme hypoxanthine-guanine phosphoribosyl transferase (HGPRT); causes mental retardation, self-mutilation, early death
Muscular dystrophy	Duchenne-type, progressive; fatal condition accompanied by muscle wasting
Testicular feminization	Insensitivity to testosterone in XY individual resulting in female sexual phenotype

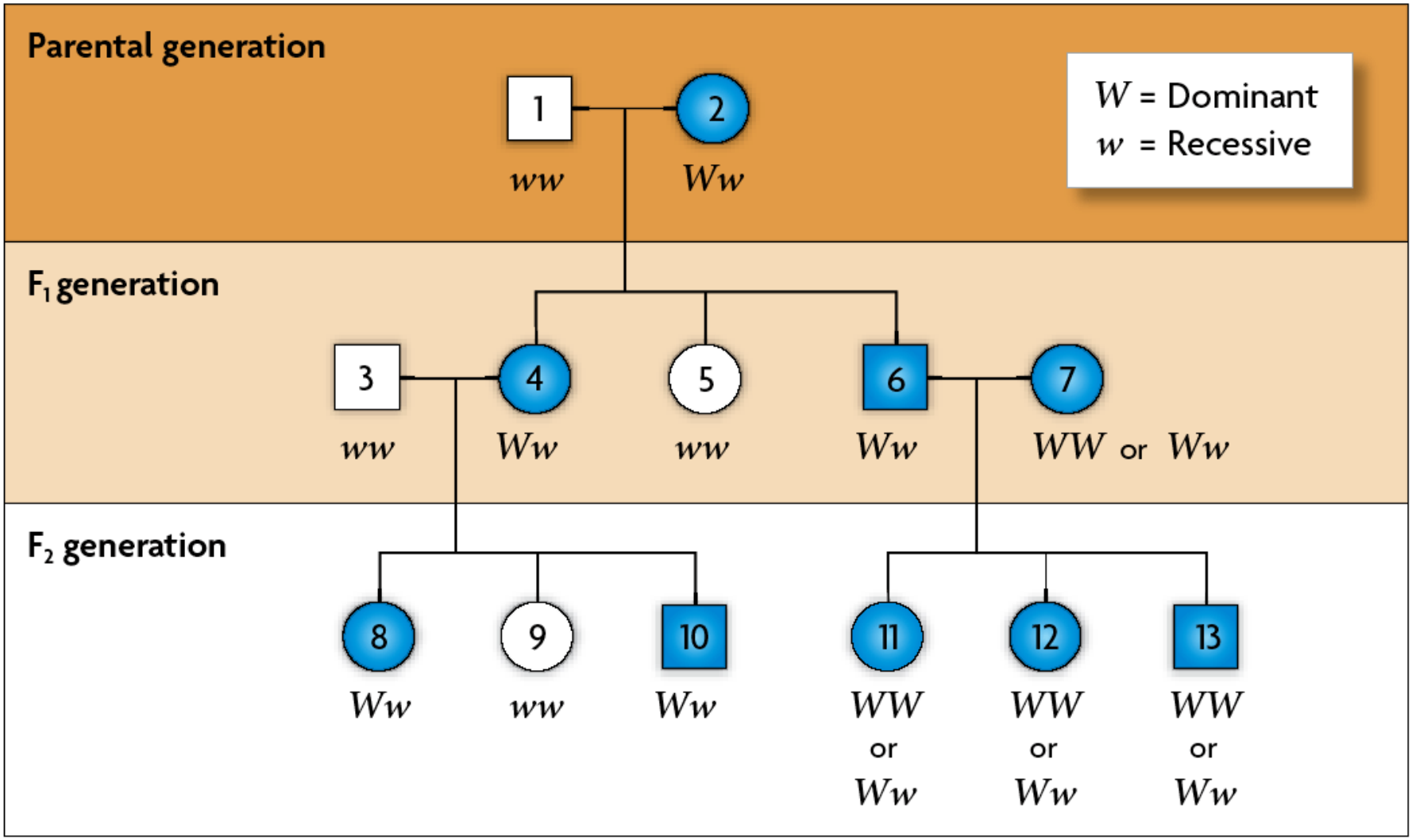
D. A pedigree is a chart for tracing genes in a family

1. Phenotypes are used to <u>infer</u> genotypes ona pedigree

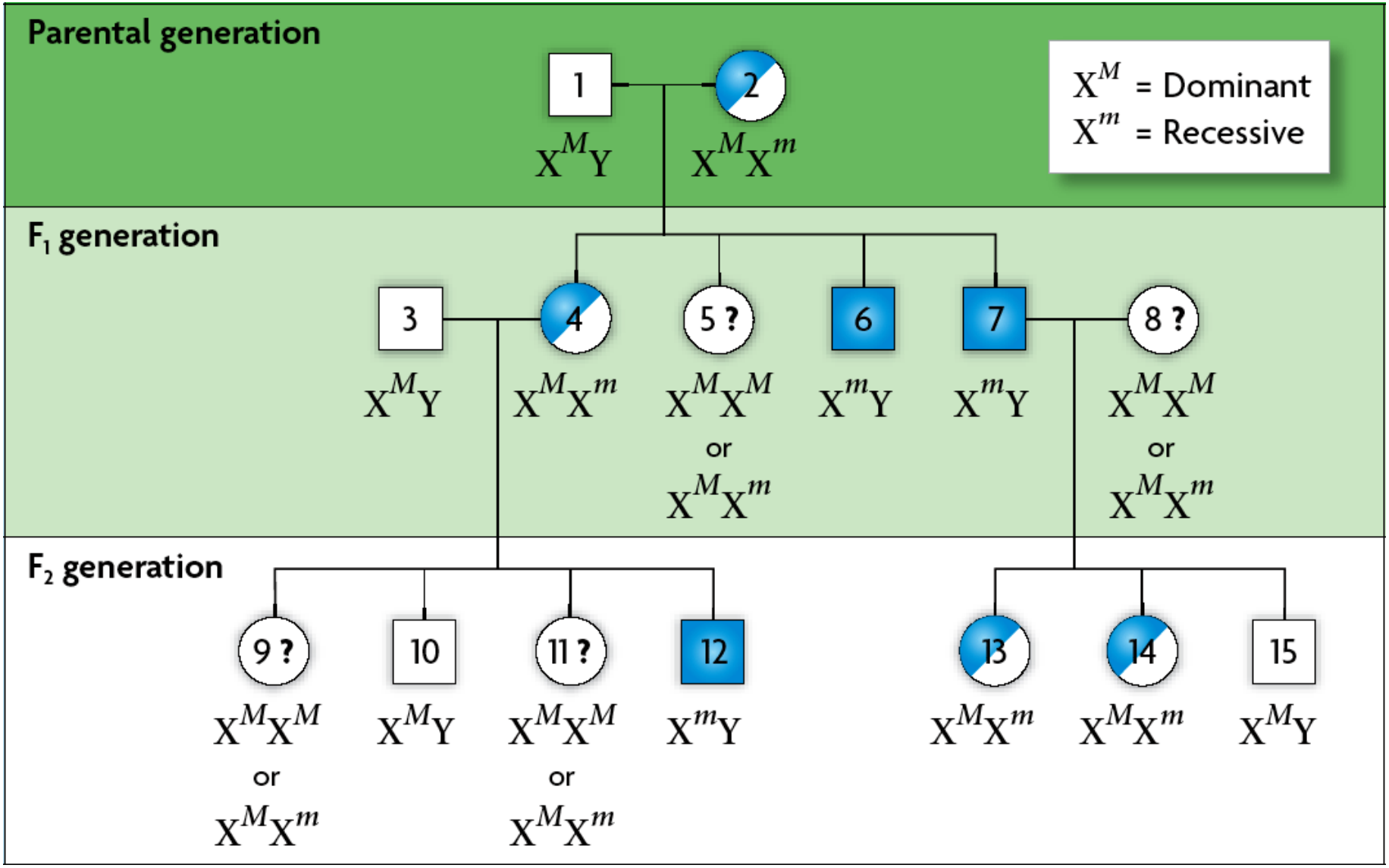
2. Autosomal genes show <u>different patterns</u> on a pedigree than sex-linked genes



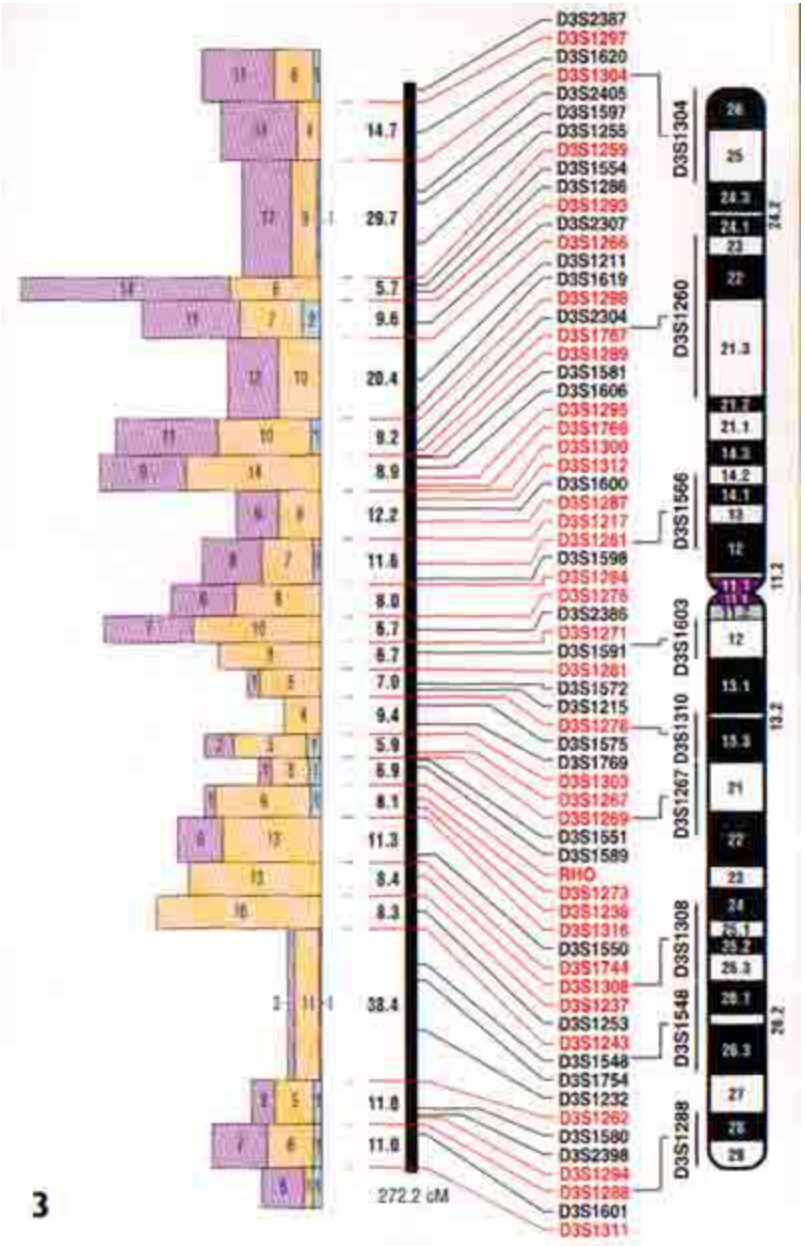
a. Autosomal genes



b. Sex-linked genes



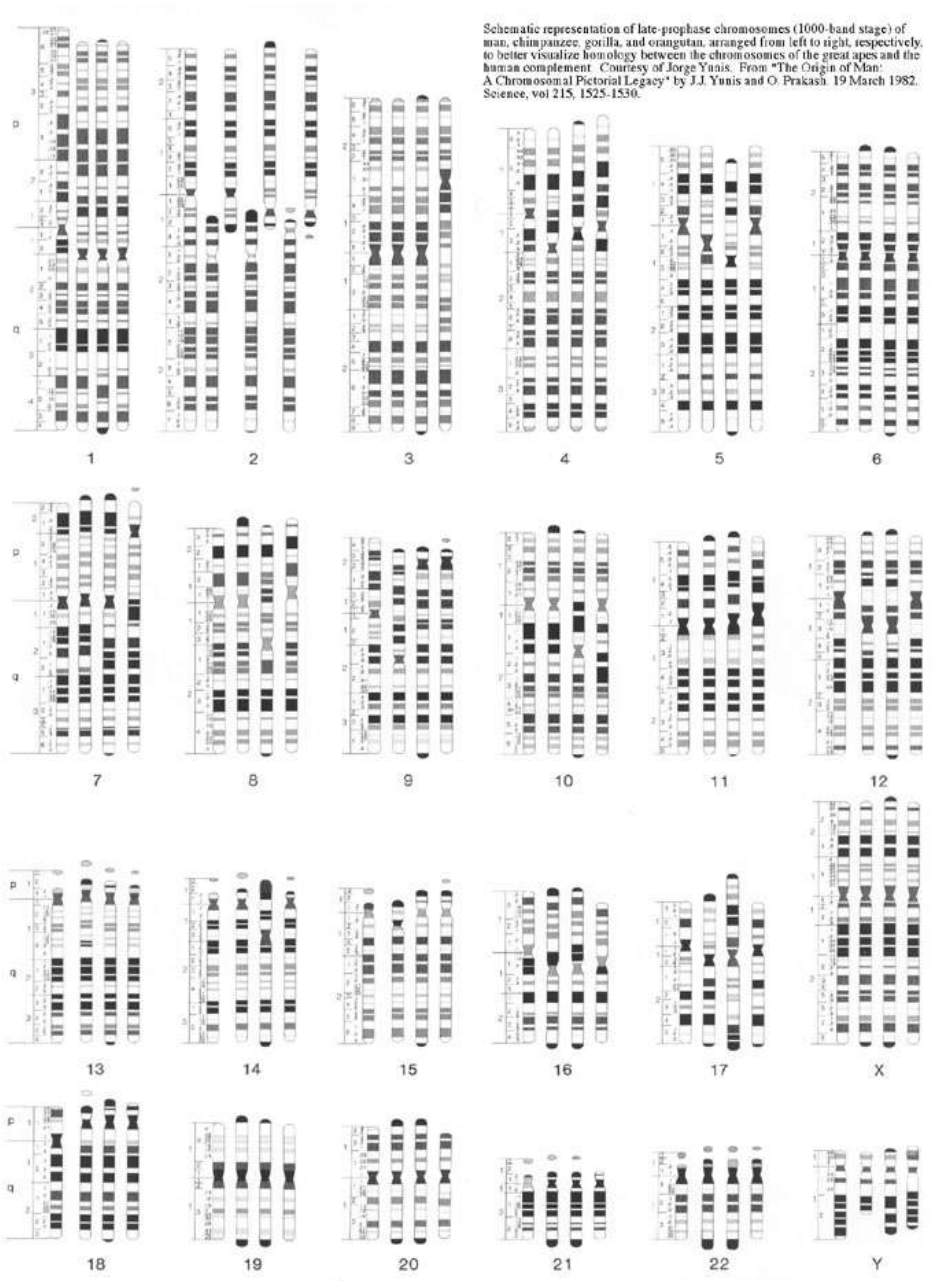
E. Several methods help map human



1. Human genome so large difficult to map

2. Several methods used

a. Pedigrees used for studying genetics in family



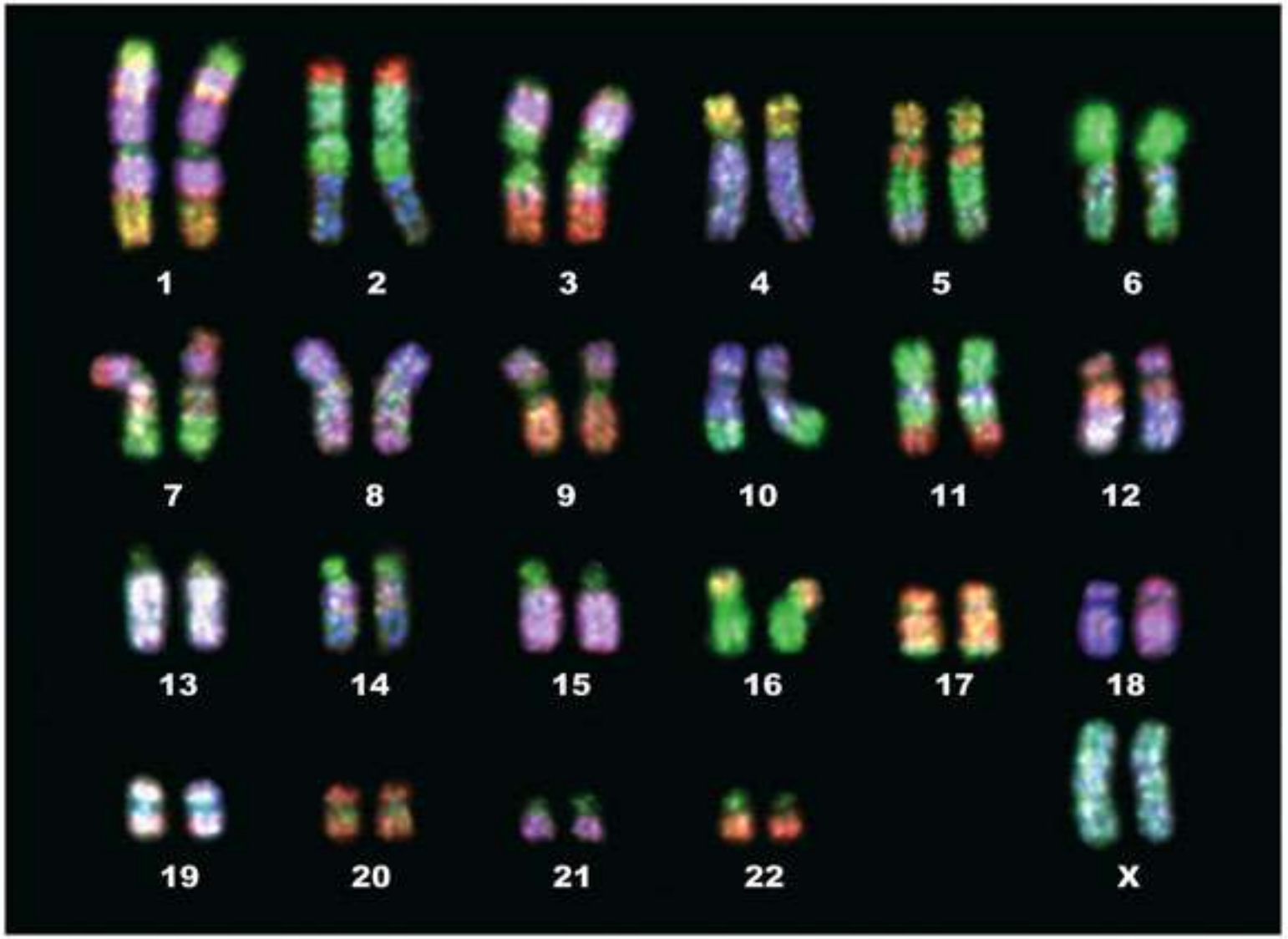
C. Human Genes-

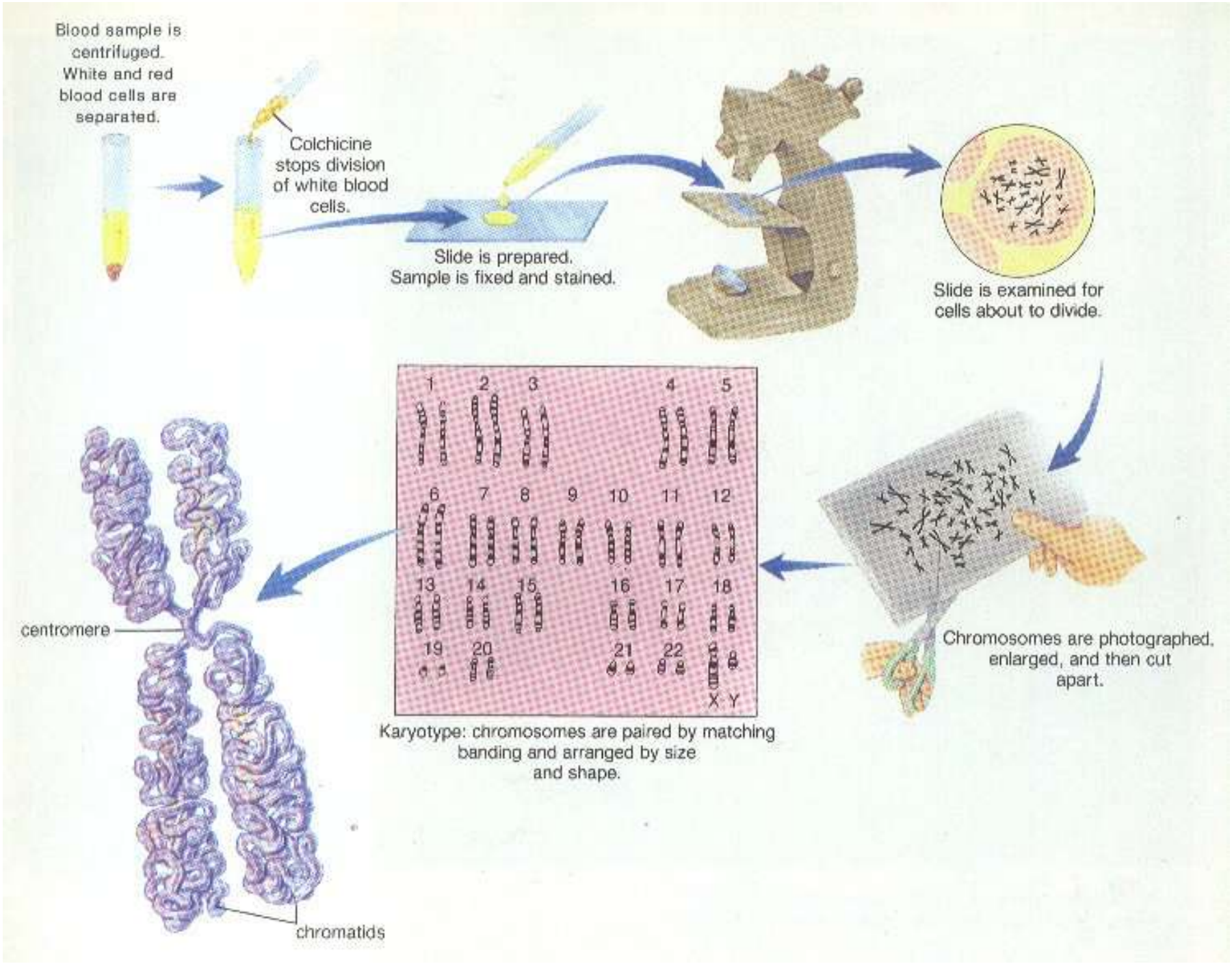
human genome contains 20-000 to 30,000 genes. Much lower than earlier estimates of 80,000 to 140,000

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**

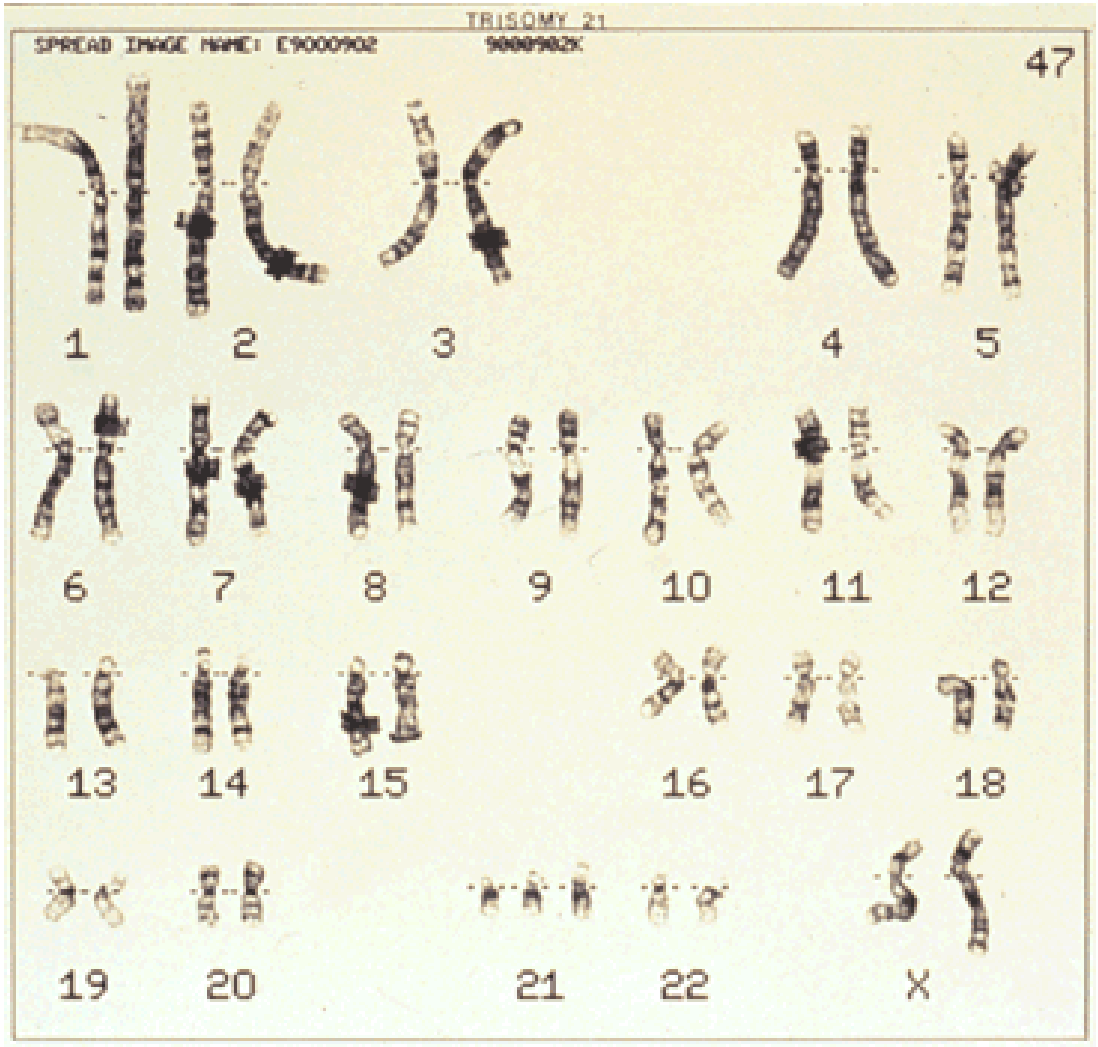
b. Karyotypes- picture of all chromosomes in a cell

1). S



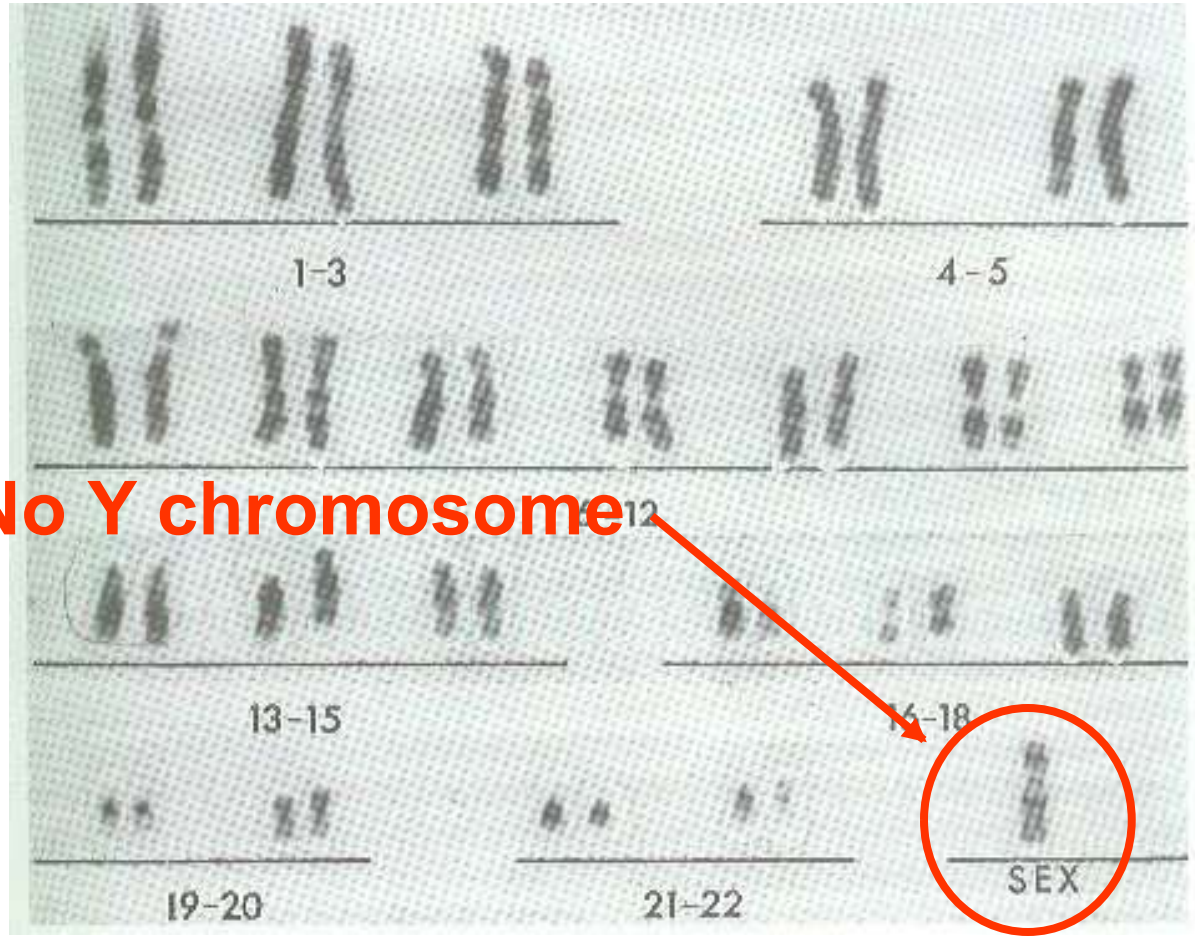

Blood sample is centrifuged. White and red blood cells are separated.
Colchicine stops division of white blood cells.
Slide is prepared. Sample is fixed and stained.
Slide is examined for cells about to divide.
Chromosomes are photographed, enlarged, and then cut apart.
1 2 3 4 5
6 7 8 9 10 11 12
13 14 15 16 17 18
19 20 21 22
X Y
Karyotype: chromosomes are paired by matching banding and arranged by size and shape.
centromere
chromatids

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

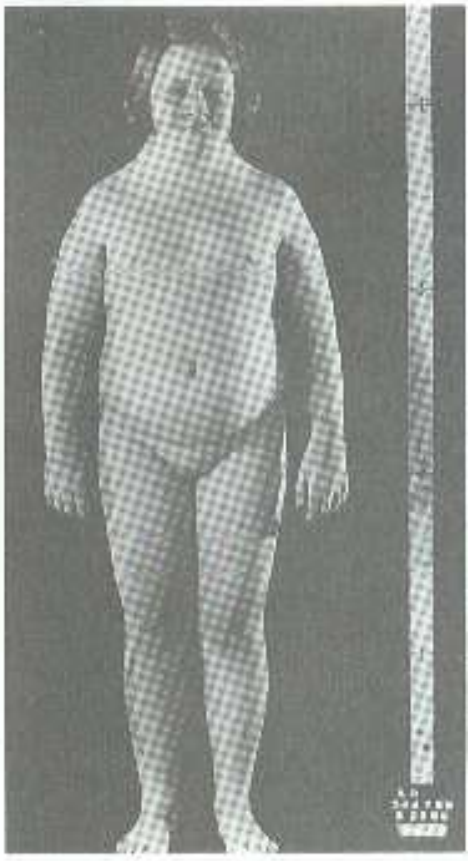


Sex Chromosome disorders

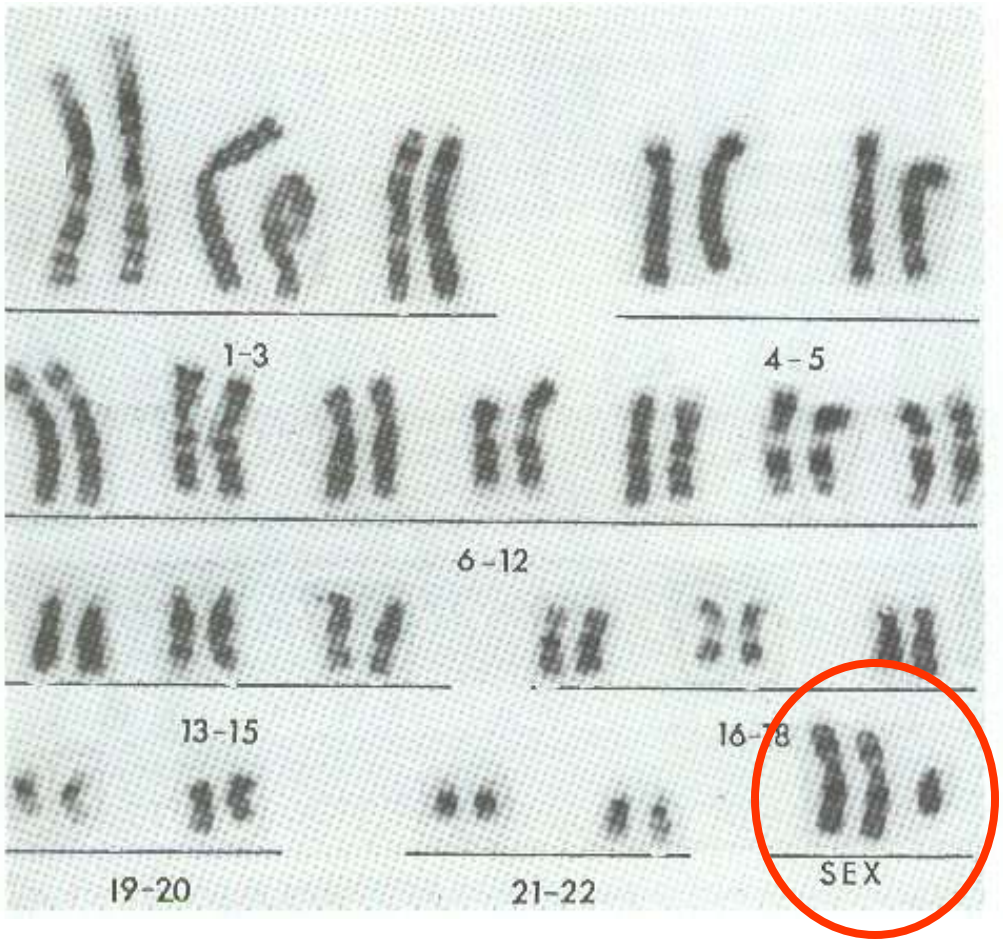
•In females, nondisjunction can lead to **Turner's Syndrome** (XO) sex organs fail to develop at puberty



No Y chromosome



In males, nondisjunction causes **Klinefelter's syndrome** (XXY) extra X interferes with meiosis and usually prevents individuals from reproducing



Review Quiz

Chapter 7
Extending Mendelian Genetics

How many chromosomes are shown in a normal human karyotype?

a.2

b.23

c.44

d.46

Review Quiz

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Review Quiz

Which of the following are shown in a karyotype?

a.homologous chromosomes

b.sex chromosomes

c.autosomes

d.all of the above

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Review Quiz

Which of the following can be observed in a karyotype?

a.a change in a DNA base

b.an extra chromosome

c.genes

d.alleles

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Review Quiz

In humans, a male has

a.one X chromosome only.

b.two X chromosomes.

c.one X chromosome and one Y chromosome.

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Human females produce egg cells that have

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c.one X or one Y chromosome.

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Review Quiz

What is the approximate probability that a human offspring will be female?

a.10%

b.25%

c.50%

d.75%

Review Quiz

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c.50%

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Review Quiz

What is the probability that a human sperm cell will carry an X chromosome?

a.0%

b.25%

c.50%

d.100%

Review Quiz

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Review Quiz

In a pedigree, a circle represents a(an)

a.male.

b.female.

c.child.

d.adult.

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Review Quiz

Which of the following is determined by multiple alleles?

a.Rh blood group

b.ABO blood group

c.Widows peak

d.Huntington's disease

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If a man with the genotype $I^A i$ and a woman with the genotype $I^B i$ produce an offspring, what might be the offspring's blood type?

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b.A, B, or O

c.A, B, AB, or O

d.AB only

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Review Quiz

Sex-linked genes are located on

a.the autosomes.

b.the X chromosome only.

c.the Y chromosome only.

d.both the X chromosome and Y chromosome.

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Review Quiz

Colorblindness is more common in males than in females because

a.fathers pass the allele for colorblindness to their sons only.

b.the allele for colorblindness is located on the Y chromosome.

c.the allele for colorblindness is recessive and located on the X chromosome.

d.males who are colorblind have two copies of the allele for colorblindness.

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Review Quiz

Which of the following statements is true?

a.Females cannot have hemophilia.

b.The father of a colorblind boy may be colorblind.

c.A sex-linked allele cannot be dominant.

d.The mother of a colorblind boy must be colorblind.

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Review Quiz

A cat that has only orange spots

a.has no Barr bodies.

b.must be a male.

c.must be a female.

d.may be a male or a female.

Review Quiz

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Review Quiz

The failure of chromosomes to separate during meiosis is called

a.nondisjunction.

b.X-chromosome inactivation.

c.Turner's syndrome.

d.Down syndrome.

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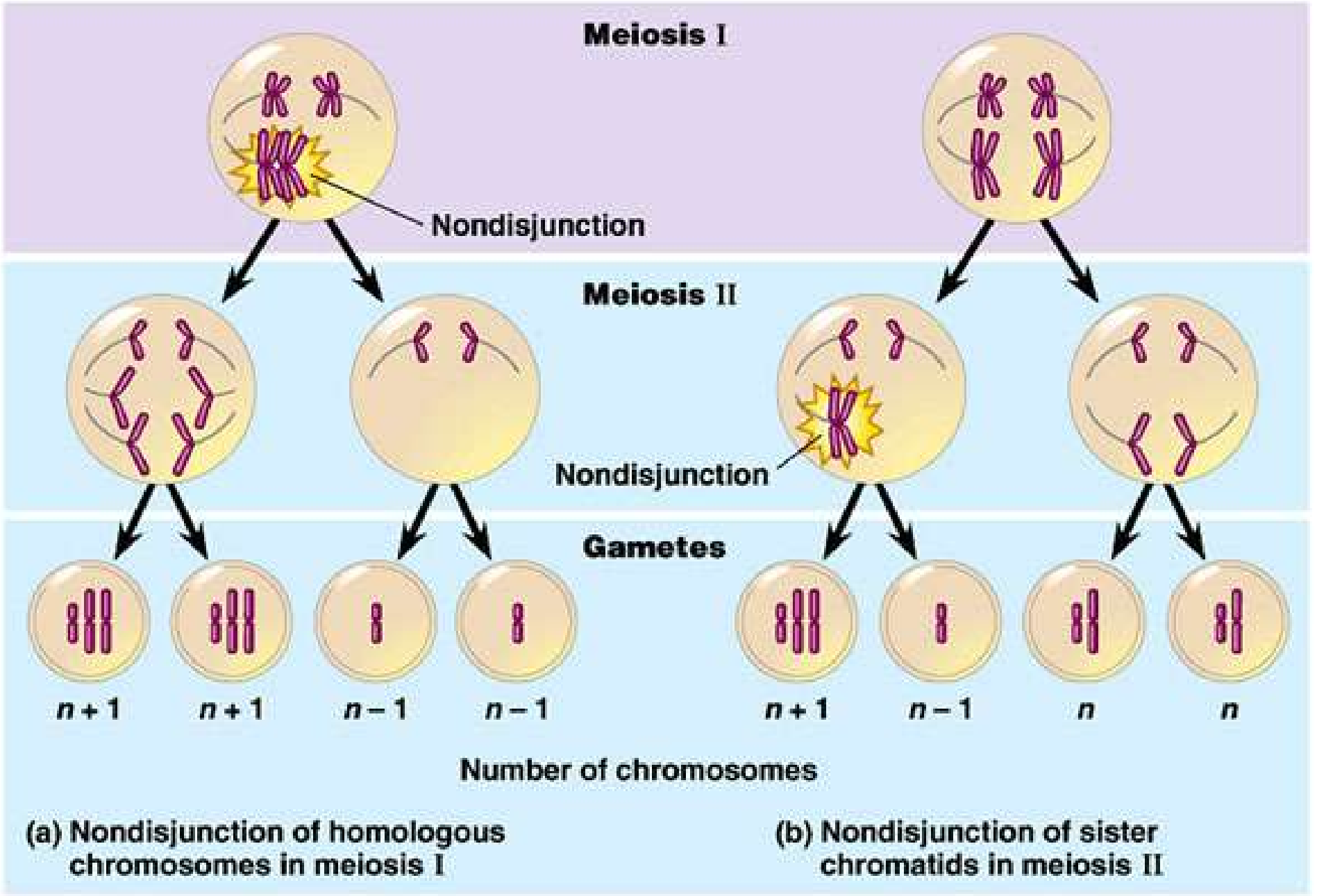

The male testes are responsible for producing sperm in which there are _____

a.44 autosomes and 2 sex chromosomes

b.22 autosomes and 2 sex chromosomes

c.44 autosomes and 1 sex chromosomes

d.22 autosomes and 1 sex chromosomes

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Review Quiz