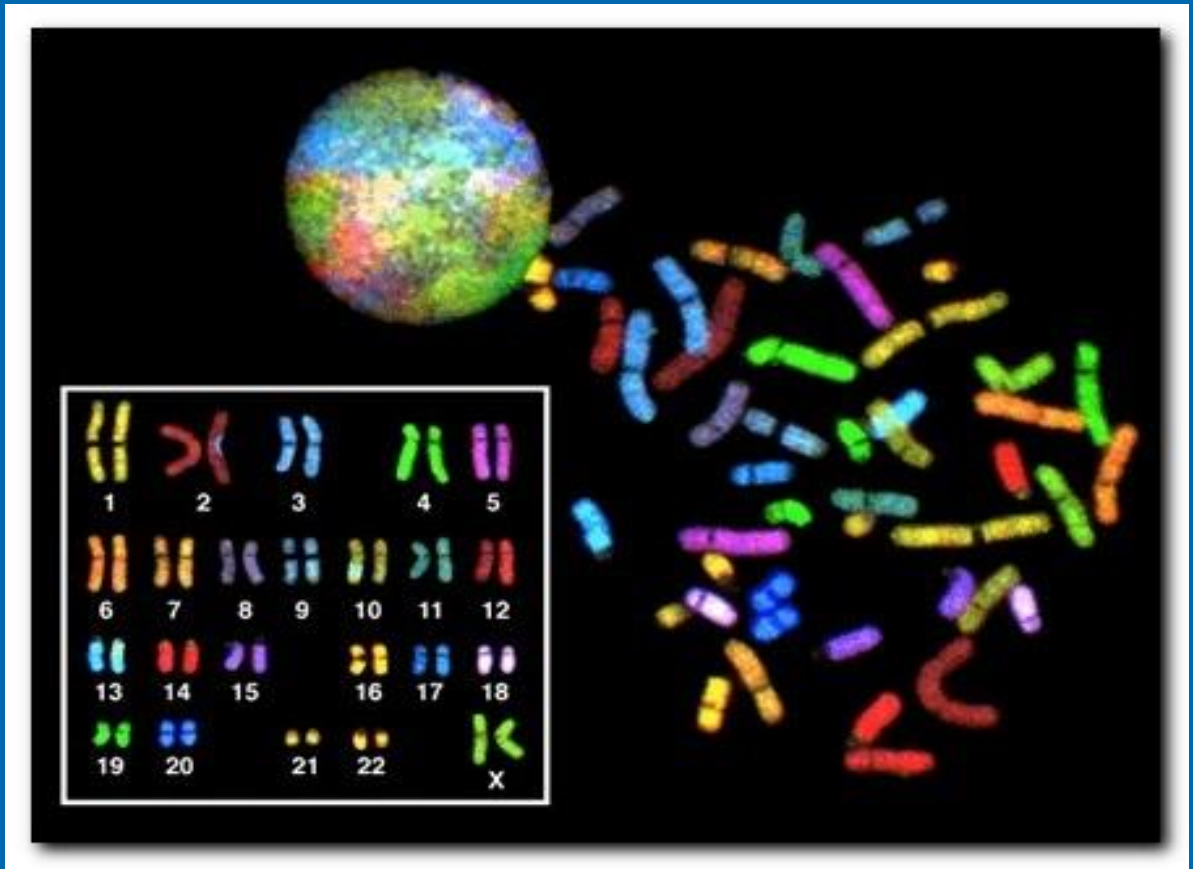
Go to the "Slide Show" shade above

Click on "Play from Beginning"

Chapter 15: Human Genetics



Spectral Karyotyping (SKY)

Preparing for Genetic Testing

1. Which four letters represent the nucleotides of the DNA?
2. In which phase of the cell cycle do the chromosomes form?
3. What protein does the DNA wrap itself around in the beginning stages of chromosome formation?
4. In which phase of mitosis are the chromosomes most easily seen?
5. Which letter represents the small arm of a chromosome?
6. Which letter represents the long arm of the chromosome?
7. How would you describe what a karyotype looks like?
8. What two types of chromosomes do you have?
9. Which chromosomes determine the gender of the person?
10. The chromosomes for Males are ___ and females are ___.

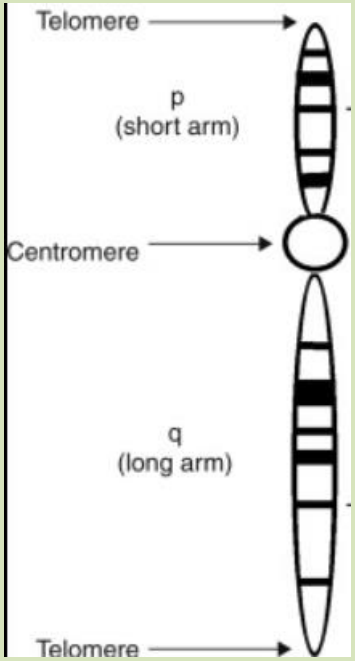
Preparing for Genetic Testing

WARM-UP

1. Which four letters represent the nucleotides of the DNA? A T G C
2. In which phase of the cell cycle do the chromosomes form?
   S phase of interphase
3. What protein does the DNA wrap itself around in the beginning stages of chromosome formation? histones
4. In which phase of mitosis are the chromosomes most easily seen?

During metaphase, the nucleus dissolves and the cell's chromosomes condense and move together, aligning in the center of the dividing cell. At this stage, the chromosomes are distinguishable when viewed through a microscope.

5. Which letter represents the small arm of a chromosome?
6. Which letter represents the long arm of the chromosome?



WARM-UP

Preparing for Genetic Testing

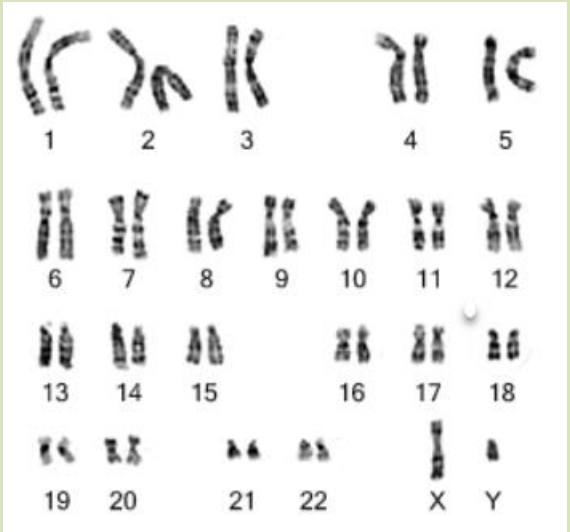
7. How would you describe what a karyotype looks like?

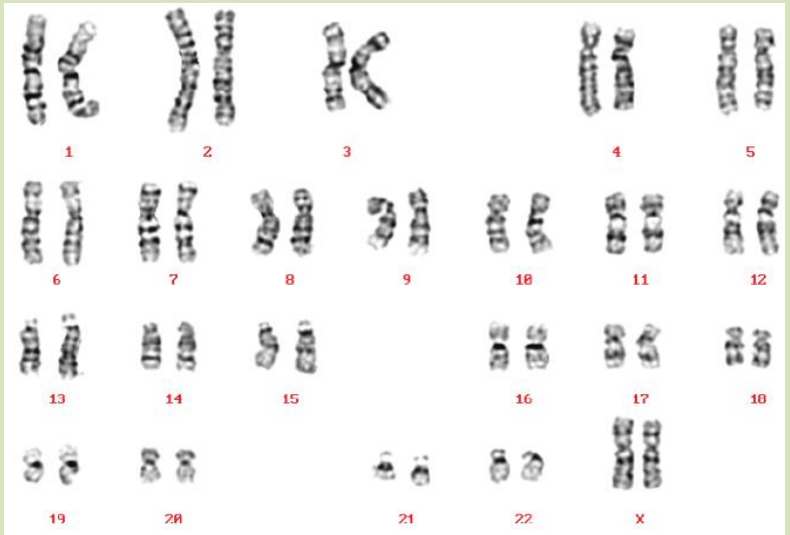
   Karyotyping is the process of pairing and ordering all the chromosomes of an organism (like a police line up), thus providing a genome-wide snapshot of an individual's chromosomes.

8. What two types of chromosomes do you have? **Autosomal & sex cells**

9. Which chromosomes determine the gender of the person? **sex cells (gametes)**

10. The chromosomes for Males are **Xy** and females are **XX**.





Lesson Objectives

By the end of this lesson, you should be able to:

- Understand and build pedigree charts.
- Describe technologies used for genetic diagnosis.
- Define and identify Karyotype mapping in terms of techniques and components.
- Understand and describe human patterns of inheritance (Autosomal Recessive, Autosomal Dominant, Sex-linked).
- Define and give examples of inherited human traits based on one gene, including heterozygous advantage.
- Define and give examples of congenital genetic diseases, including non-disjunction.

- **Science Practice: Genetics with a Smile**

Human Genetics

The study of inheritance as it occurs in human beings.

Human genetics encompasses a variety of overlapping fields including: classical genetics, cytogenetics, molecular genetics, biochemical genetics, genomics, population genetics, developmental genetics, clinical genetics, and genetic counseling.

Human Genetics

The development of human genetics has been limited because of

- ethics,
- limited offspring,
- lack of ability to select mates,
- ~18 years between generations,
- Environmental influences.

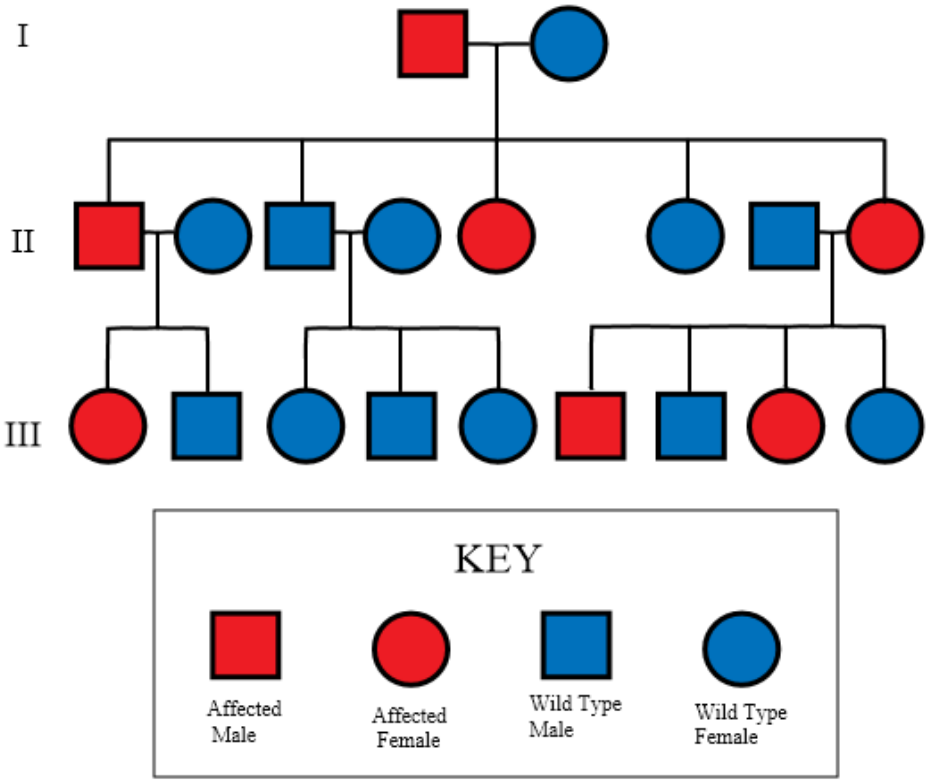
Genetic Traits in Humans can be tracked through Family Pedigrees

- The inheritance of human traits follows Mendel's laws.
- A **Pedigree**
  - shows the **inheritance of a trait** in a family through multiple generations
  - demonstrates **dominant** or **recessive** inheritance, and
  - can also be used to deduce genotypes of family members.

Pedigree Chart

Pedigree

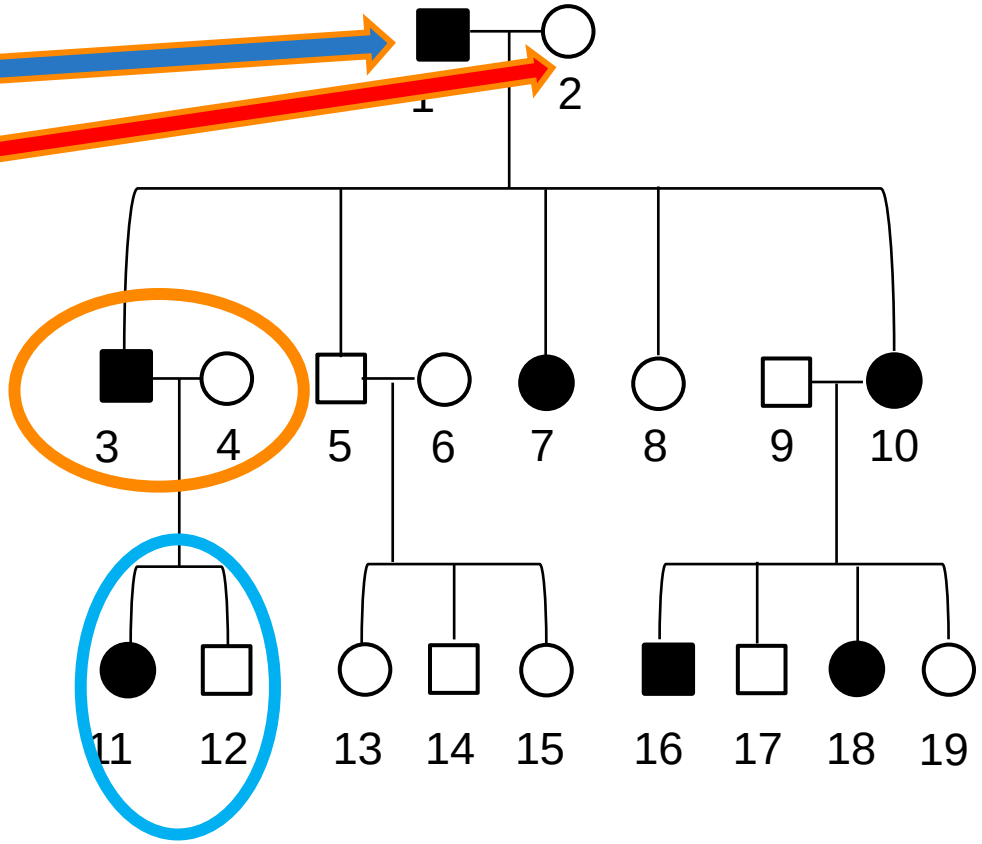
A graphic model showin inheritance patterns in families or breeding groups, including the genetic information for the organisms.



How to Read a Pedigree Chart

- Generation: row
- **Males**: squares
- **Females**: circles
- Horizontal lines between circle and square: **matings**
- Vertical lines: **offspring** of a mating

Different colors or shading are used to show individuals with and without a trait.

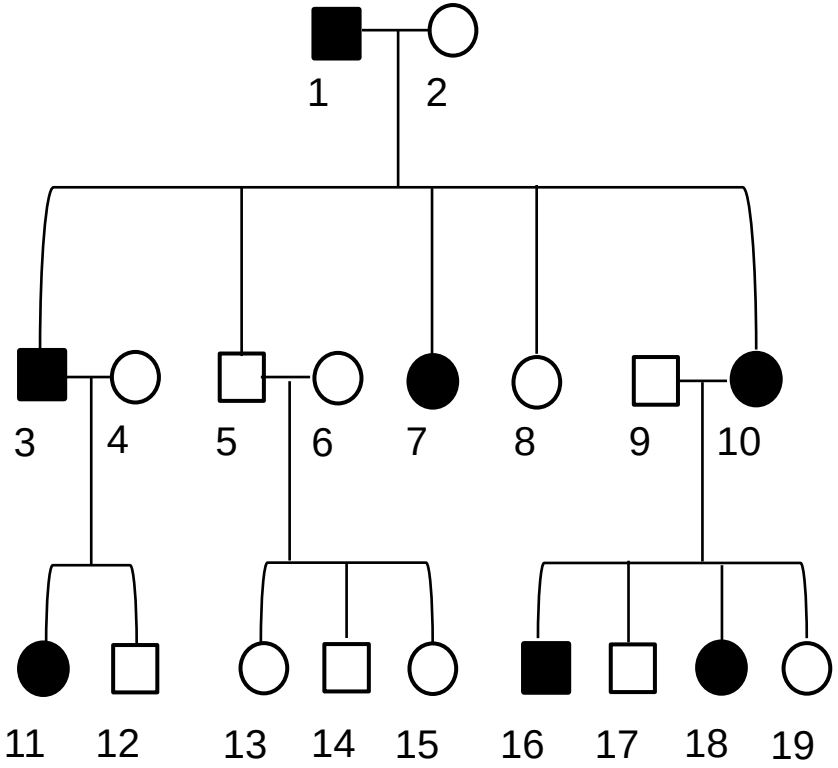


QUICK CHECK

Interpreting a Pedigree Chart

Which two individuals of generation III would have a higher probability of offspring with the trait of interest, shown in black?

- O 11 and 12
- O 13 and 14
- O 11 and 16
- O 14 and 18



The offspring of which individuals would be **least** closely related?

- O 3 and 7
- O 11 and 14
- O 17 and 18
- O 16 and 19

QUICK CHECK

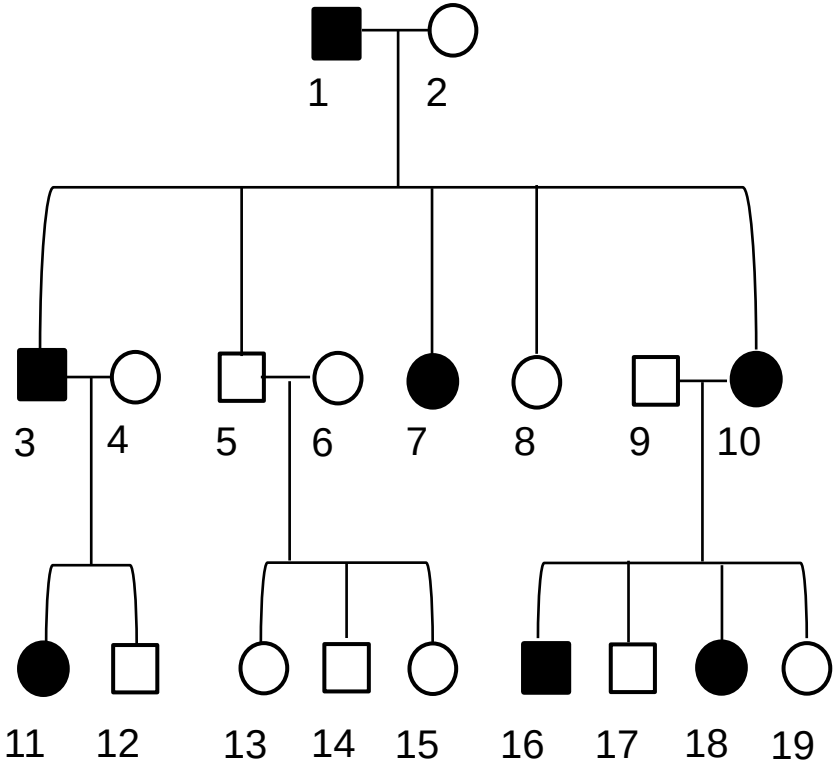
Interpreting a Pedigree Chart

Which two individuals of generation III would have a higher probability of offspring with the trait of interest, shown in black?

O 11 and 12
O 13 and 14
!!! 11 and 16
O 14 and 18

The offspring of which individuals would be **least** closely related?

O 3 and 7
!!! 11 and 14
O 17 and 18
O 16 and 19



Human Health Often Relates to Genetics

- There are major aspects related to personal health
  - **physical, emotional, social, spiritual, vocational, and intellectual**.
- In order to be considered "well“ or “healthy”, it is imperative that none of these areas be neglected.

Human Health Often Relates to Genetics

- For the scope of this course, we will confine our study to genetically related diseases.
- Techniques have been developed to detect genetic diseases or deficiencies before and after birth.

“Genetic Disorders”

http://dir.yahoo.com/Health/Diseases_and_Conditions/Genetic_Disorders/

Achondroplasia@
Achromatopsia@
Acid Maltase Deficiency@
Adrenoleukodystrophy@
Aicardi Syndrome@
Alpha-1 Antitrypsin Deficiency@
Androgen Insensitivity Syndrome@
Apert Syndrome@
Arrhythmogenic Right Ventricular Dysplasia@
Ataxia Telangiectasia@
Barth Syndrome@
Blue Rubber Bleb Nevus Syndrome@
Canavan Disease@
Cri Du Chat Syndrome@

“Genetic Disorders”

http://dir.yahoo.com/Health/Diseases_and_Conditions/Genetic_Disorders/

Cystic Fibrosis@
Dercum's Disease@
Ectodermal Dysplasia@
Fanconi Anemia@
Fibrodysplasia Ossificans Progressiva@
Fragile X Syndrome@
Galactosemia@
Gaucher Disease@
Hemochromatosis@
Hemophilia@
Huntington's Disease@
Hurler Syndrome@
Hypophosphatasia@
Klinefelter Syndrome@

“Genetic Disorders”

http://dir.yahoo.com/Health/Diseases_and_Conditions/Genetic_Disorders/

Krabbes Disease@
Langer-Giedion Syndrome@
Leukodystrophy@
Long QT Syndrome@
Marfan Syndrome@
Moebius Syndrome@
Mucopolysaccharidosis (MPS)@
Nail Patella Syndrome@
Nephrogenic Diabetes Insipidus@
Neurofibromatosis@
Niemann-Pick Disease@
Osteogenesis Imperfecta@
Porphyria@
Prader-Willi Syndrome@

“Genetic Disorders”

http://dir.yahoo.com/Health/Diseases_and_Conditions/Genetic_Disorders/

Progeria@
Proteus Syndrome@
Retinoblastoma@
Rett Syndrome@
Rubinstein-Taybi Syndrome@
Sanfilippo Syndrome@
Shwachman Syndrome@
Sickle Cell Disease@
Smith-Magenis Syndrome@
Stickler Syndrome@
Tay-Sachs@
Thrombocytopenia Absent Radius (TAR) Syndrome@
Treacher Collins Syndrome@
Trisomy@

“Genetic Disorders”

http://dir.yahoo.com/Health/Diseases_and_Conditions/Genetic_Disorders/

Tuberous Sclerosis@
Turner's Syndrome@
Urea Cycle Disorder@
von Hippel-Lindau Disease@
Waardenburg Syndrome@
Williams Syndrome@
Wilson's Disease@

How do you think a genetic disorder is discovered?

- Abnormalities can be observed
- Techniques help diagnosis

Technologies provide Insight into one's Genetic Legacy & Genetically Related Diseases

- **BLOOD TESTS** on the mother at 15–20 weeks of pregnancy can help identify fetuses at risk for certain birth defects.

Technologies provide Insight into one's Genetic Legacy & Genetically Related Diseases

- **Fetal Imaging** enables a physician to examine a fetus directly for anatomical deformities.
  - The most common procedure is **ULTRASOUND Imaging**, using sound waves to produce a picture of the fetus.

Technologies provide Insight into one's Genetic Legacy & Genetically Related Diseases

- **Newborn SCREENING** can detect diseases that can be prevented by special care and precautions.

A few drops of blood from a heel stick allows hospitals to screen newborns for genetic conditions.

Technologies provide Insight into one's Genetic Legacy & Genetically Related Diseases

- New technologies raise ethical considerations that include
  - the confidentiality and potential use of results of genetic testing,
  - time and financial costs, and
  - the determination of what, if anything, should be done as a result of the testing.

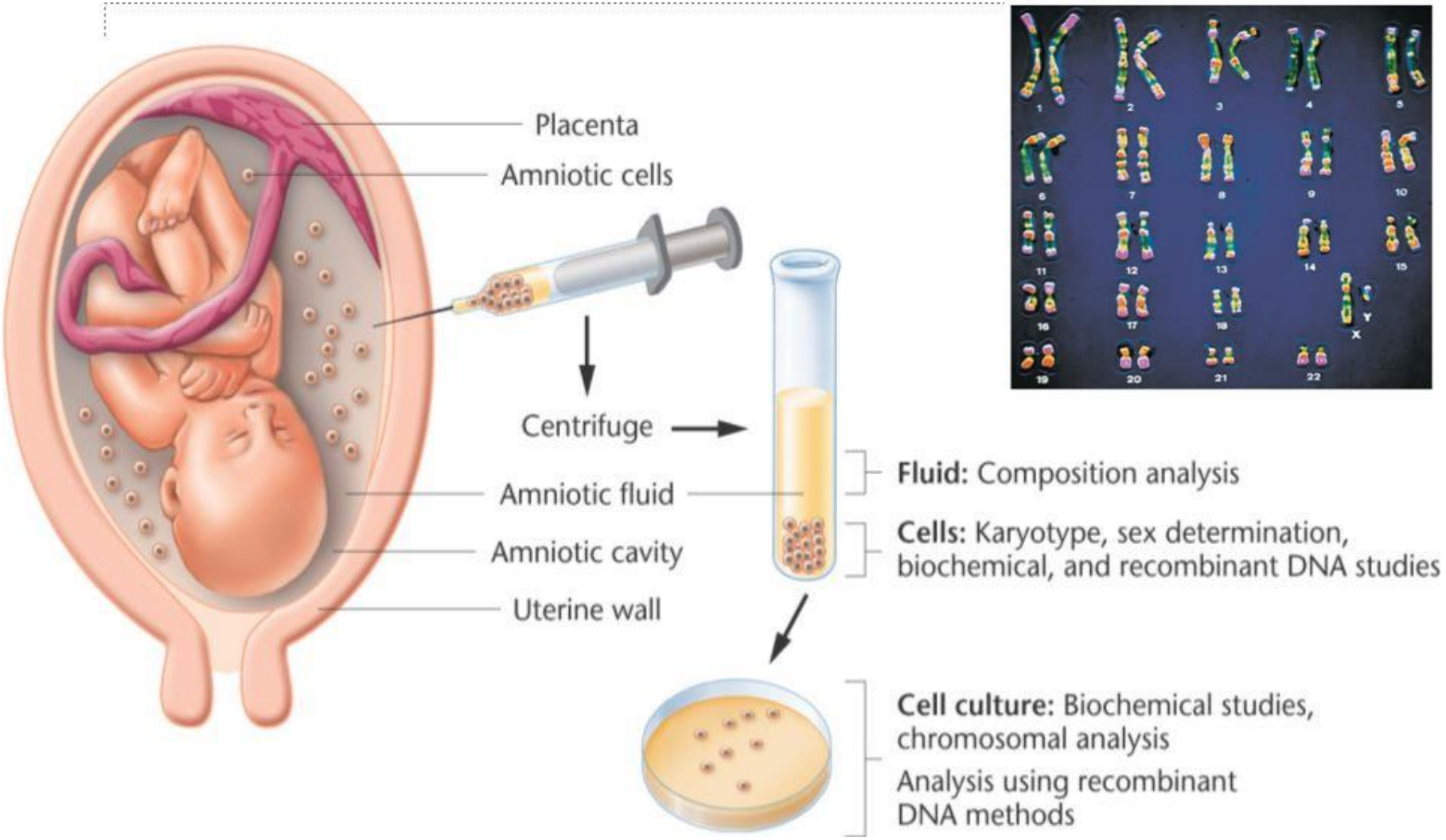
Technologies can provide Insight into one's Genetic Legacy

- Modern technologies offer ways to obtain genetic information
  - **before conception**
  - **during pregnancy**
  - **after birth**
- **Genetic Testing** can identify prospective parents who are heterozygous carriers for certain diseases.

Technologies can provide Insight into one's Genetic Legacy

- Certain technologies can be used for detecting genetic conditions in a fetus:
  - **Amniocentesis** extracts samples of **amniotic fluid containing fetal cells** and permits:
    - karyotyping to detect chromosomal abnormalities such as Down syndrome.
    - biochemical tests on cultured fetal cells to detect other conditions.

Technologies can provide Insight into one's Genetic Legacy



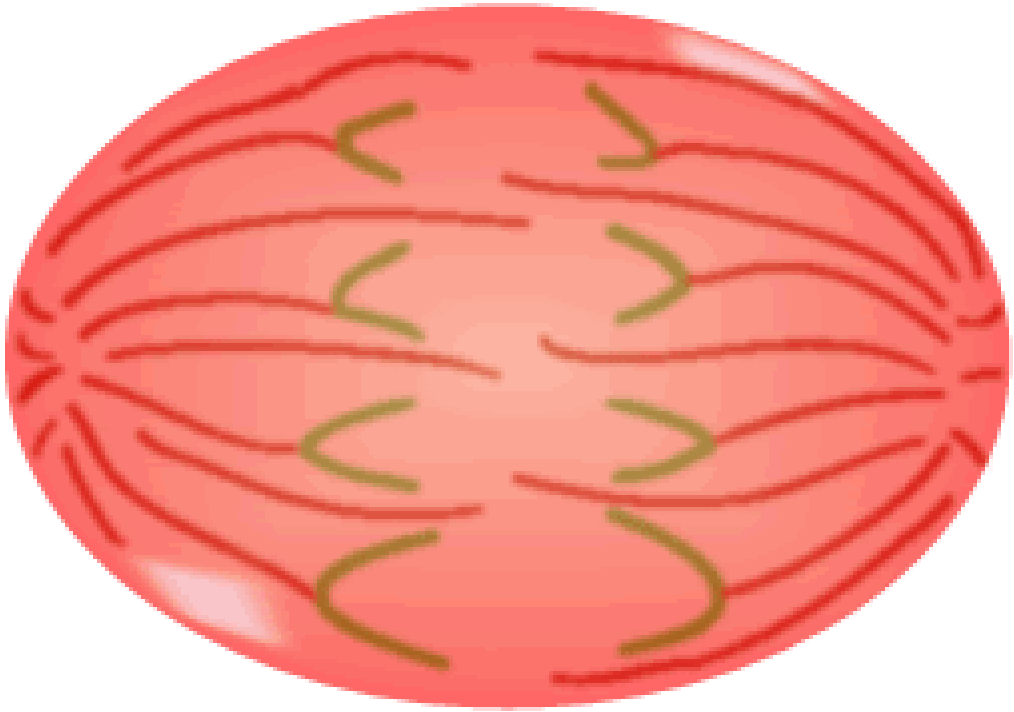
Human Genetic Patterns of Inheritance

Different patterns of inheritance are observed depending on the condition:

- **Autosomal Recessive Inheritance** (Sickle Cell, Phenylketonuria, Cystic Fibrosis)
- **Autosomal Dominant Inheritance** (Huntington's, Achondroplasia, Aneuploidy)
- **Sex-linked inheritance** (e.g. hemophilia, pattern baldness)

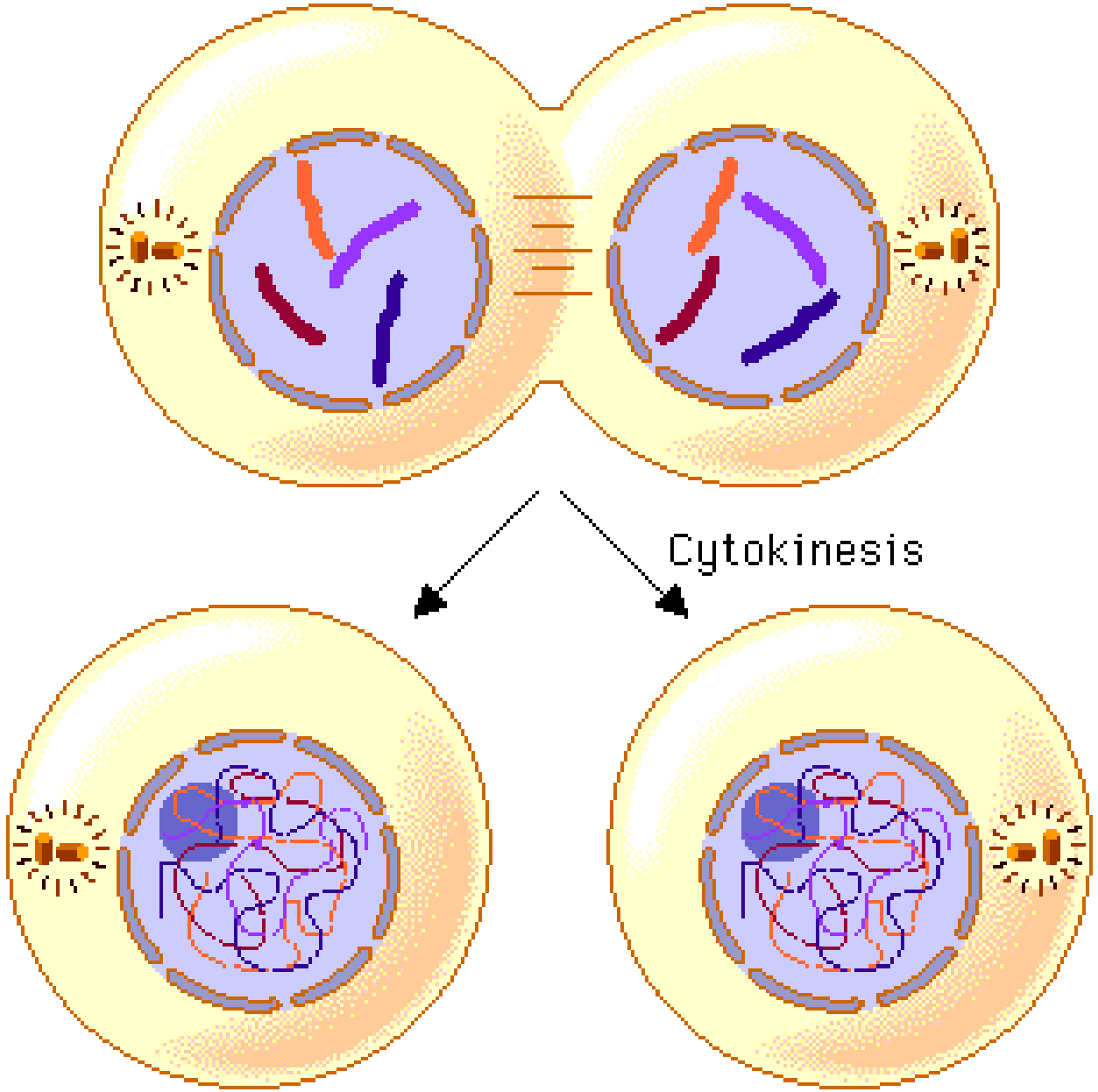
How do these patterns get diagnosed or discovered?

Recall Anaphase



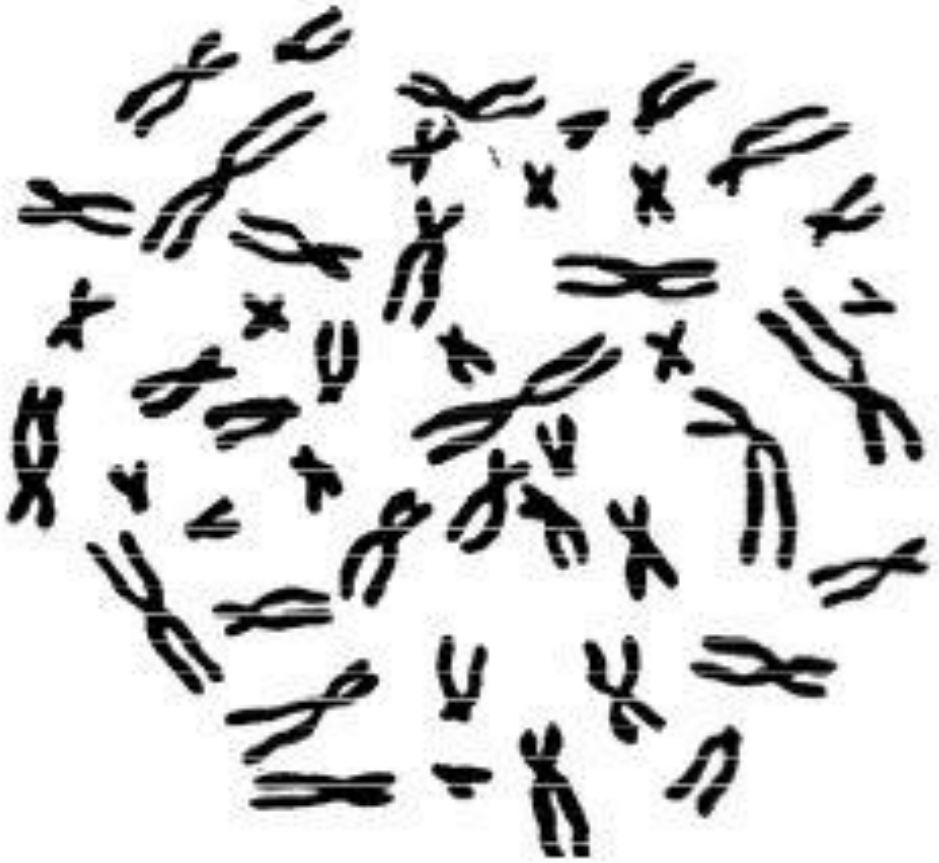
The duplicate chromosomes are pulled apart at the **centromere** just prior to **cytokinesis.**

What happens during cytokinesis?

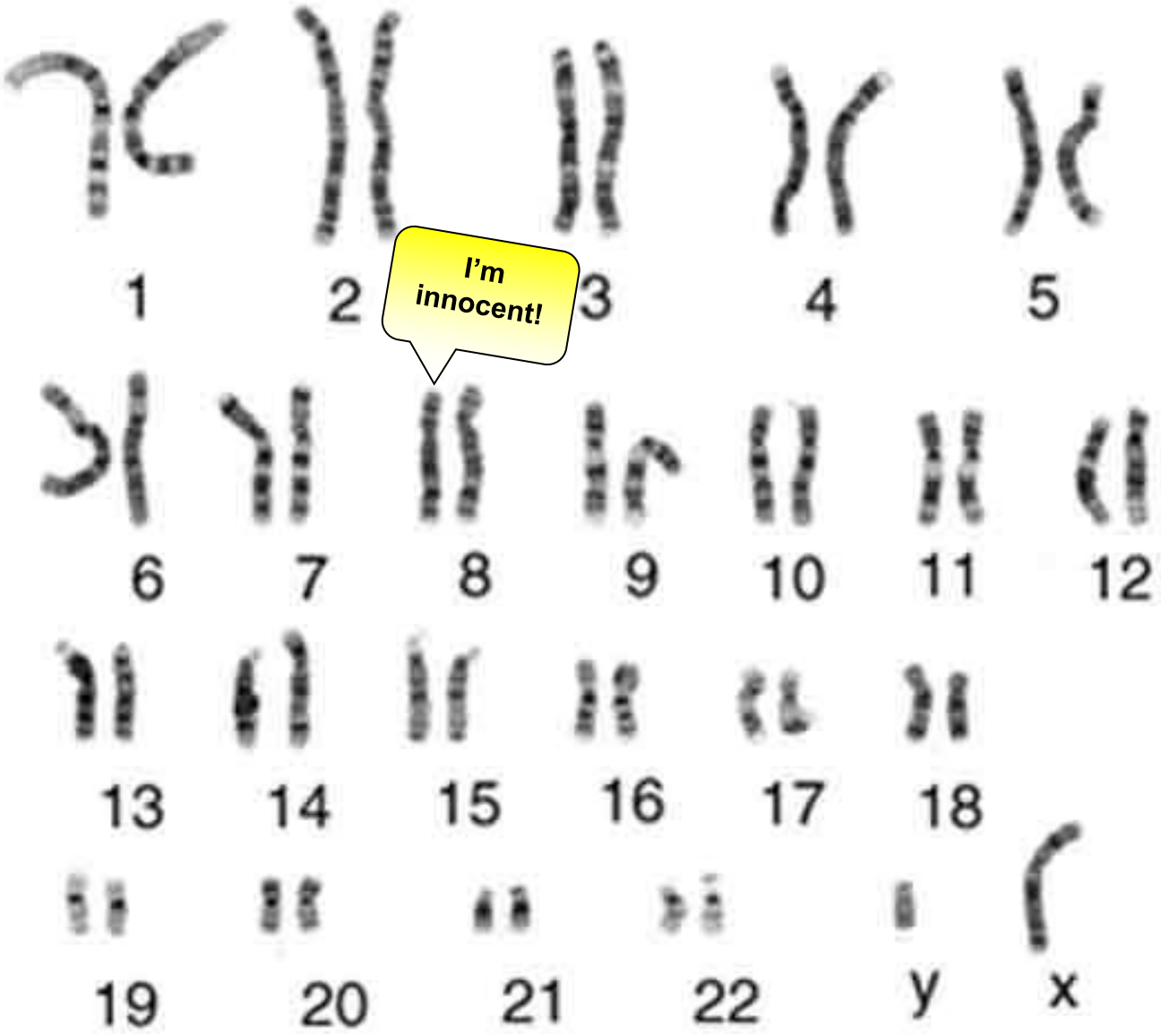


The cytoplasm pinches together to form the cell membrane of two distinct cells.

Prior to anaphase the chromosomes are dyed and removed for analysis.

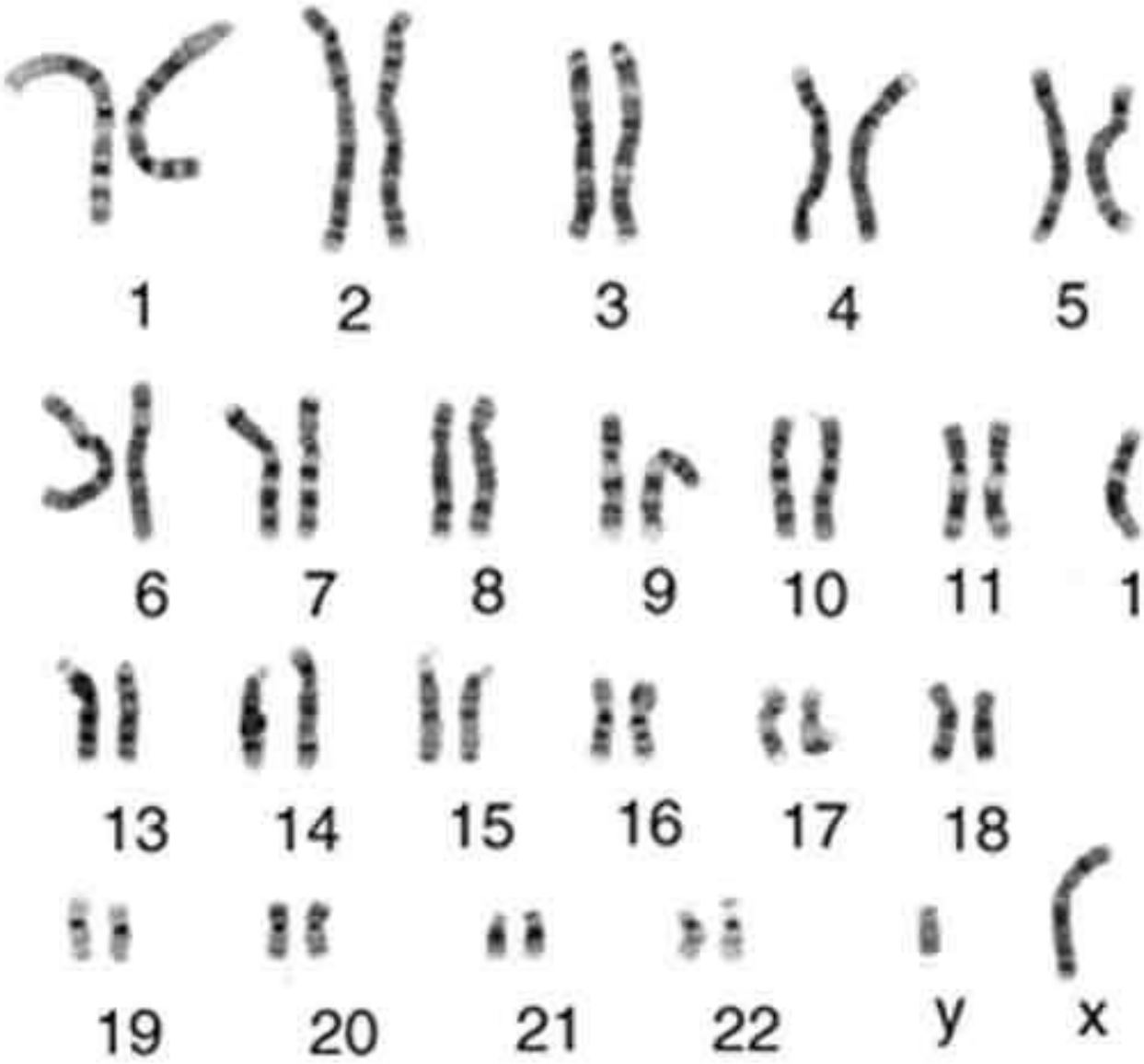


A Prepared Human Karyotype.



In this arrangement, the chromosomes are organized like a police line up.

A Prepared Human Karyotype.



Where are the 46 chromosomes?

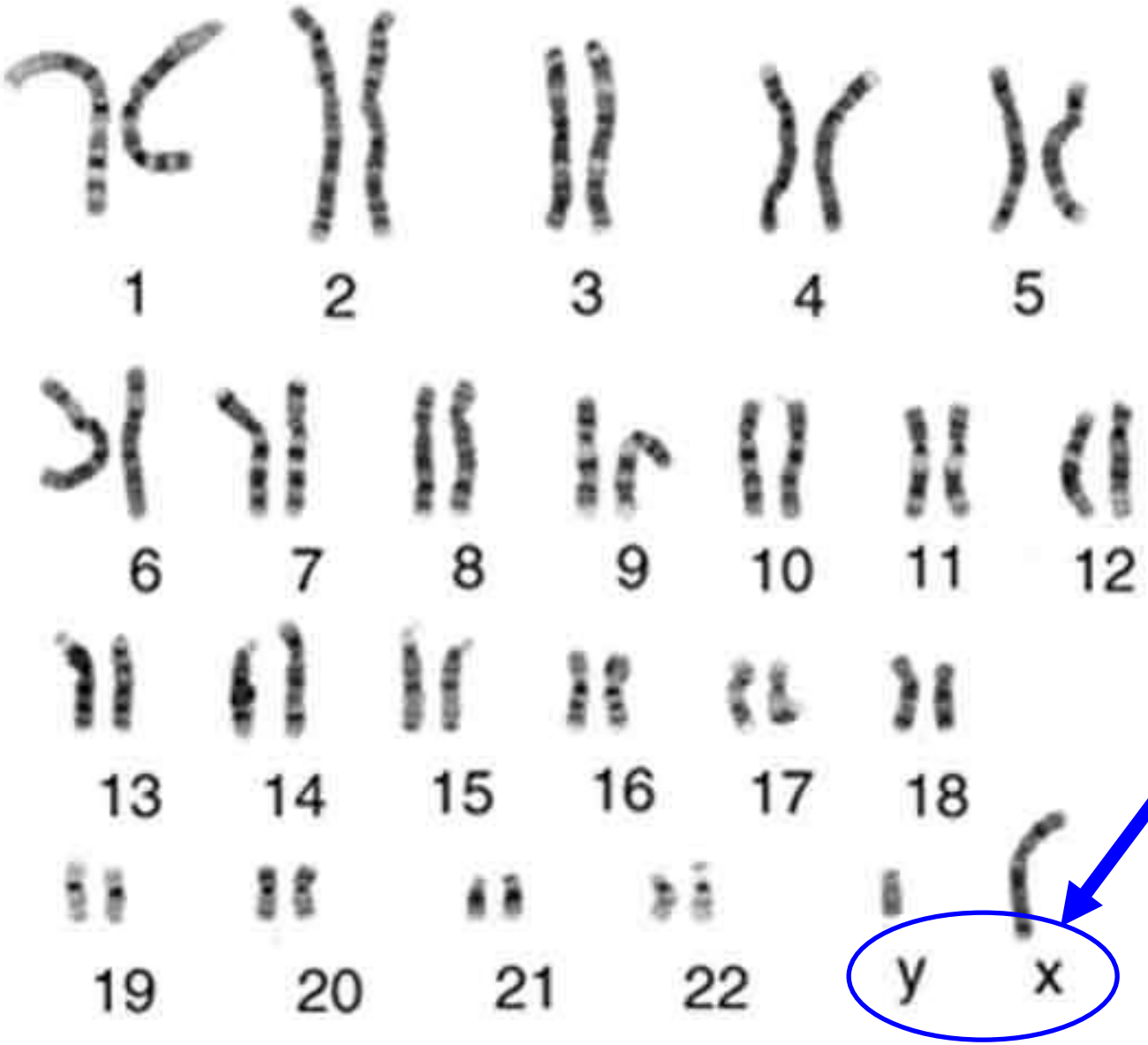
22 pairs of **autosomes**

and

2 sex chromosomes

(**x** and **y**)

A Prepared Human Karyotype.



The sex chromosomes determine the gender.

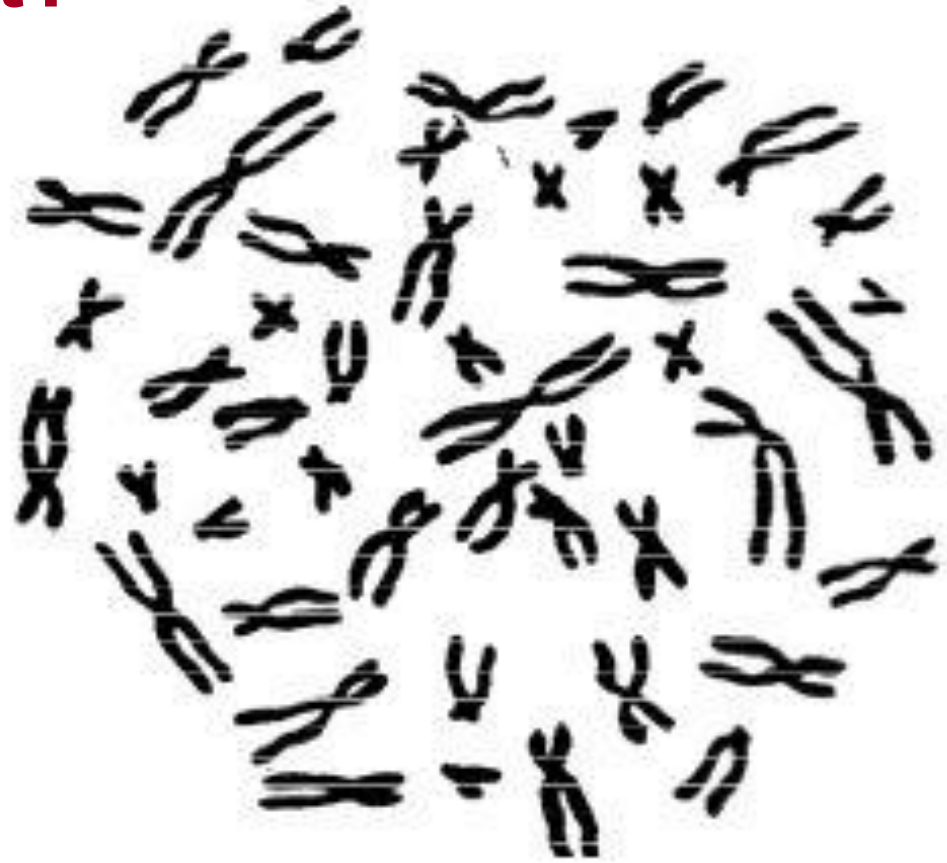
This is the karyotype of a male human.

Males (XY)

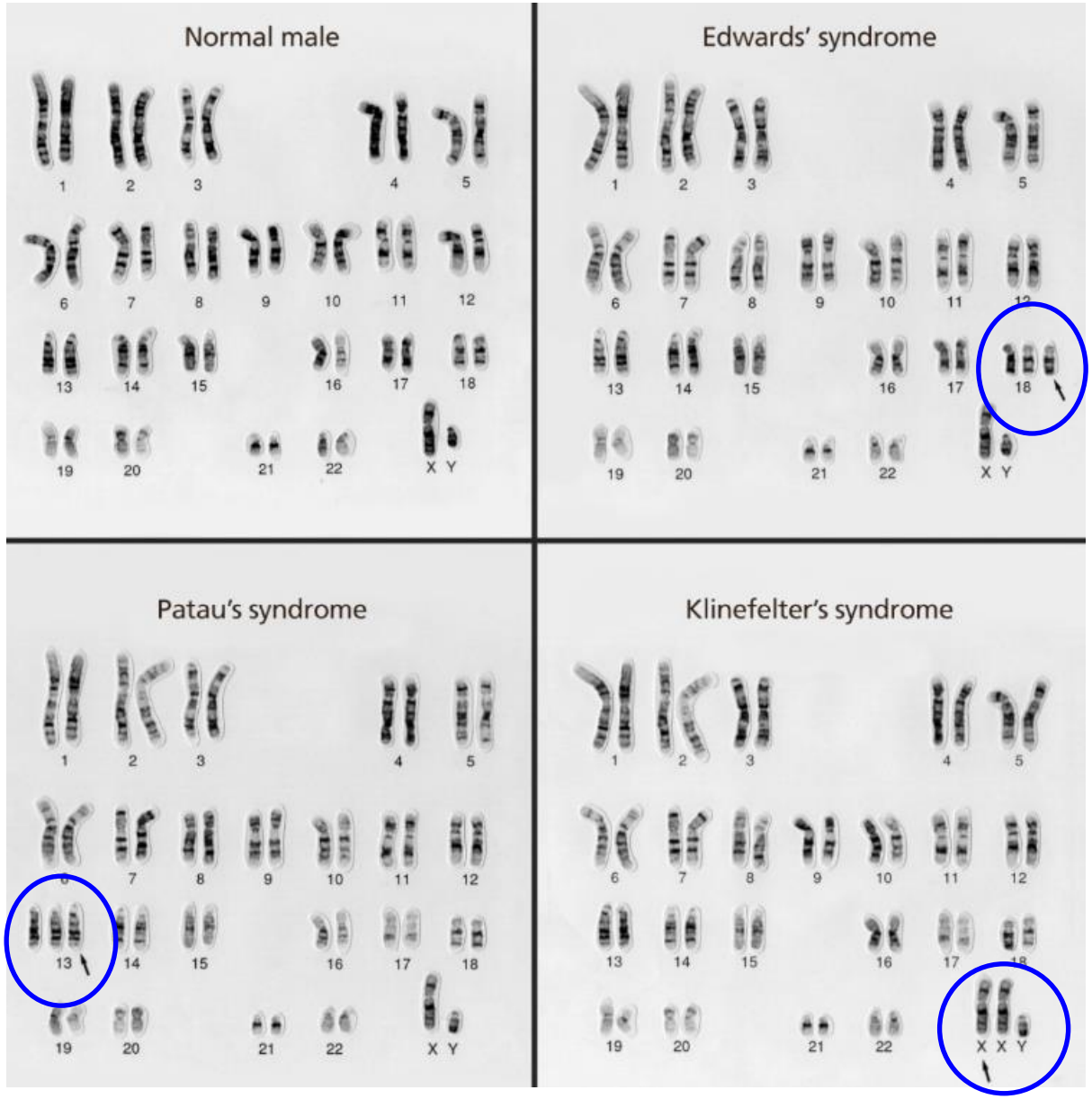
Females (XX)

Can you identify which of the chromosomes are autosomes and which are the sex chromosomes

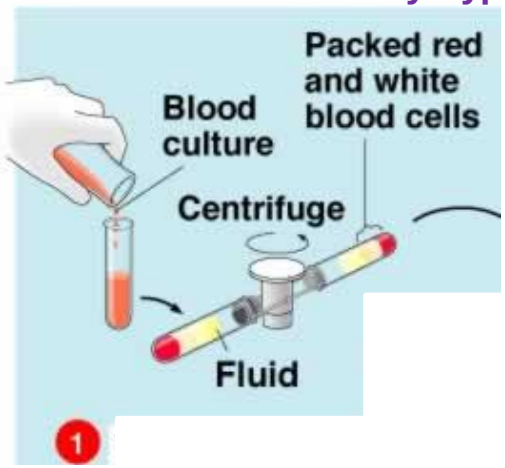
…by sight?



A karyotype is an image that maps the chromosomes and allows the detection of abnormalities.



Former Method of Karyotyping



1

The blood culture is centrifuged
(spun at a high speed)
to separate the blood cells from the culture fluid (blood plasma).

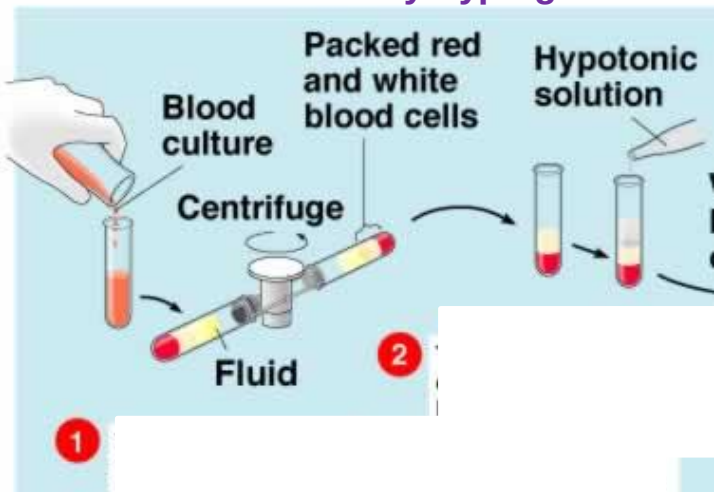
2

The fluid is discarded and a hypotonic solution is mixed with the cells.

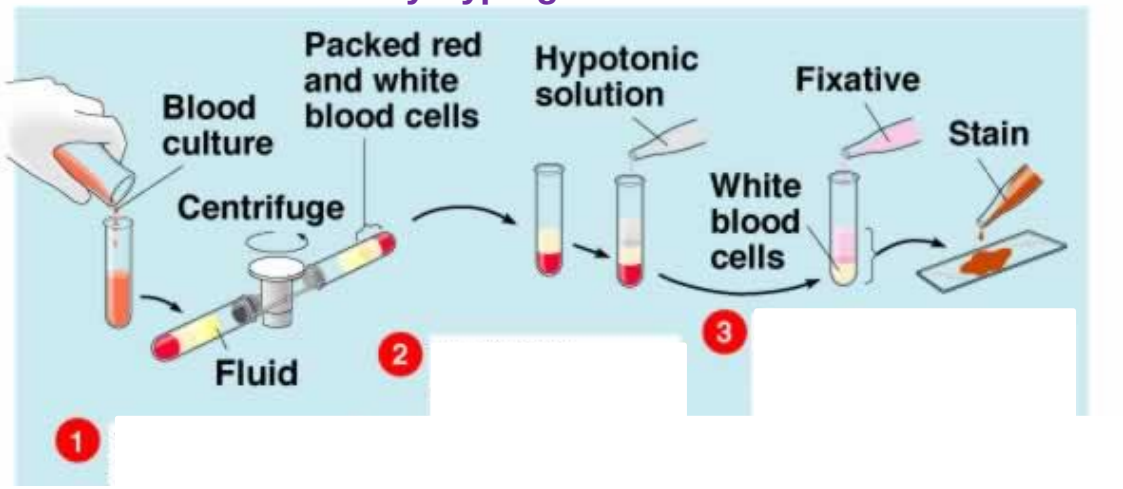
This makes the red blood cells (RBC's) swell and burst.

The white blood cells (WBC's) swell but do not burst, and their chromosomes spread out.

Former Method of Karyotyping



Former Method of Karyotyping



3

Another centrifugation step separates the swollen WBC's.

The fluid containing the remnants of the RBC's is poured off.

A fixative (a preservative) is mixed with the WBC's.

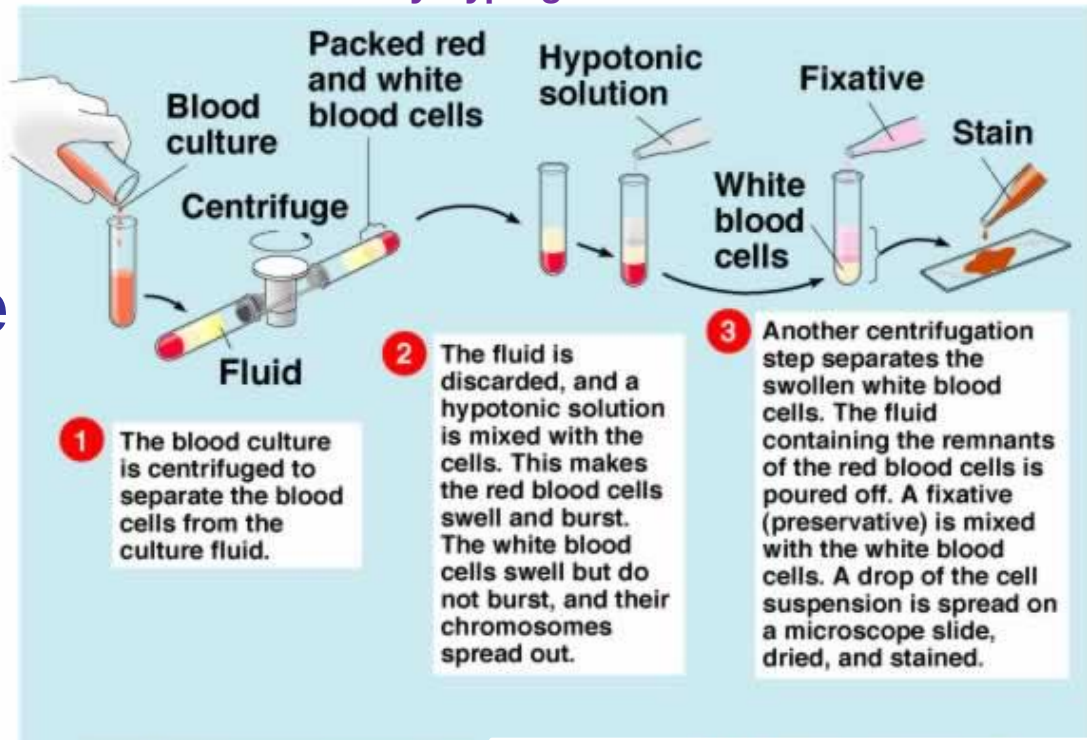
A drop of the cell suspension is spread on a microscope slide, dried and stained.

4

The slide with the WBC's DNA is viewed with a microscope,

and the images of the metaphase chromosomes are sorted by size and shape on a computer.

Former Method of Karyotyping



4 The slide is viewed with a microscope, and images of the metaphase chromosomes are sorted by size and shape on a computer.

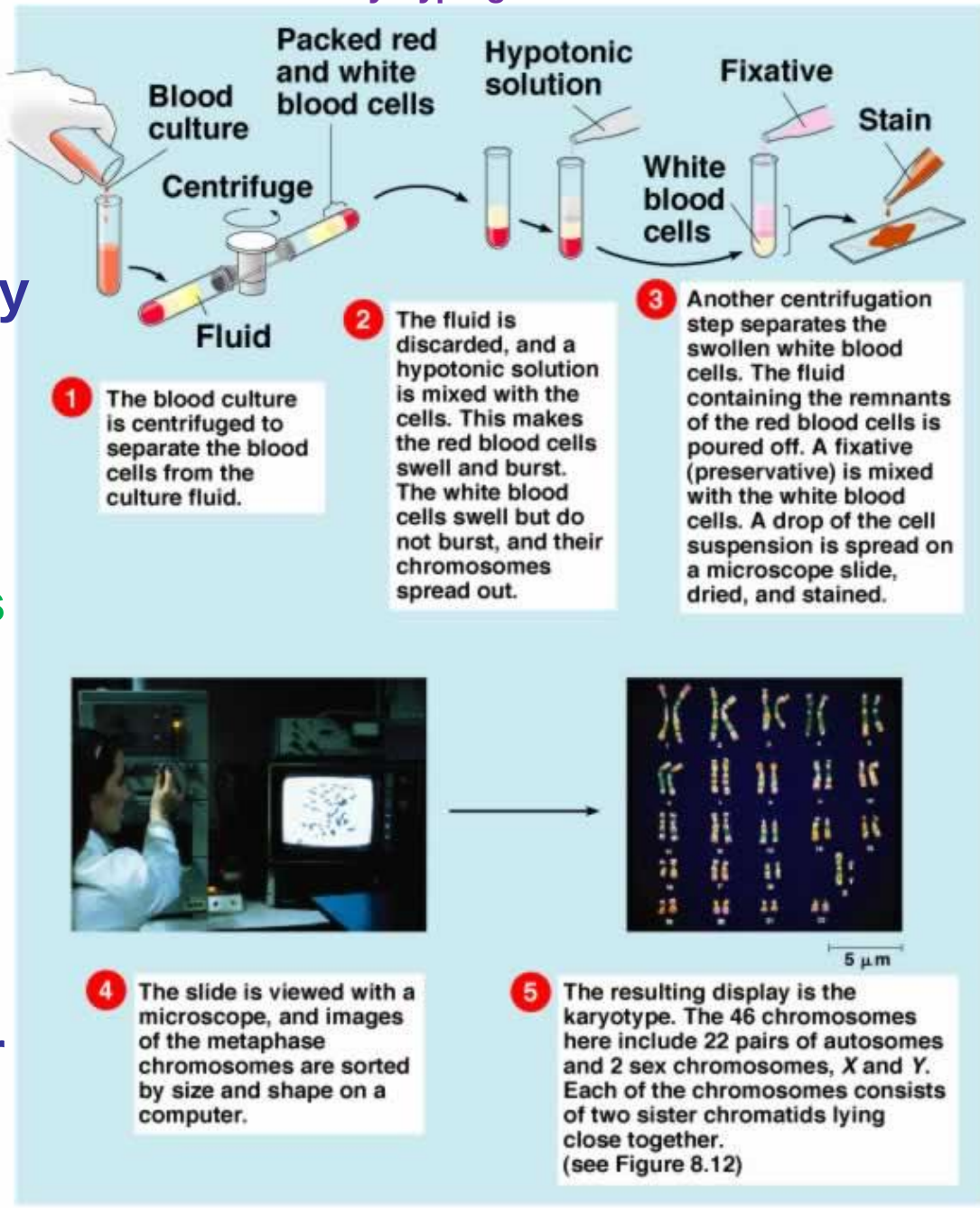
5

The resulting display is the karyotype.

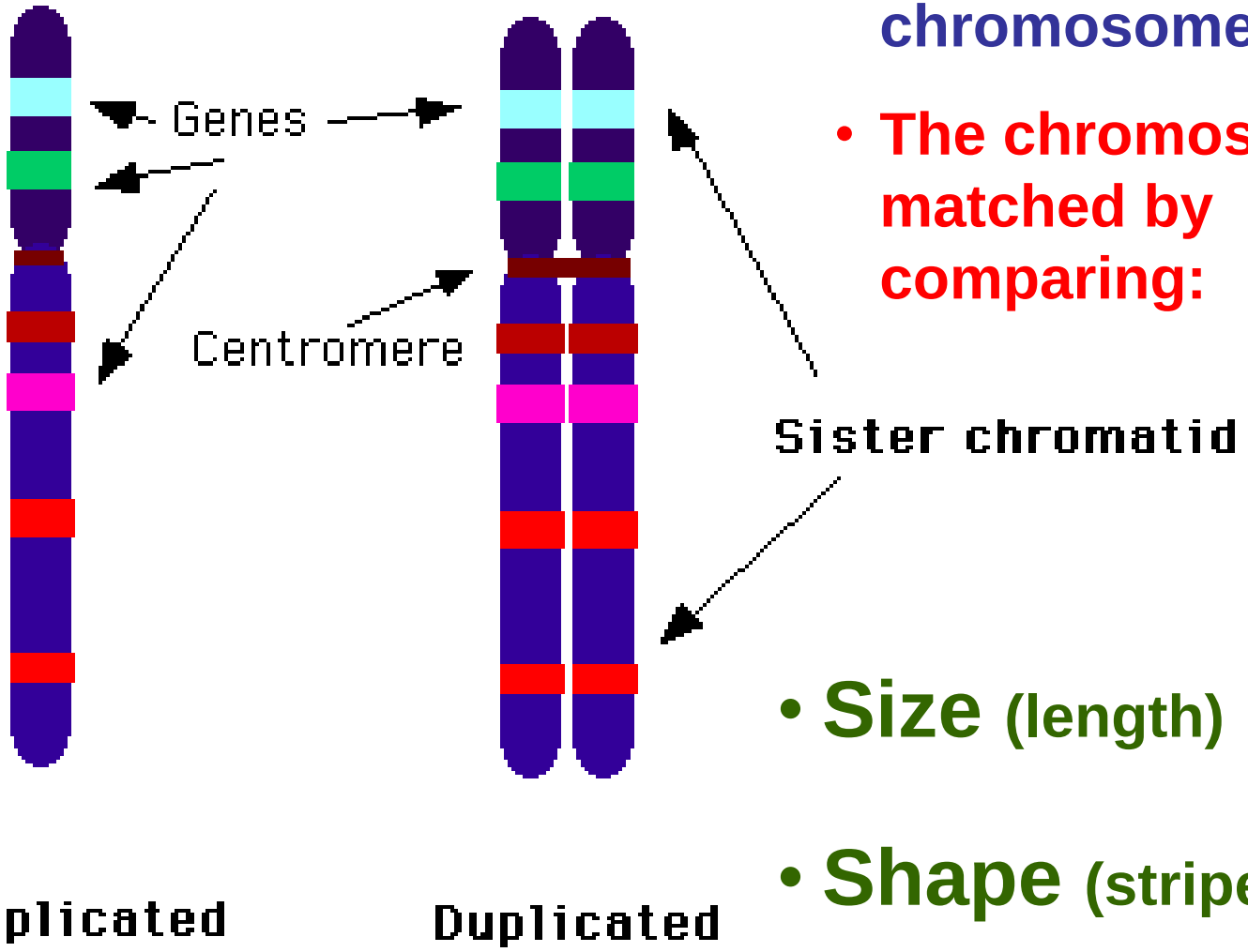
The 46 chromosomes found indicates:
22 pairs of autosomes and 2 sex chromosomes: X and Y.

Each of the chromosomes consist of two sister chromatids lying close together.

Former Method of Karyotyping



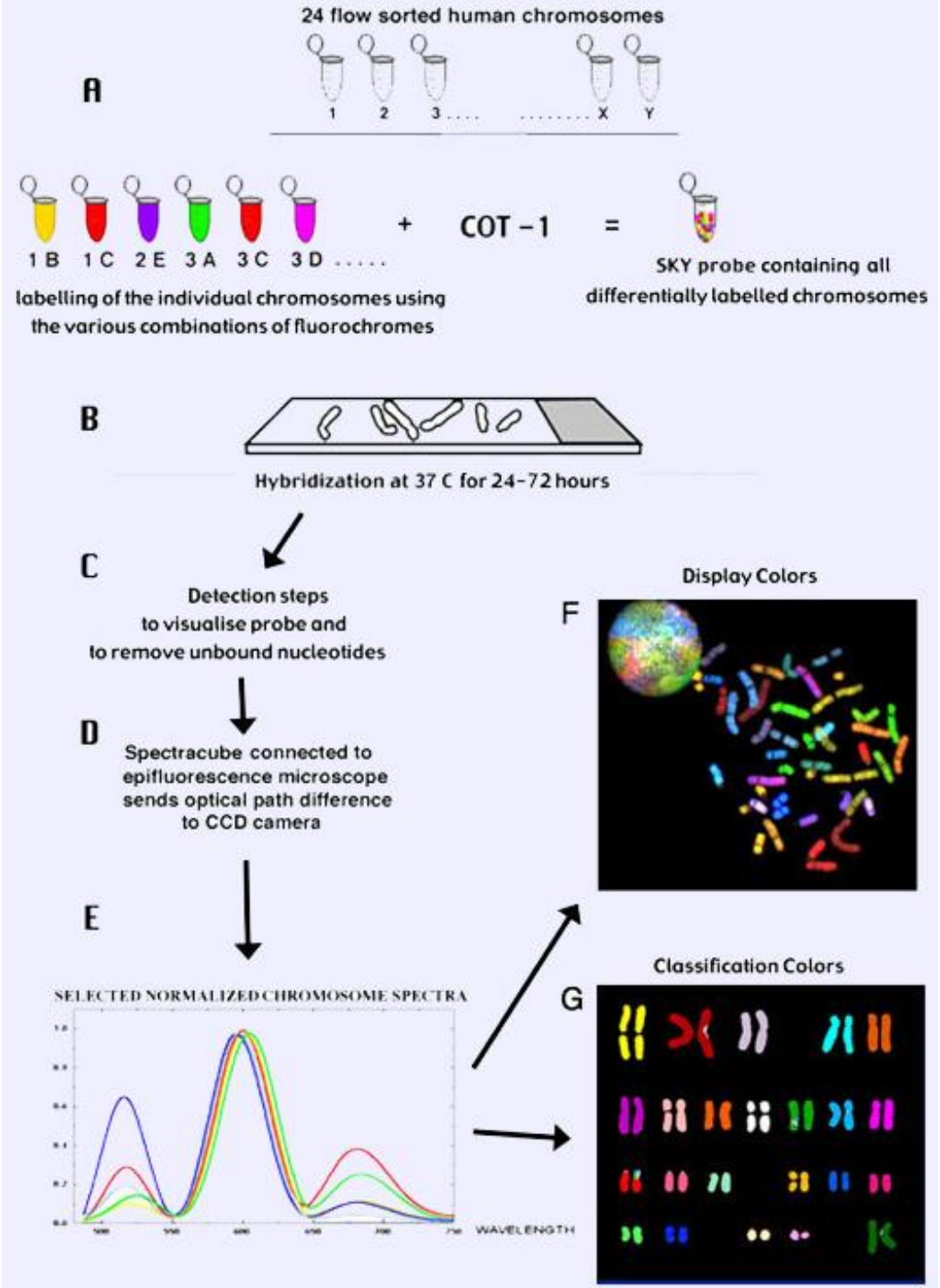
Karyotype Mapping



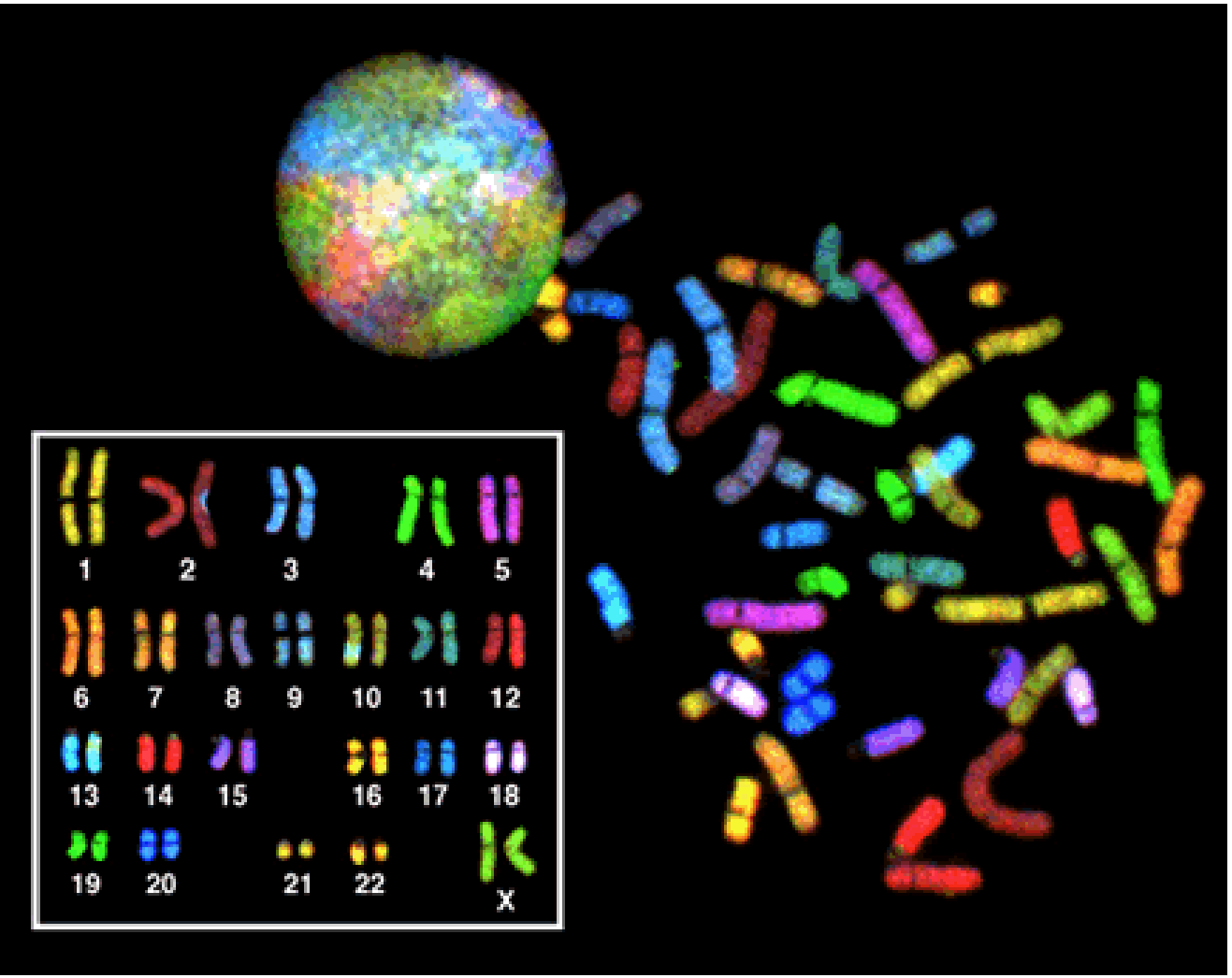
- Another karyotyping method uses different dyes to color the chromosomes.
- The chromosomes are matched by comparing:
  - **Size** (length)
  - **Shape** (stripe pattern)

A Modern Karyotyping Method

The SKY process (spectral karyotyping) dyes the chromosomes different colors for ease of matching.



Spectral Karyotyping (SKY)



SKY adds color to the sorting process.

Other than color, what are the other two sorting criteria or how are chromosomes grouped?

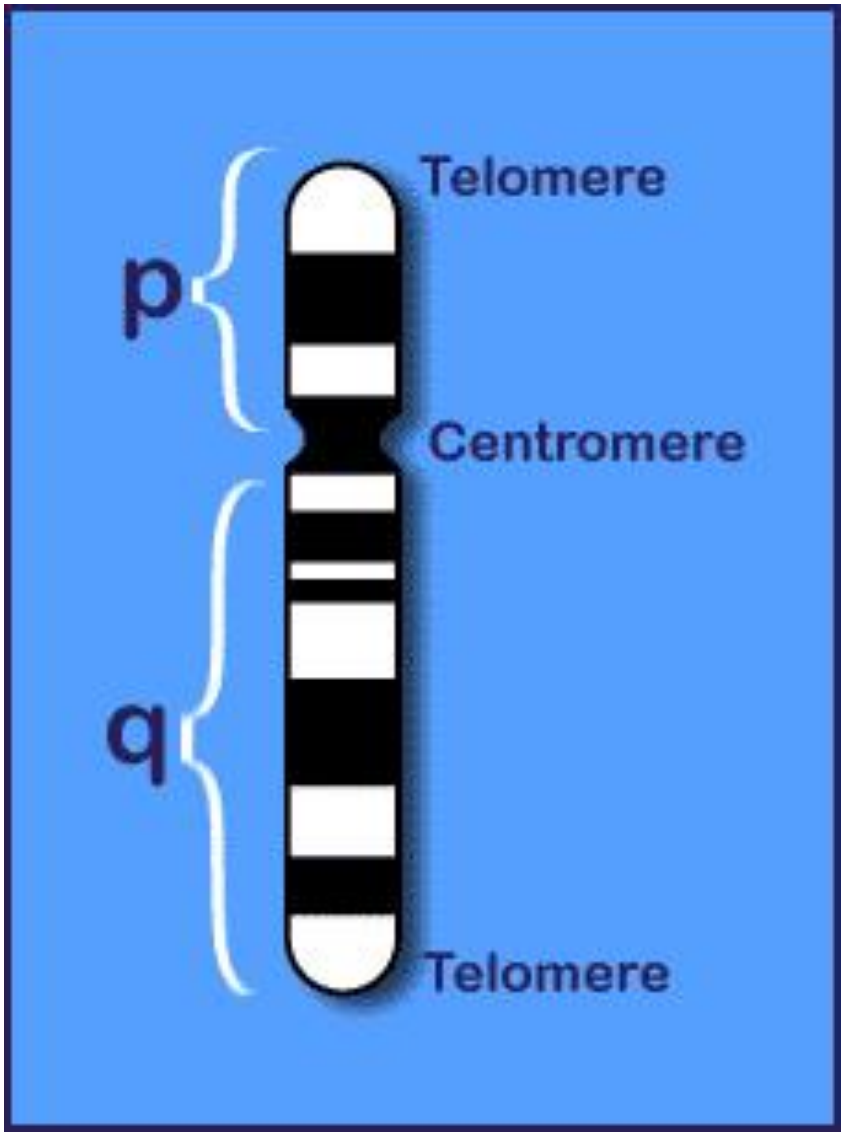
The Telomere

The **telomere** is the small section of DNA on the end of a chromosome.

Every time the chromosome duplicates the telomere slightly degrades.

When the telomere completely disappears, the chromosome can no longer duplicate.

What could happen to a cell if its ability to duplicate its chromosomes **never ended**?



QUICK CHECK

Karyotype Mapping

How many chromosomes should a human have?

What are the two types of chromosomes?

What is the name of the process that sorts chromosomes?

By which two basic criteria are chromosomes sorted into a karyotype?

Which advantage does the Spectral Karyotyping (SKY) have over the standard method?

Where are the Telomeres located on a chromosome?

What happens to the Telomeres every time the chromosome divides?

What can happen to the cell if the Telomeres do NOT eventually disintegrate?

QUICK CHECK

Karyotype Mapping

How many chromosomes should a human have? 46

What are the two types of chromosomes? autosomes, gametes

What is the name of the process that sorts chromosomes? karyotyping

By which two basic criteria are chromosomes sorted into a karyotype? Size (length) and Shape (stripe pattern)

Which advantage does the Spectral Karyotyping (SKY) have over the standard method? Easier to match chromosome pairs

Where are the Telomeres located on a chromosome? End of chromosome

What happens to the Telomeres every time the chromosome divides? Partial disintegration

What can happen to the cell if the Telomeres do NOT eventually disintegrate? Defect or abnormality

Genetic Disorders

According to **National Human Genome Research Institute** a genetic disorder is a disease caused by a different form of a gene called a **variation**, or an alteration of a gene called a **mutation**.

Many diseases have a genetic aspect. Some, including many **cancers**, are caused by a mutation in a gene or group of genes in a person's cells.

These mutations can occur randomly or because of an **environmental exposure** such as **cigarette smoke**.

Genetic Disorders

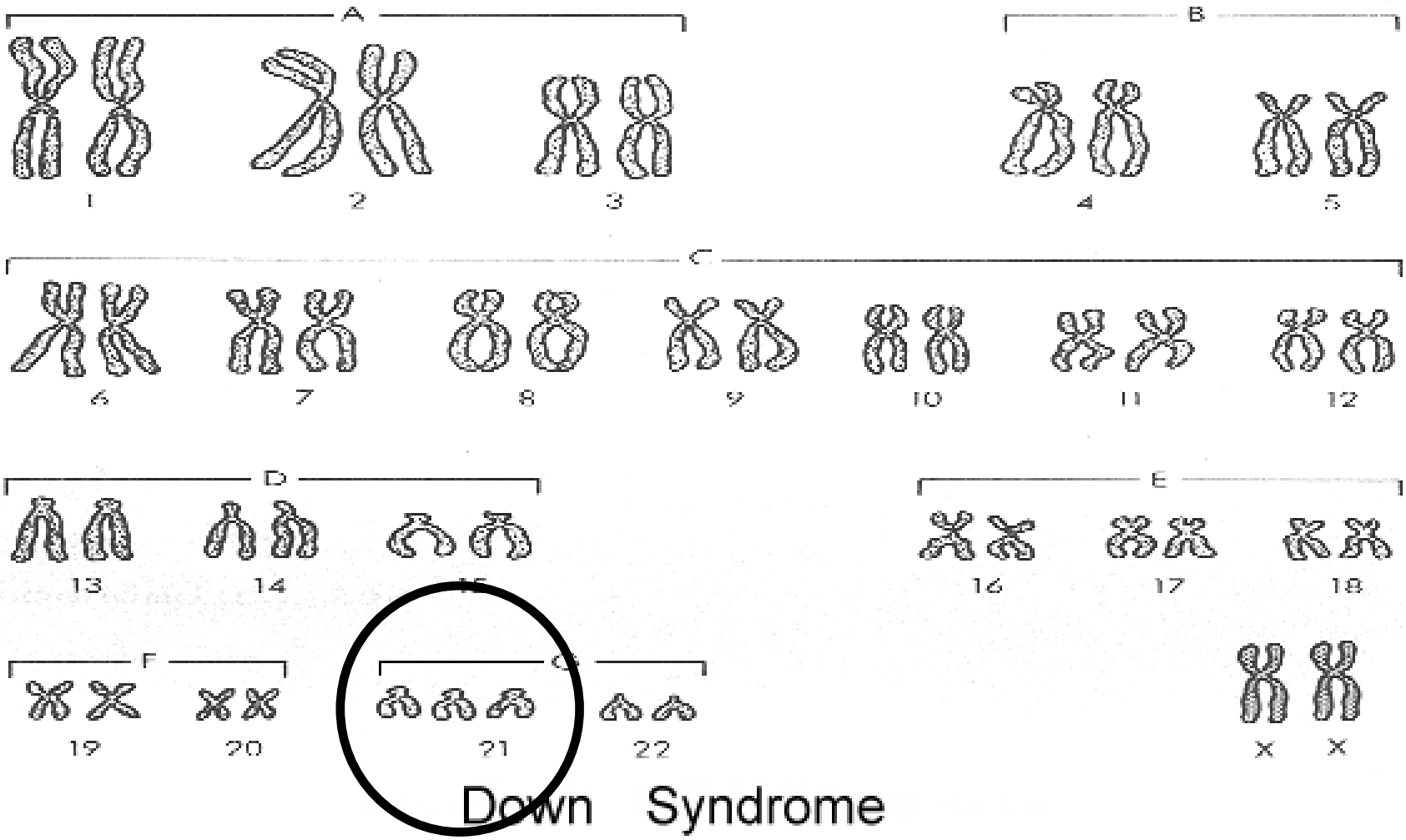
Other genetic disorders are **inherited**. A mutated gene is passed down through a family and each generation of children can inherit the gene that causes the disease.

Still other genetic disorders are due to **problems with the number of packages of genes** called chromosomes.

- In Down syndrome, for example, there is an extra copy of chromosome 21.

In the following karyotpe, can you identify **chromosome 21**?

A Karyotype of a Person with Down Syndrome



Commonly Used Terms:
Unaffected, Carriers & Affected

- **Affected** persons have a genetic disease or condition.
- **Carriers** do not have a genetic disease, but carry the gene which causes the disease.
  - Carriers have **one normal/dominant** allele + **one recessive** allele for a disease.
  - There is **NO** effect of the recessive allele on a carrier, because it has a normal/dominant allele that is expressed.
- **Unaffected** persons do not have the disease and do not carry the gene for it either.

Straight hairline

Widow's peak

TRY IT

KEY

Female Male

Widow's peak hairline trait — *H*: widow's peak allele

Straight hairline trait — *h*: straight allele

Mating

1ST GENERATION

Al Beth

Charles Debbie

Children

2ND GENERATION

Evelyn Frank Gary Henry Isabel Juliana

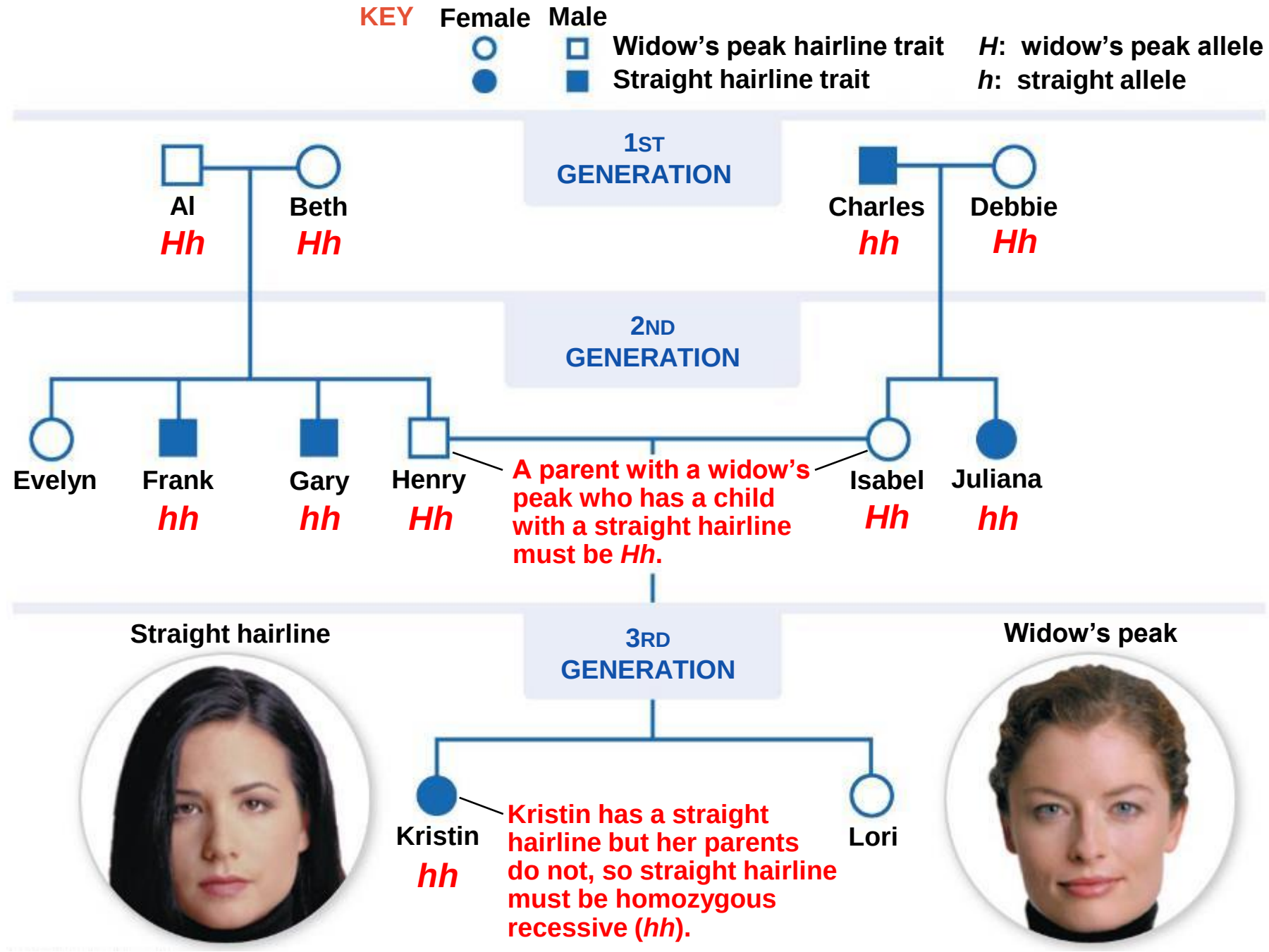
Straight hairline

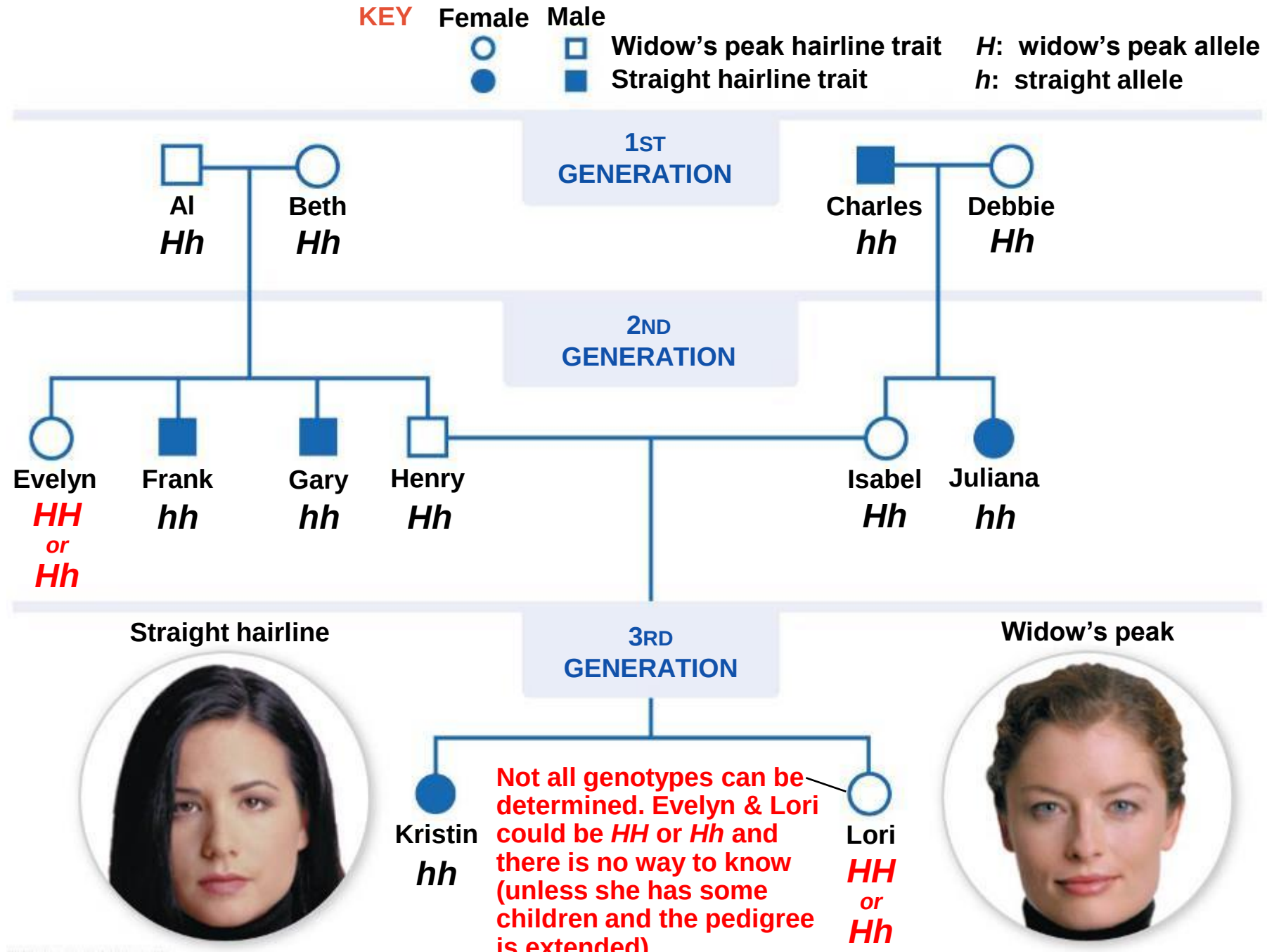
3RD GENERATION

Kristin Lori

Widow's peak

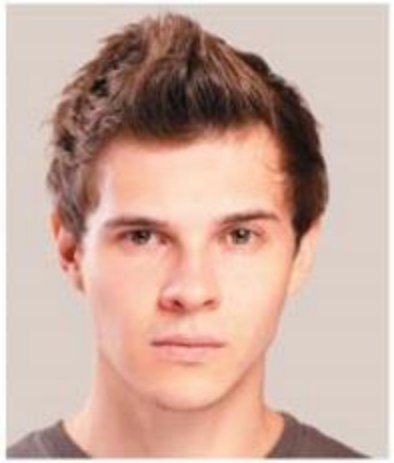
GENOTYPES??


KEY
Female
Male
Widow's peak hairline trait
Straight hairline trait
H: widow's peak allele
h: straight allele
1ST GENERATION
Al
Hh
Beth
Hh
Charles
hh
Debbie
Hh
2ND GENERATION
Evelyn
hh
Frank
hh
Gary
hh
Henry
Hh
A parent with a widow's peak who has a child with a straight hairline must be Hh.
Isabel
Hh
Juliana
hh
Straight hairline
3RD GENERATION
Widow's peak
Kristin
hh
Kristin has a straight hairline but her parents do not, so straight hairline must be homozygous recessive (hh).
Lori


KEY
Female
Male
Widow's peak hairline trait
Straight hairline trait
H: widow's peak allele
h: straight allele
1ST GENERATION
Al
Hh
Beth
Hh
Charles
hh
Debbie
Hh
2ND GENERATION
Evelyn
HH
or
Hh
Frank
hh
Gary
hh
Henry
Hh
Isabel
Hh
Juliana
hh
Straight hairline
3RD GENERATION
Widow's peak
Kristin
hh
Not all genotypes can be determined. Evelyn & Lori could be HH or Hh and there is no way to know (unless she has some children and the pedigree is extended).
Lori
HH
or
Hh

Many Inherited Traits in Humans are controlled by a Single Gene

- Because a trait is dominant does NOT mean that it is
  - "**normal**" or
  - **more common than a recessive trait**.
- **Wild-type Traits** are
  - those **most often seen in nature** and
  - not necessarily specified by dominant alleles.

Dominant Traits	Recessive Traits
	
Freckles	No freckles
	
Normal pigmentation	Albinism

Key

- Wild-type (more common) trait
- Less Common trait

Many Inherited Traits in Humans are controlled by a Single Gene

- The genetic disorders listed in the following table are known to be inherited as dominant or recessive traits controlled by a single gene.
- These human disorders therefore show simple inheritance patterns like the traits Mendel studied in pea plants.
- These genes are all located on autosomes.

SOME AUTOSOMAL DISORDERS IN HUMANS

Disorder	Major Symptoms	Incidence	Comments
Recessive Disorders			
Albinism	Lack of pigment in the skin, hair, and eyes	1/22,000	
Cystic fibrosis	Excess mucus in the lungs, digestive tract, liver; increased susceptibility to infections; death in early childhood unless treated	1/2,500 Caucasians	
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; developmental disabilities	1/10,000 in United States and Europe	
Sickle-cell disease	Sickled red blood cells; damage to many tissues	1/400 African Americans	
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	1/3,600 Jews from central Europe	

AUTOSOMAL RECESSIVE

Many Inherited Traits in Humans are controlled by a Single Gene

- Thousands of human genetic disorders — ranging in severity from relatively mild, such as albinism, to invariably fatal, such as cystic fibrosis — are inherited as **recessive traits**.
- Most people who have recessive disorders are born to normal parents who
  - are both **heterozygotes**, **carriers** of the recessive allele for the disorder, but
  - are phenotypically normal.

Parents

Normal ***Aa*** × **Normal** ***Aa***

	Sperm ***A***	Sperm ***a***
Eggs ***A***	***AA*** Normal	***Aa*** Normal (carrier)
Eggs ***a***	***Aa*** Normal (carrier)	***aa*** Albinism

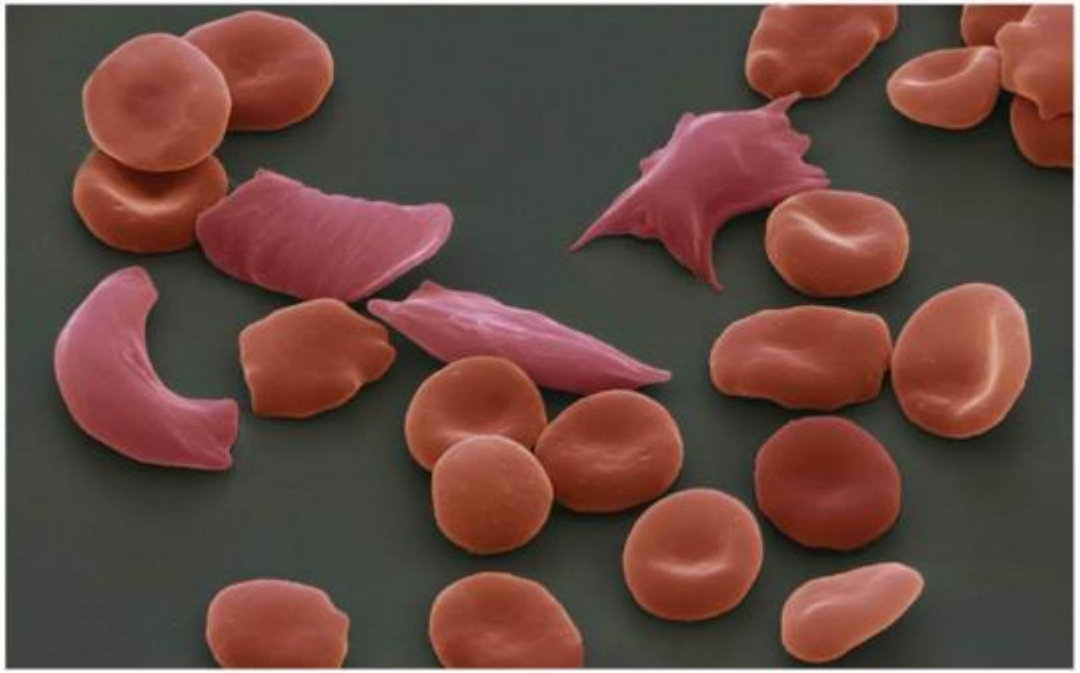
Offspring

Sickle Cell Disease

- Transmitted in an **autosomal recessive** fashion.
- Most common inherited disease seen in individuals of African descent.
- This disease affects the type of **hemoglobin** produced and the shape of red blood cells.
- Caused by a **mutation** in one of the proteins that makes up hemoglobin.
- This is a **substitution mutation** in one of the protein chains of hemoglobin.
- This results in the amino acid **valine** being inserted into this chain instead of the normal **glutamic acid**.

Sickle Cell Disease

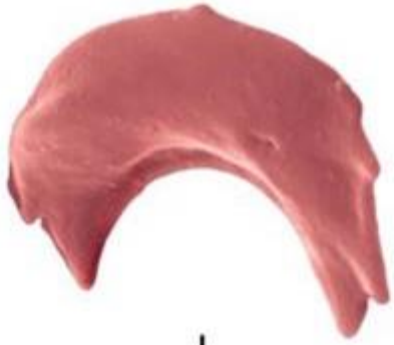
- This replacement of glutamic acid with valine results in a hemoglobin molecule with abnormal structure distorting the shape of the red blood cells.
- As a result hemoglobin cannot carry oxygen properly.



An individual homozygous for the sickle-cell allele

Produces sickle-cell (abnormal) hemoglobin

The abnormal hemoglobin crystallizes, causing red blood cells to become sickle-shaped



Damage to organs

Kidney failure
Heart failure
Spleen damage
Brain damage (impaired mental function, paralysis)

Other effects

Pain and fever
Joint problems
Physical weakness
Anemia
Pneumonia and other infections

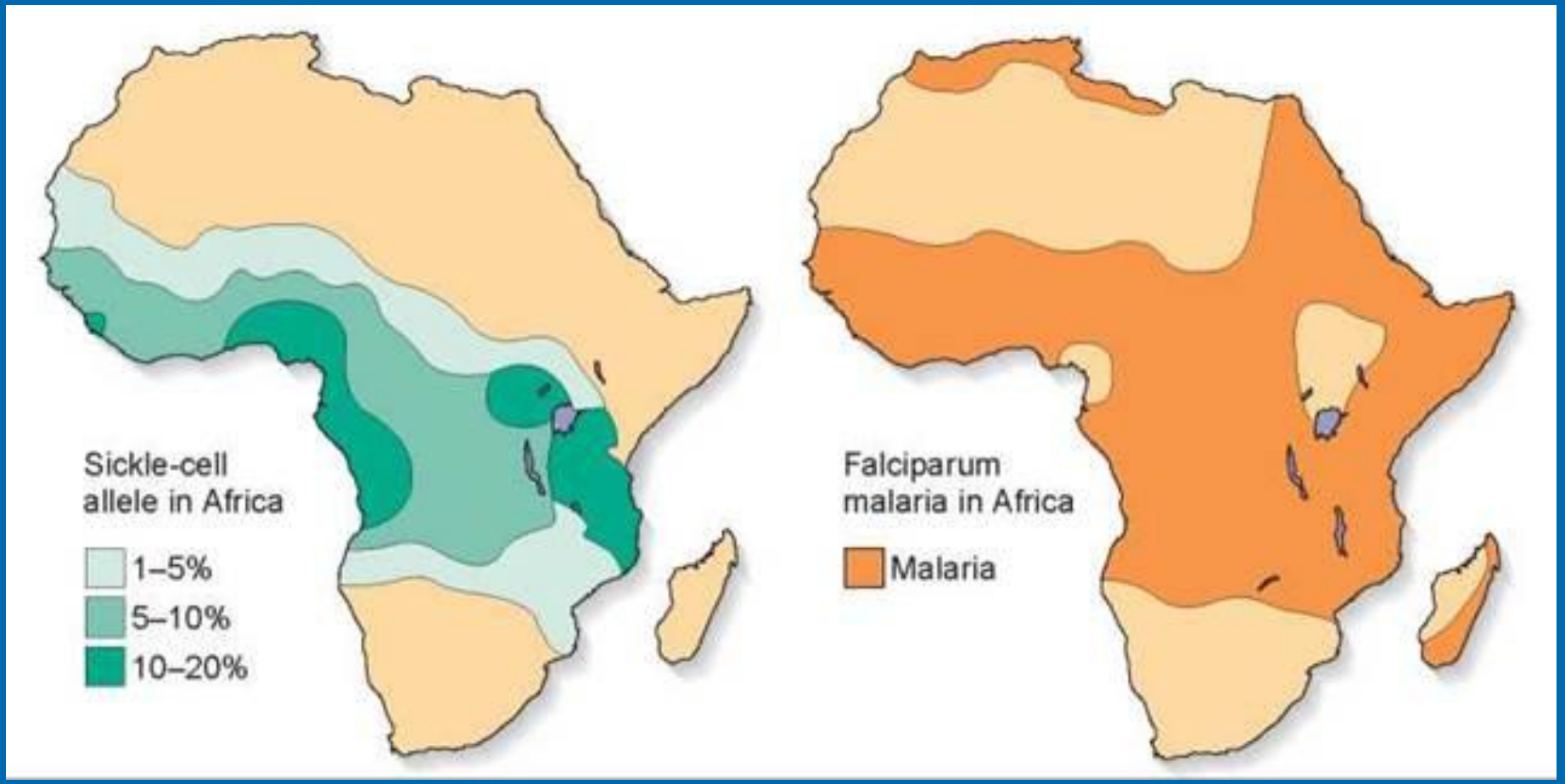
Heterozygous Advantage

- The condition of the heterozygous form being **protective** against some disease or illness.
- This is the protection that the heterozygous condition can give to people who are **carriers of a recessive allele**.
- Example:
  - **Sickle Cell Disease** and **Malaria**
    - **Carriers** of sickle-cell disease have **increased resistance to malaria**.

Heterozygous Advantage

- The **recessive sickle-cell allele** produces hemoglobin with reduced capacity to carry oxygen.
- This mutation also confers malaria resistance in **heterozygotes**. This heterozygote advantage leads to a larger portion of the recessive allele than usual in areas where malaria is widespread.
- High frequency of the **mutant allele** in people of African descent.
- The mutant allele results in **defective hemoglobin molecules** which alter the red blood cells so that they are **less susceptible to infection by the malarial** parasite.

Heterozygous Advantage

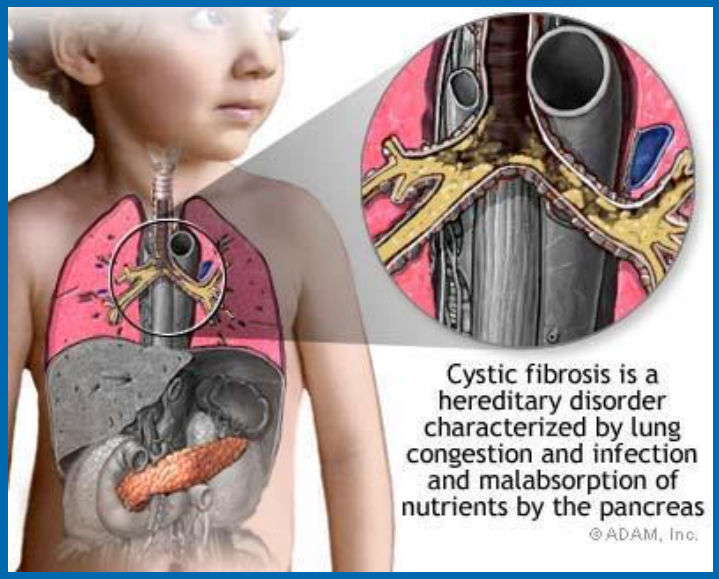


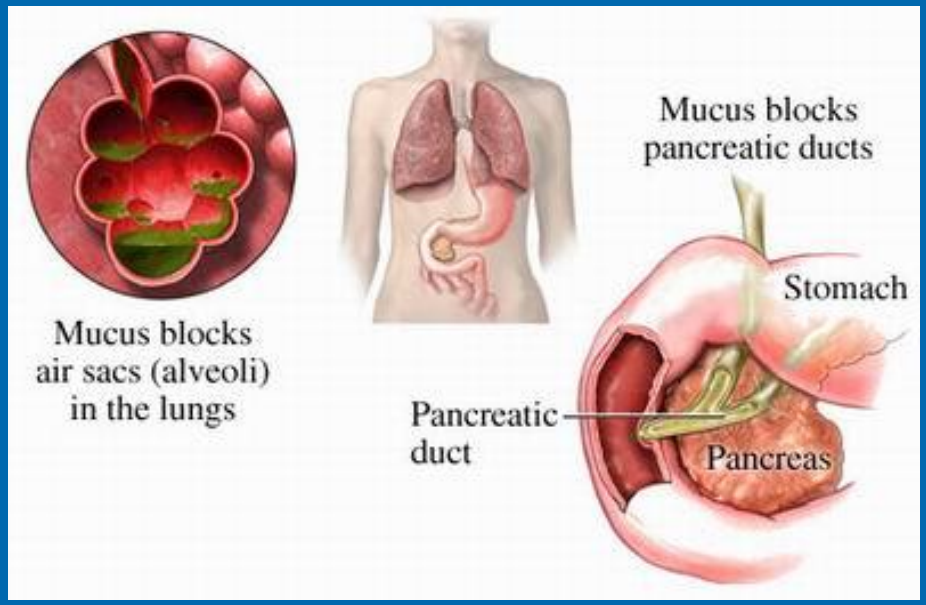
Phenylketonuria: PKU

- Transmitted in an **autosomal recessive** fashion.
- Inability to break down the amino acid **phenylalanine**.
- Enzyme which breaks down the amino acid phenylalanine is defective.
- Caused by a mutated gene which codes for this enzyme.
- Therefore phenylalanine builds up in the brain, causing **mental retardation** and other problems.
- Requires **elimination of this amino acid from the diet** or serious mental retardation will result.

Cystic Fibrosis

- Transmitted in an **autosomal recessive** fashion.
- Single most common inherited disease among **Caucasians**.
- Results in **increased (thick) airway and digestive tract mucus** that causes breathing problems and recurrent infections; Lethal.
- Caused by a **defective** membrane chloride channel **protein**.





SOME AUTOSOMAL DISORDERS IN HUMANS

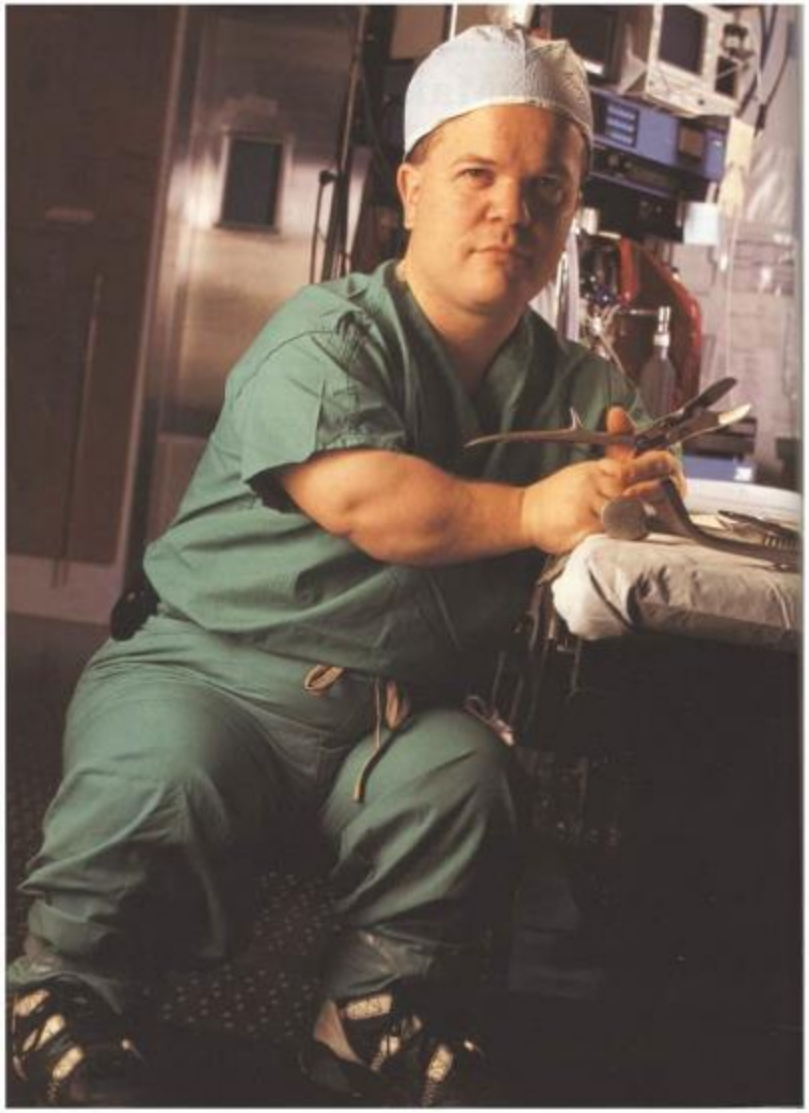
Disorder	Major Symptoms	Incidence	Comments
Dominant Disorders			
Achondroplasia	Dwarfism	1/25,000	See Module 9.9
Huntington's disease	Developmental disabilities and uncontrollable movements; strikes in middle age	1/25,000	See Module 9.9
Hypercholesterolemia	Excess cholesterol in the blood; heart disease	1/500 are heterozygous	See Module 9.11

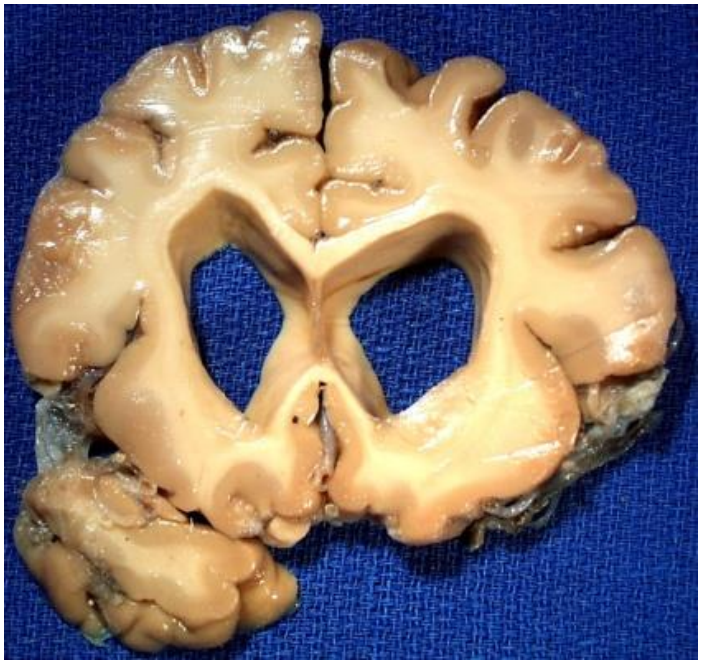
AUTOSOMAL DOMINANT

Autosomal Dominant

- Based on a **Dominant allele**
- Human disorders include
  - **Huntington's Disease**, a degenerative disorder of the nervous system and
  - **Achondroplasia**, a form of dwarfism in which
    - the head and torso of the body develop normally but
    - the arms and legs are short.

Achondroplasia





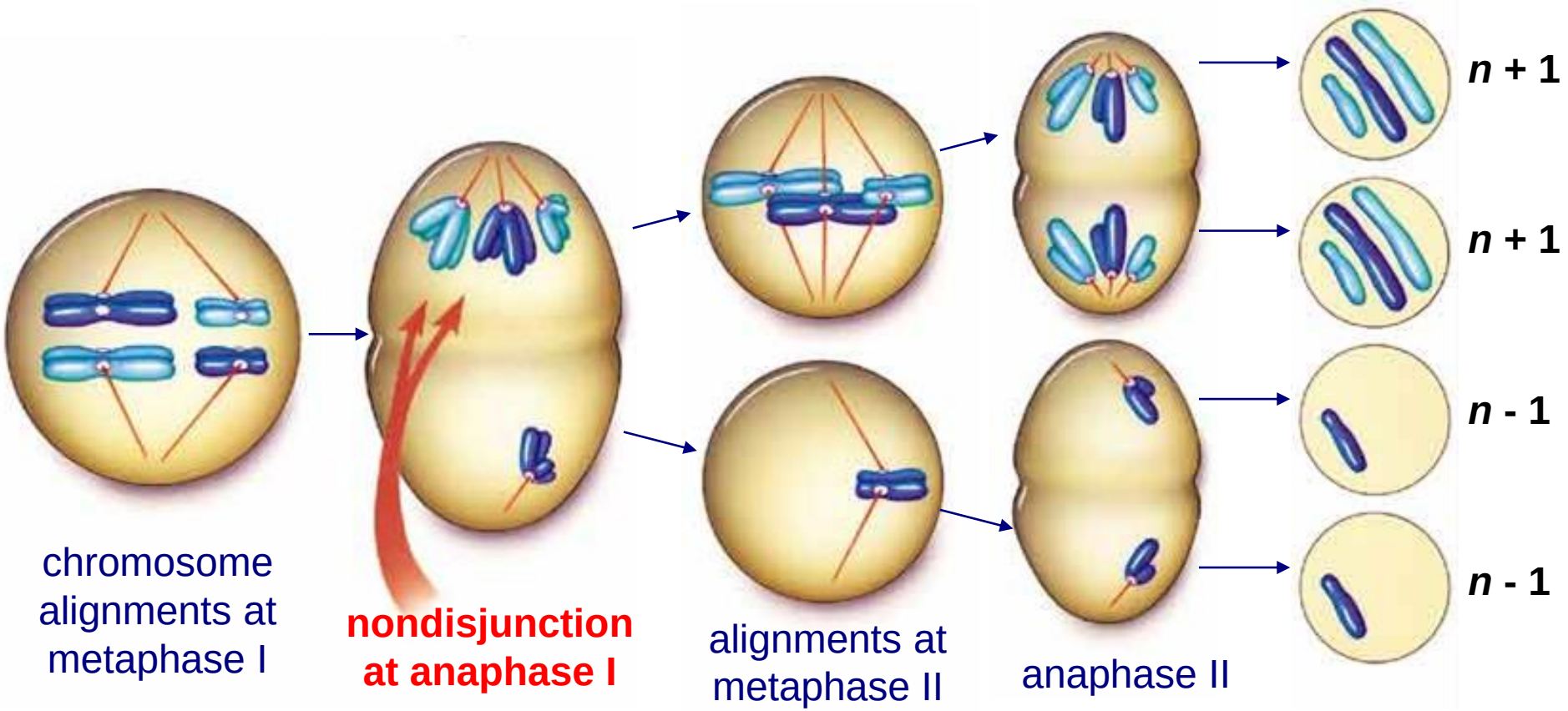
Congenital Genetic Diseases

- Disease a child is **born with** due to a mutation in the DNA.
- May be a **Point** or **Chromosomal** Mutation and based on dominant allele in chromatid pair.
- Many are called **Syndromes** because the chromosome abnormalities result in common problems associated with the specific chromosomal mutation.
- There are many examples of dominant allele genetic disorders called: **Aneuploidy**

Aneuploidy (autosomal dominance)

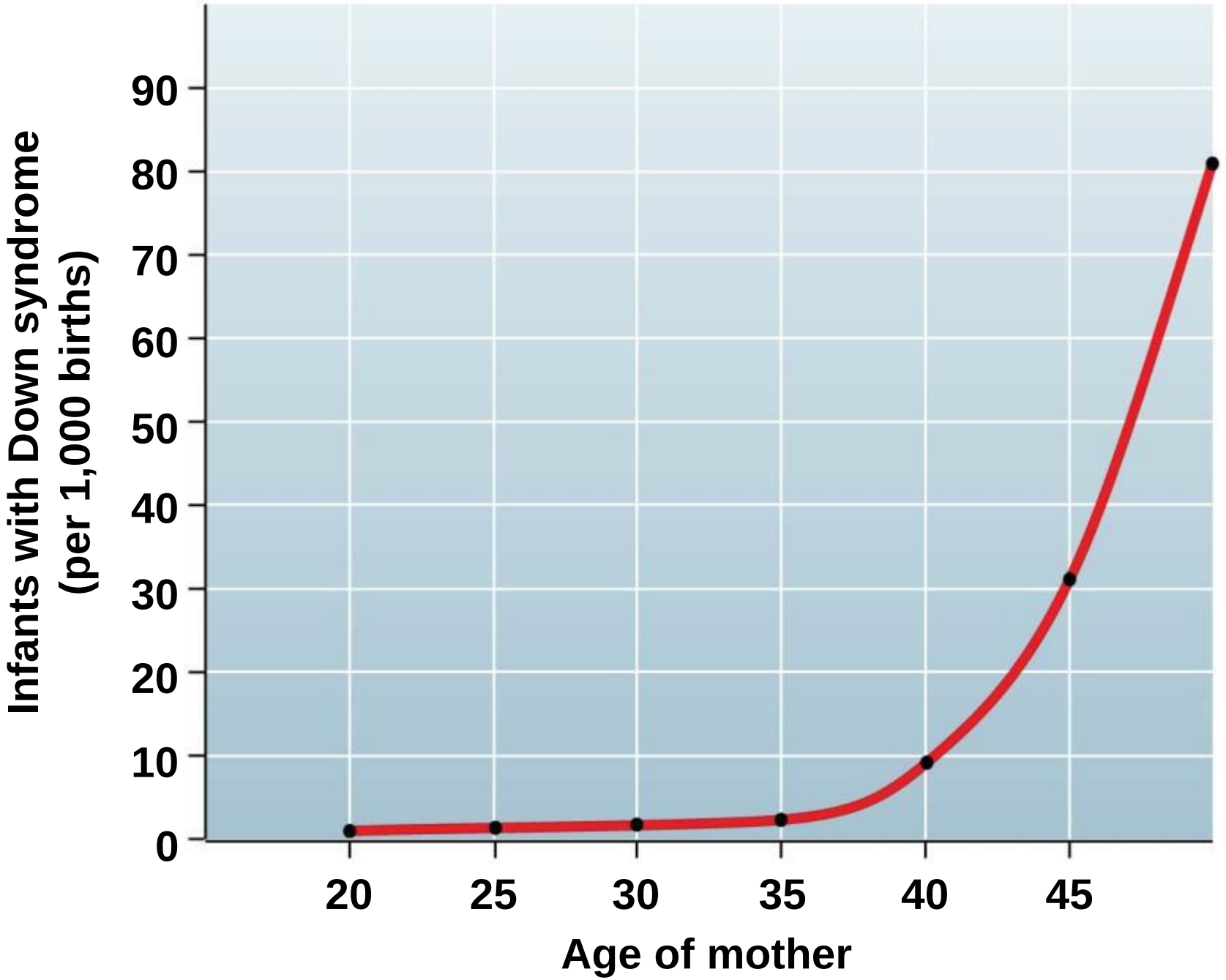
- Individuals have one extra or less chromosome
- ($2n + 1$ or $2n - 1$)
- Caused by non-disjunction during meiosis
- Major cause of human reproductive failure
- Most human miscarriages are aneuploids

Aneuploidy caused by Nondisjunction



Down Syndrome

- Trisomy of chromosome 21
- Mental impairment and a variety of additional defects.
- Can be detected before birth.
- Risk of Down syndrome increases dramatically in mothers over age 35.


Infants with Down syndrome
(per 1,000 births)
90
80
70
60
50
40
30
20
10
0
20
25
30
35
40
45
Age of mother

How Does Down Syndrome Affect a Child?

Kids with Down syndrome tend to share certain physical features such as:

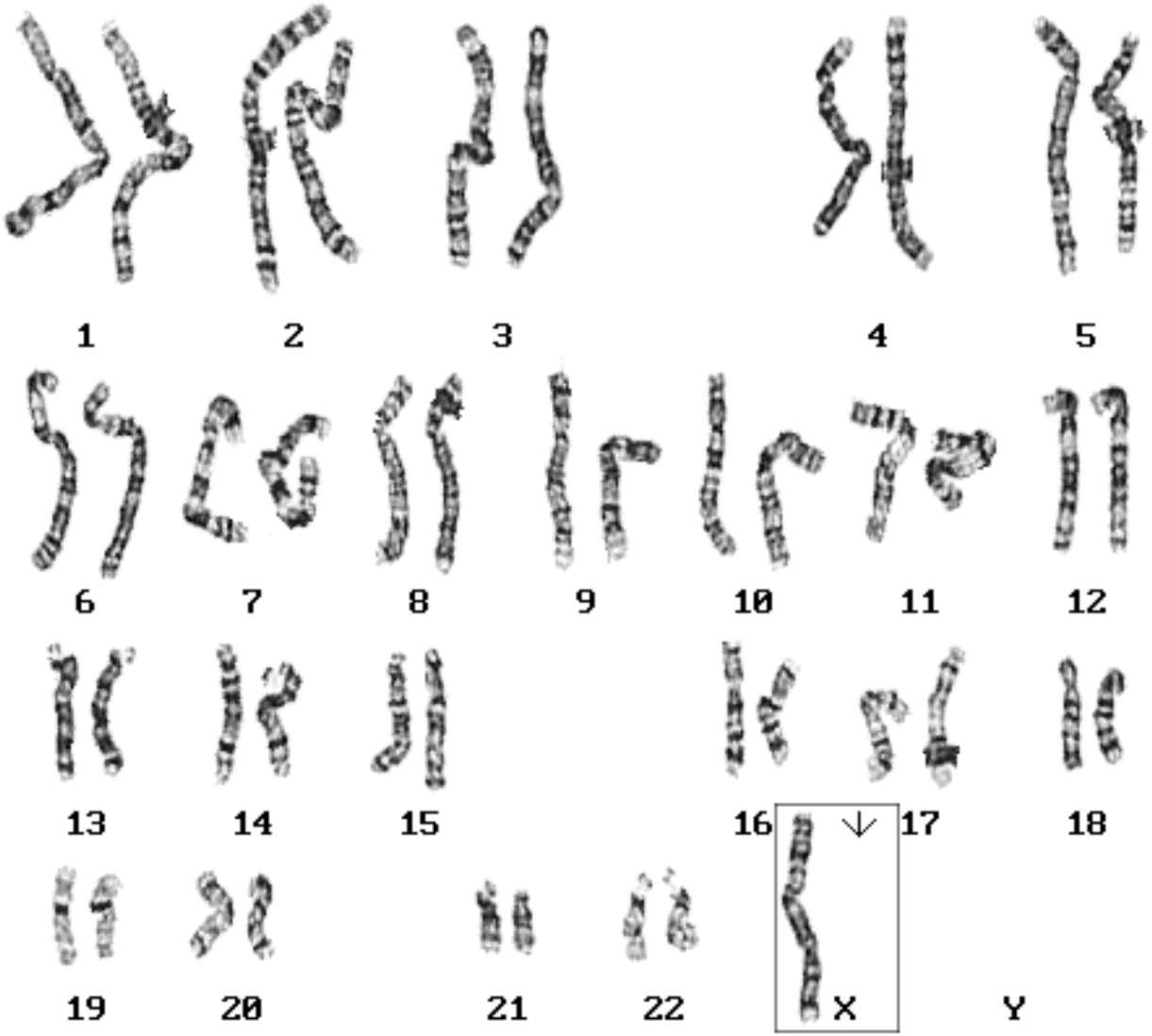
- a flat facial profile,
- an upward slant to the eyes,
- small ears,
- a single crease across the center of the palms, and
- an enlarged tongue.

A doctor can usually tell if a newborn has the condition through a physical exam.

Turner Syndrome

- Inheritance of only one X (XO)
- 98% spontaneously miscarried
- Survivors are short, infertile females
  - No functional ovaries
  - Secondary sexual traits reduced
  - May be treated with hormones, surgery

A Karyotype of a Person with **Turner's Syndrome**

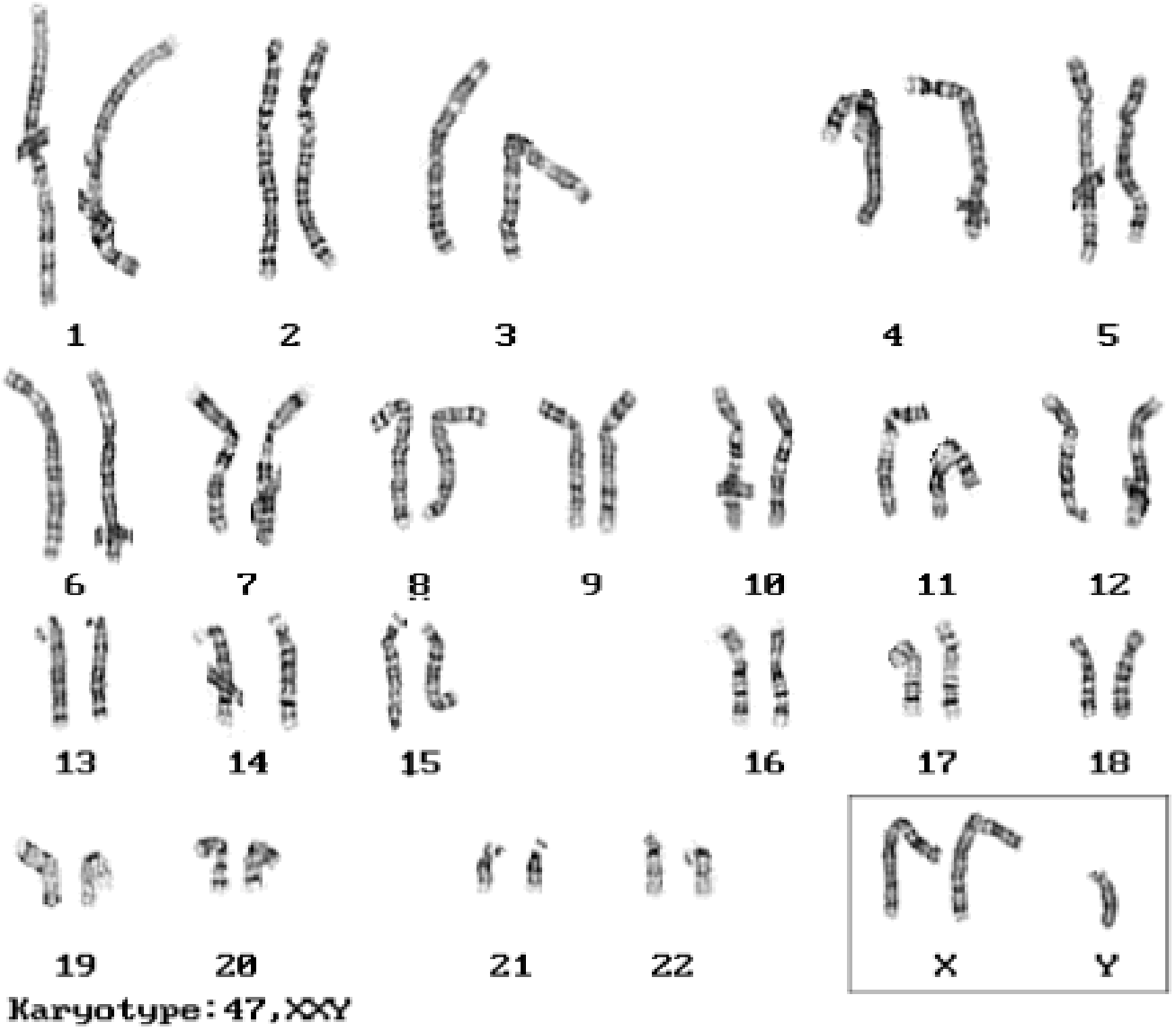


This karyotype lacks of an entire sex chromosome (Y).

Klinefelter Syndrome

- XXY condition
- Results mainly from nondisjunction in mother (67%)
- Phenotype is tall males
  - Sterile or nearly so
  - Feminized traits (sparse facial hair, somewhat enlarged breasts)
  - Treated with testosterone injections

A Karyotype of a Person with **Klinefelter Syndrome**

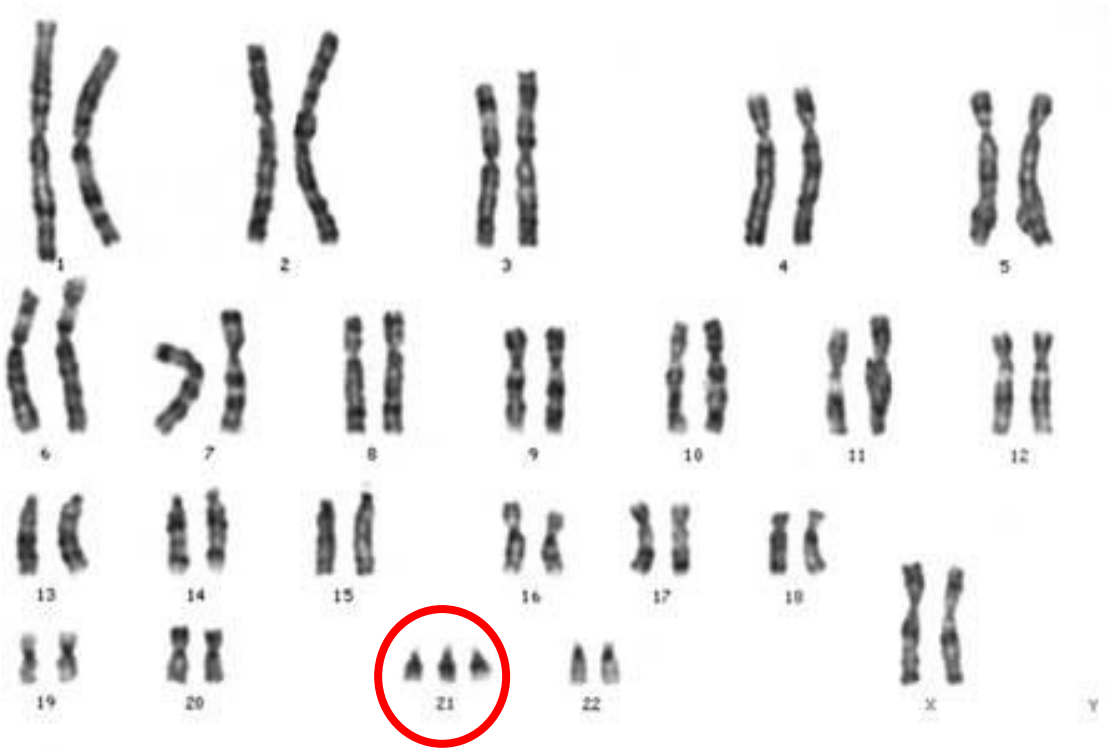


This karyotype shows an extra X chromosome.

Aneuploidy

Having one extra or one less chromosome

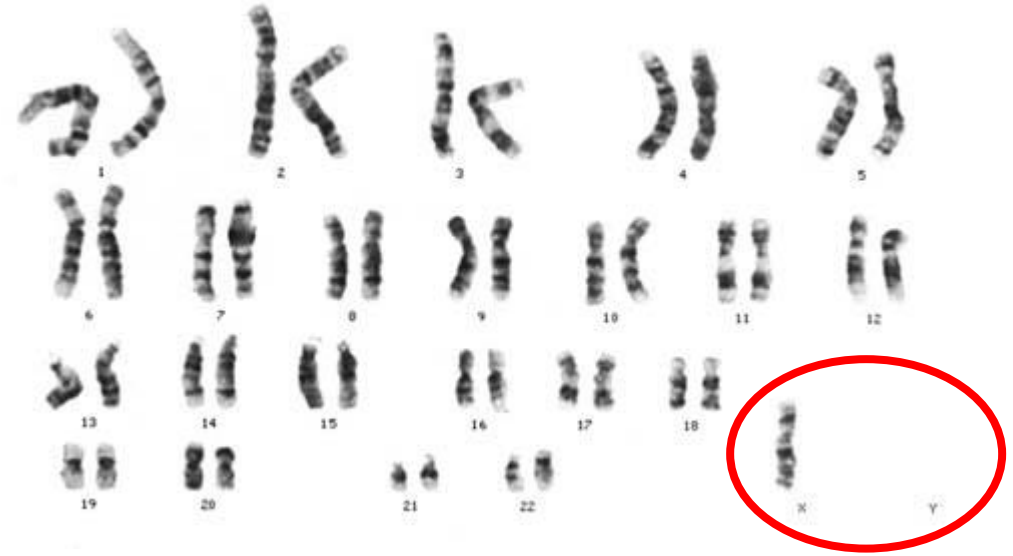
Down Syndrome Trisomy 21



Aneuploidy

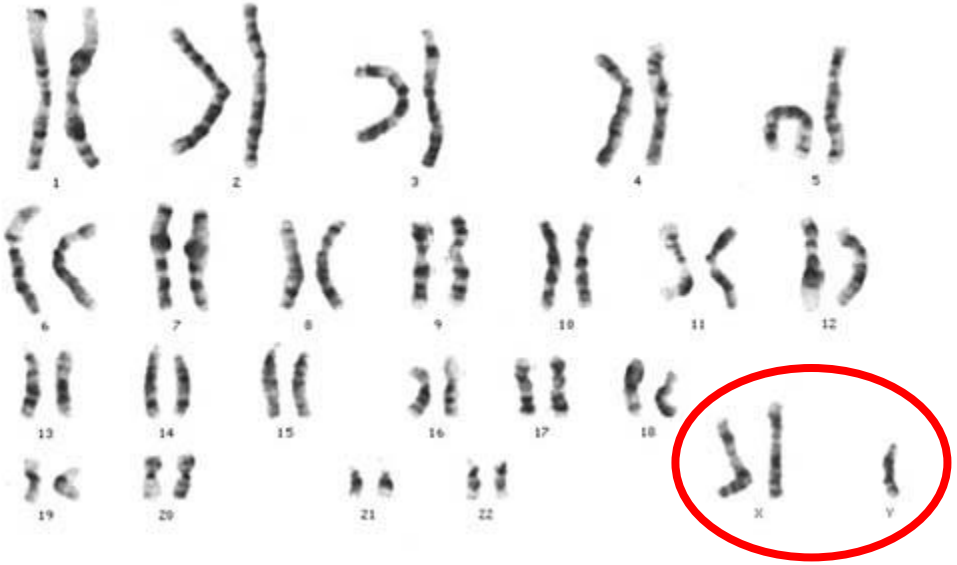
Turner Syndrome

XO

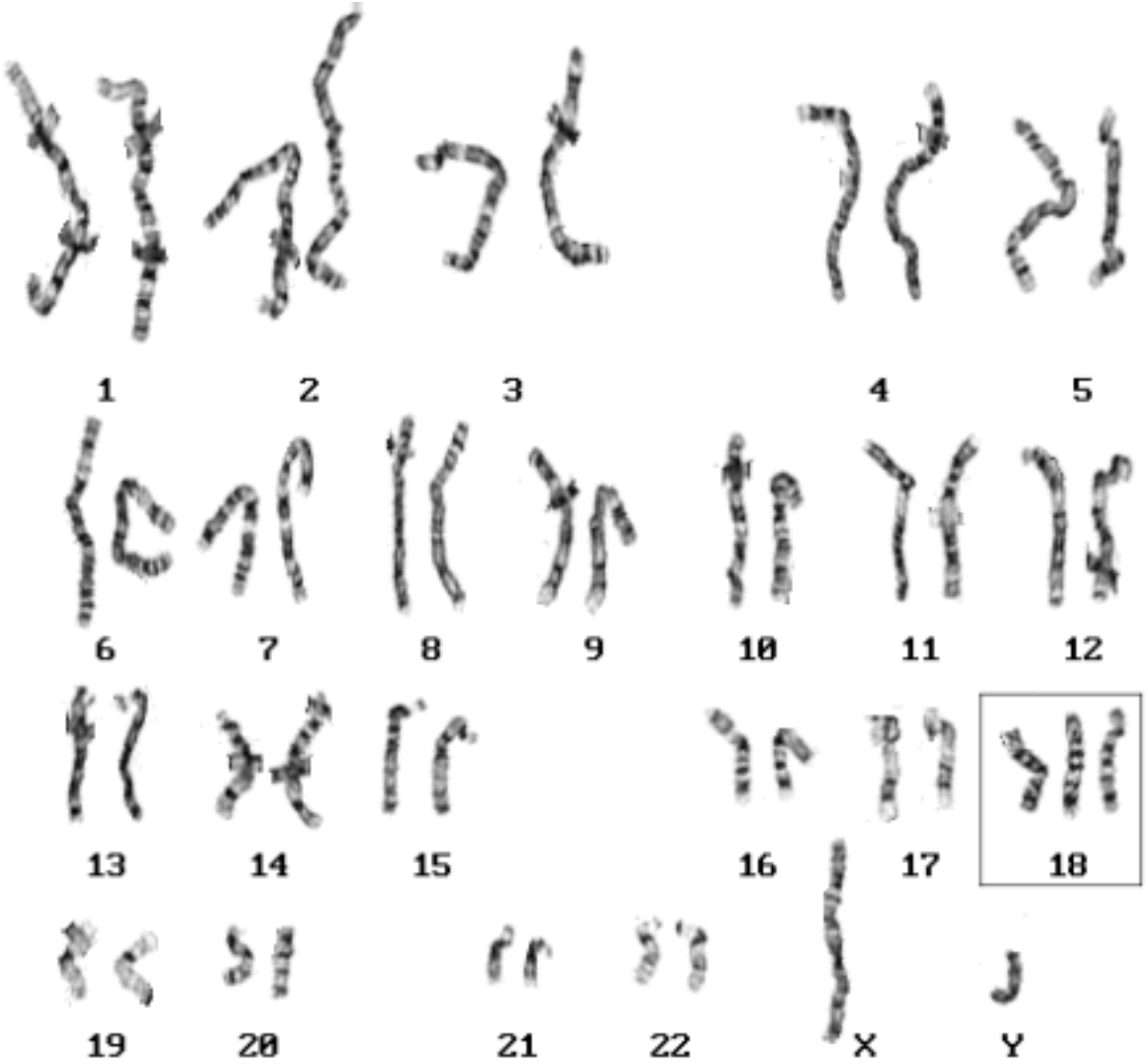


Klinefelter Syndrome

XXY

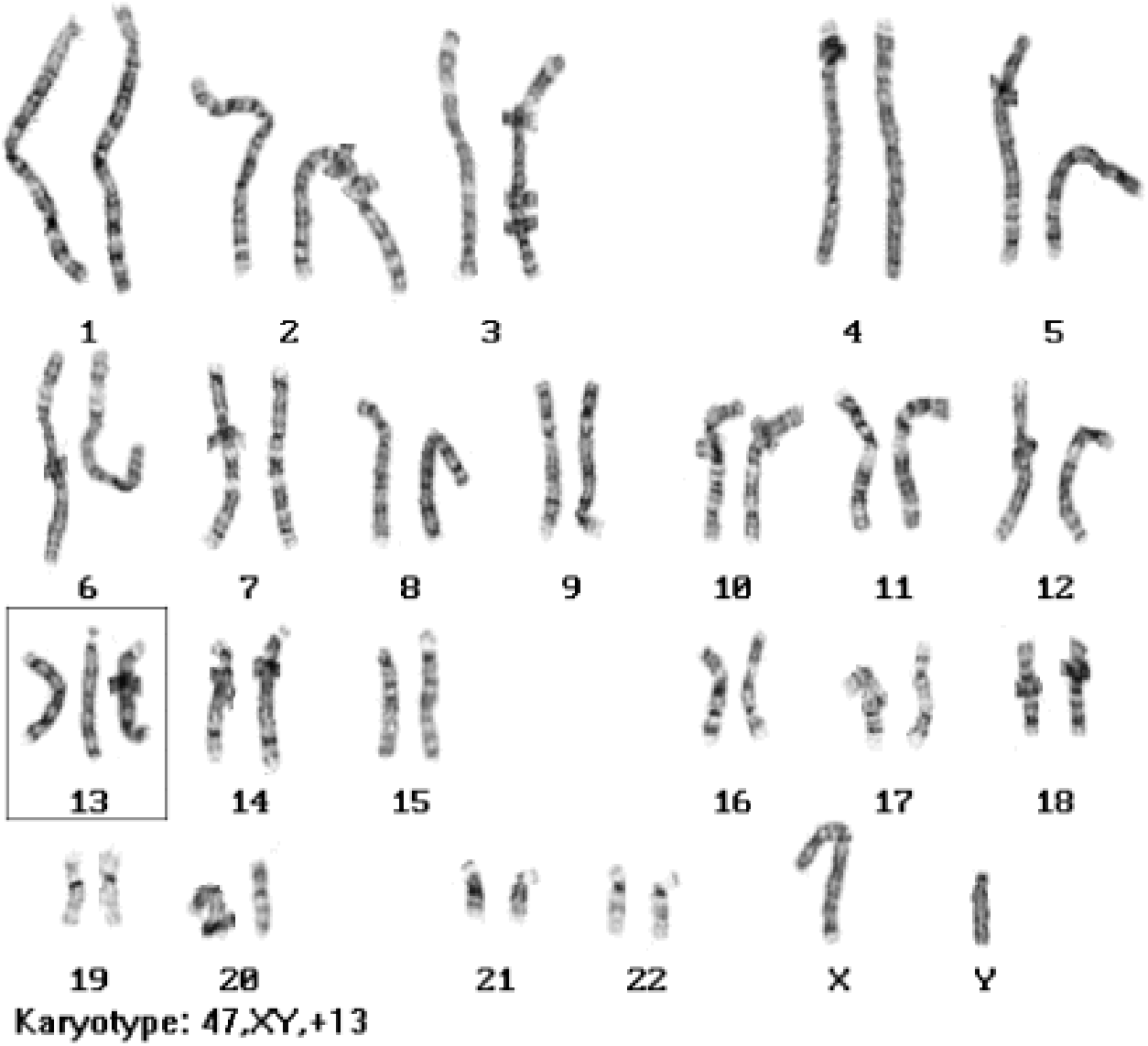


A Karyotype of a Person with **Edward's Syndrome**



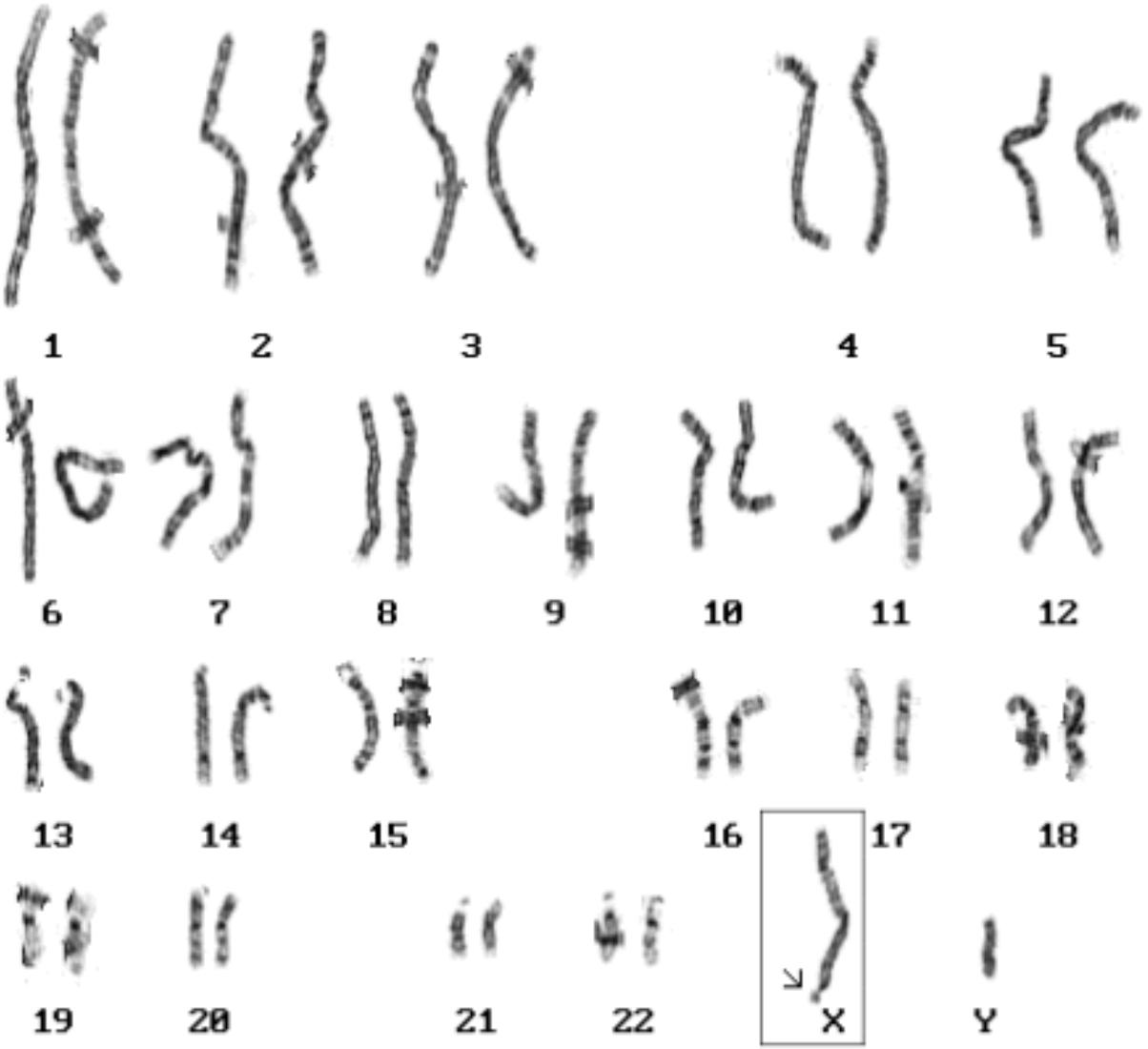
Also called Trisomy 18
(3 of chromosome 18)

A Karyotype of a Person with **Patau's Syndrome**



Also called
Trisomy 13
(3 of chromosome 13)

A Karyotype of a Person with **Fragile X Syndrome**



Karyotype: 46,Y,fra(X)(q27.3)

A gene on the X chromosome has mutated and does not make a necessary protein.

Human Genetic Patterns of Inheritance

REVIEW:

- **Autosomal Recessive Inheritance**
  (Sickle Cell, Phenylketonuria, Cystic Fibrosis)
- **Autosomal Dominant Inheritance**
  (Huntington's, Achondroplasia, Aneuploidy)
- **Sex-linked inheritance** (e.g. hemophilia, pattern baldness)

Sex-Linked Traits Review

A. Sex-linked traits are produced by genes only on the sex chromosomes (gametes).

B. They can be Dominant or Recessive.

1. A = dominant a = recessive
2. What would be the genotypes of a male and female that have a Sex-linked Dominant trait and do not express the trait?
   a) Expresses Trait: Male - $X^A Y$ Female - $X^A X^A$ or $X^A X^a$
   b) No Expression: Male - $X^a Y$ Female - $X^a X^a$
3. What would be the genotypes of a male and female that have a Sex-linked Recessive trait and do not express the trait?
   a) Expresses Trait: Male - $X^a Y$ Female - $X^a X^a$
   b) No Expression: Male - $X^A Y$ Female - $X^A X^A$ or $X^A X^a$ (Carrier)
4. Most Sex-linked traits are Recessive.