APPLICATIONS and Complications OF DNA

DNA Warm Up - Interactive Website

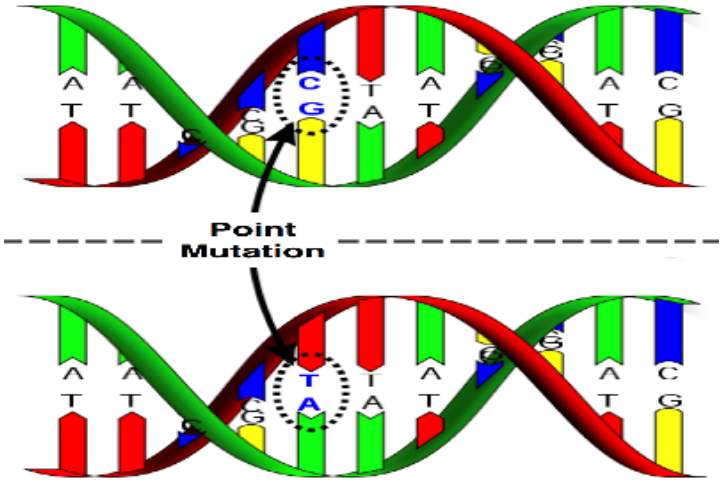
Mutations

A-T, C-G is desired

- The specific protein that a gene codes for depends on the order of the DNA bases (A, C, T, G) in the gene.
- Any change in the usual order of the A, C, T, G bases is called a **mutation**.

A mutation in a gene can alter the structure of the protein it produces.

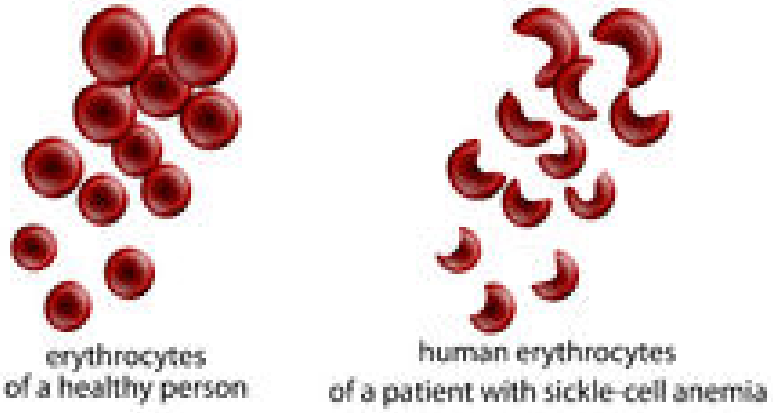
Such a change can then affect how well the protein does its job.

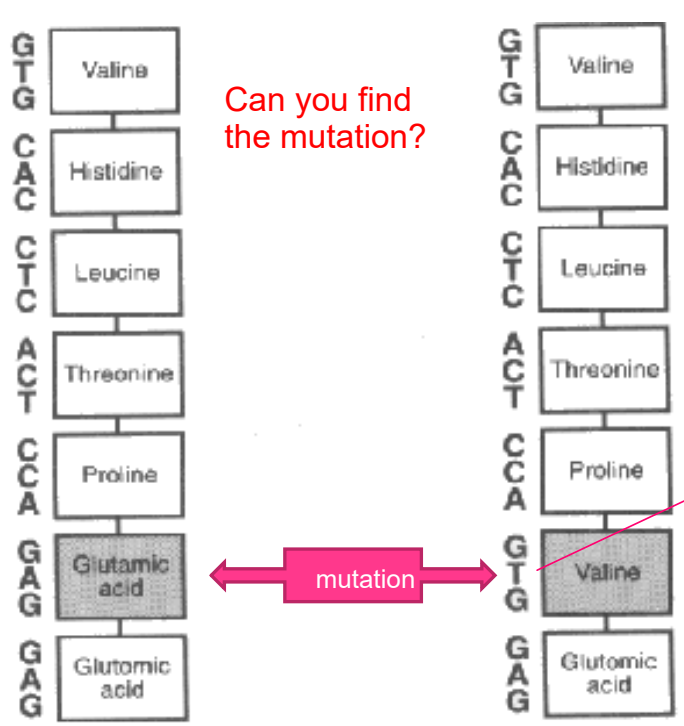


Example:

People who have a condition called sickle cell anemia have a mutation in the gene that codes for hemoglobin, a type of protein in red blood cells.

The mutation changes the protein in such a way that it is less able to perform its function, which is to help the blood carry oxygen to body cells.





Can you find the mutation?

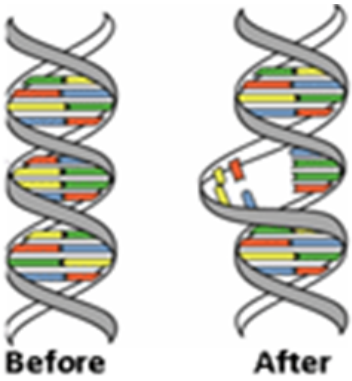
The sickle cell mutation. One amino acid out of a total of 287 differs in sickle cell haemoglobin compared with normal haemoglobin.

Substitution

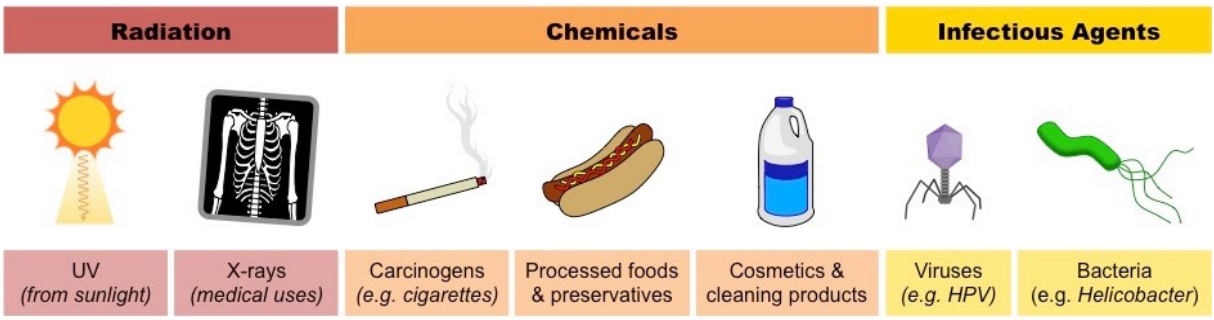
Sickle cell anemia causes chronic anemia, joint pain, and crescent-shaped red blood cells.

- Many mutations can be caused by a **mutagen**.

  A substance or factor that can physically damage and mutate DNA



- Mutagens include:
  - Radiation such as X rays and UV rays
  - Chemicals such as mercury, tar in cigarettes
  - Infectious agents including certain viruses and bacteria



Did You Know?...


Butane
Lighter Fluid
Toluene
Industrial
Solvent
Nicotine
Insecticide
Acetic Acid
Vinegar
Methanol
Rocket Fuel
Hexamine
Barbecue
Lighter
Stearic Acid
Candle Wax
Ammonia
Toilet Cleaner
Benzene
Petrol Fumes
Carbon
Monoxide
Arsenic
Poison
Cadmium
Batteries
Acetone
Paint
Stripper/
Nail Varnish

- Not all mutations are harmful.
- Some mutations may help an organism adapt to a particular environment.

  Example: mutations in bacteria that allow them to resist antibiotics

- Other mutations may be neutral – neither causing harm nor helping the organism.

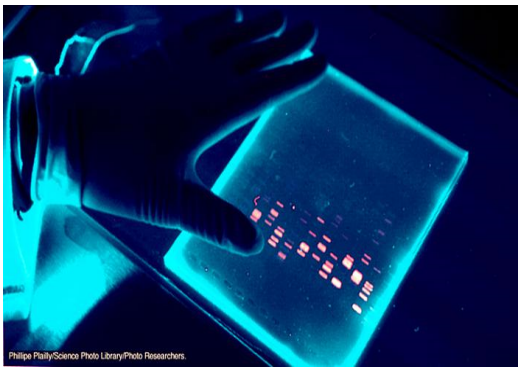
Not all mutations are passed from one individual to their offspring. An individual suffering from skin cancer, often caused by sun's UV, will not pass that mutation on to their children.

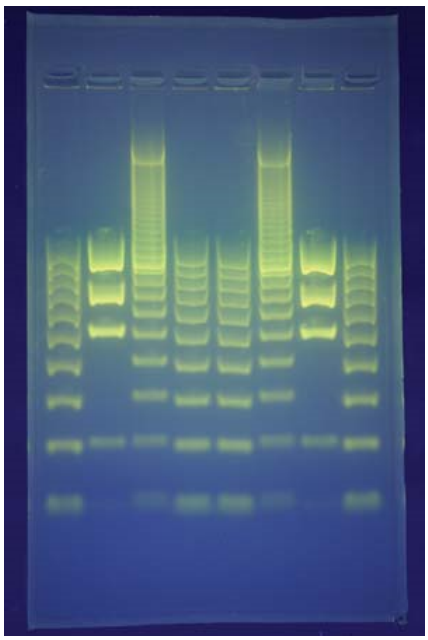
Mutations Link

Applications

DNA Fingerprinting

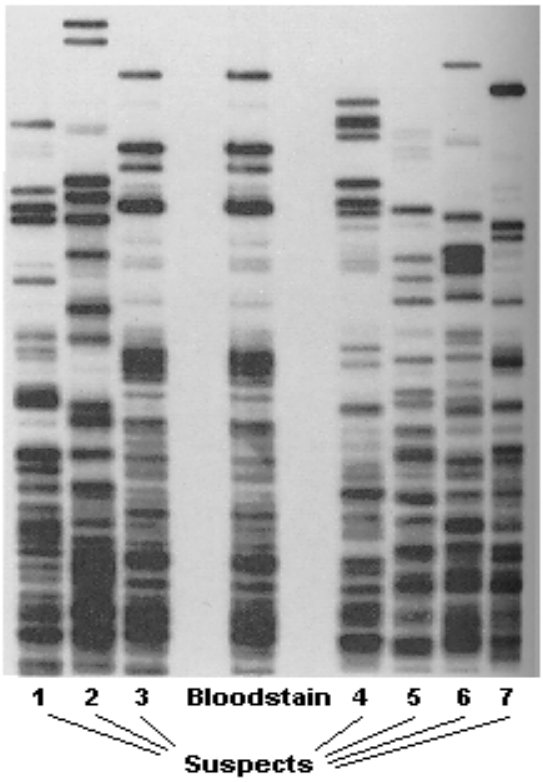
- DNA Fingerprinting (also known as DNA sequencing) is a method of isolating and identifying a specific nucleotide sequence in the DNA of an individual.
- In DNA fingerprinting, fragments of DNA are separated on a gel, producing a specific pattern of bands that is unique to that individual.





Applications of DNA Fingerprinting

- An early, and well known, application of DNA fingerprinting is in criminal identification and forensics to help solve crimes.



- Many new practical applications have been made, including:
  - Determining the identify of biological parents and siblings
  - Helping find cures to genetic diseases by pinpointing segments of genes that cause the disease
  - Mapping the human genome
  - Engineering drought-resistant plants to increase crop production in Agriculture
  - Producing biological drugs from genetically altered bacteria
  - Detecting genetic diversity in various herbs and plants for medical uses

  ALL genetic applications should undergo a rigorous ethical screening process, but few do. Just because something CAN be done, doesn't mean it SHOULD be done.

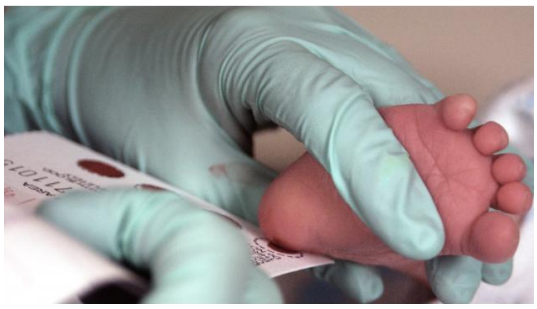
DNA Screening

- Testing for the presence of genetic disorders is referred to as *genetic screening*, or **DNA screening**.
- Some types of genetic disorders can be detected by examining a blood sample.

  In such tests, the presence or absence of specific proteins in the blood can indicate whether a person's genes are functioning normally.

One example of a genetic condition that can be detected using this type of testing is PKU (phenylketonuria).

In Canada, babies are normally tested soon after birth for this condition which, if uncorrected, can lead to severe brain damage.



Another disorder that is detected using DNA screening is Huntington disease.

Huntington disease is a genetic disorder that affects nerve cells.

Symptoms, which normally appear in a person's 40s, include:

- loss of muscular control
- loss of brain function

The symptoms worsen for approximately 15 years before the disease causes death.

Scientists have identified which gene causes this disease. Individuals who have the gene will, with certainty, develop the disease. Therefore, finding out if the gene is present in a person leads to a definite diagnosis.

Should Individuals at risk get tested?

There is an ongoing debate whether individuals at risk of Huntington disease (that is, people who have a parent with the disease) should get tested.

Arguments "against" include:

- diagnosing the disease can cause people needless emotional pain – since there is no cure
- testing is too expensive, especially since it will not save lives

Arguments "for" include:

- diagnosing reduces the stress of uncertainty
- knowing whether a person has or does not have the gene might change many of their life decisions

So…To Test or Not to Test?

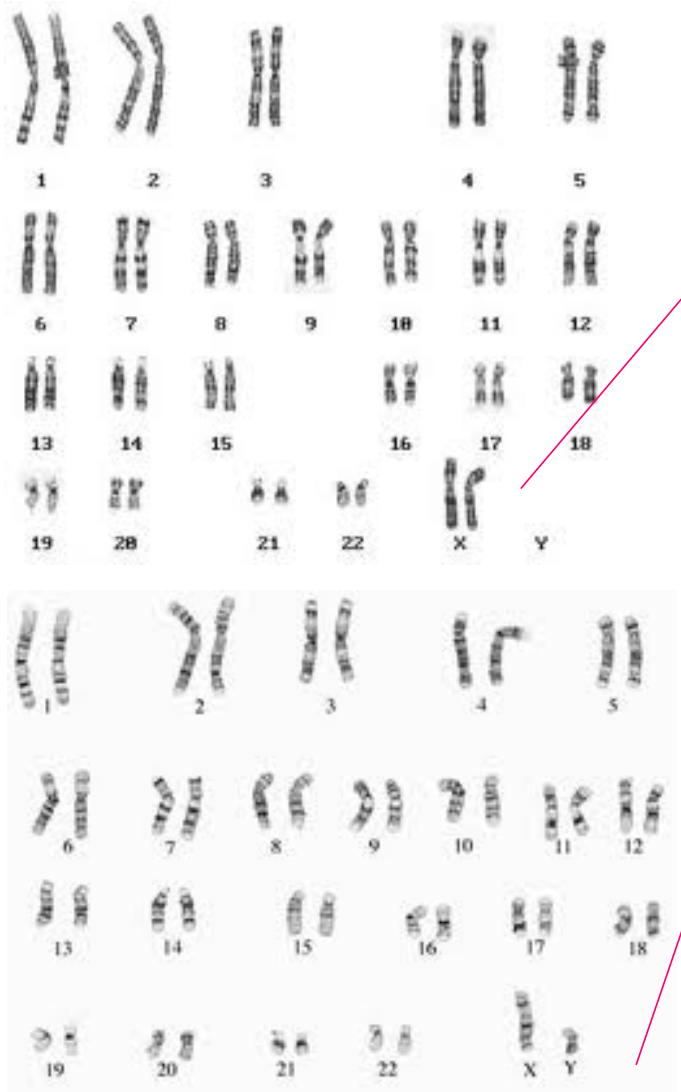
Karyotypes

- Other types of genetic disorders can be observed by looking at a micrograph of a person's chromosomes.

This micrograph is referred to as a **karyotype** and is developed using a technique called *amniocentesis*.

A needle is inserted through a pregnant woman's abdominal wall to withdraw a sample of fluid from the amniotic sac. Cells from the fetus are then isolated and a micrograph of the chromosomes in these cells is taken.

Normal human karyotype



Is this a male or female?

FEMALE – 23rd chromosomes (sex chromosomes) are XX

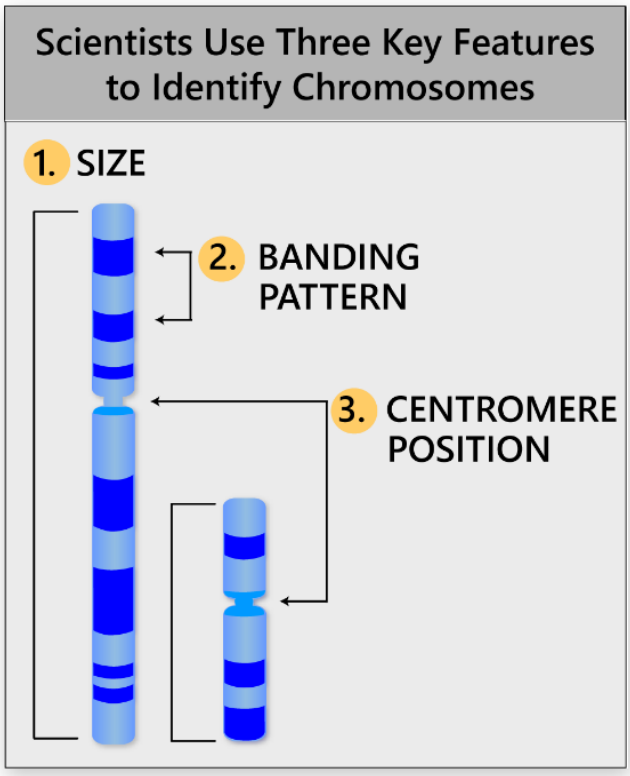
MALE – 23rd chromosomes (sex chromosomes) are XY

How do Scientists Read Chromosomes?

To "read" a set of chromosomes, scientists use three key features to identify their similarities and differences:

1. **Size.** This is the easiest way to tell chromosomes apart.
2. **Banding pattern.** The size and location of Giemsa bands make each chromosome unique.
3. **Centromere position.** Centromeres appear as a constriction. They have a role in the separation of chromosomes into daughter cells during cell division (mitosis and meiosis).

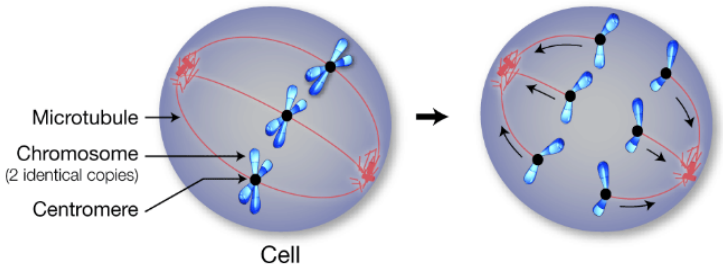
Using these key features, scientists can identify all 46 chromosomes one set of 23 from each parent



What are centromeres for?

Centromeres are required for chromosome separation during cell division. The centromeres are attachement points for microtubules, which are protein fibers that pull duplicate chromosomes toward opposite ends of the cell before it divides. This separation ensures that each daughter cell will have a full set of chromosomes.

Each chromosome has only one centromere.



During cell division, microtubules attach to centromeres and pull the chromosomes to opposite ends of the cell.

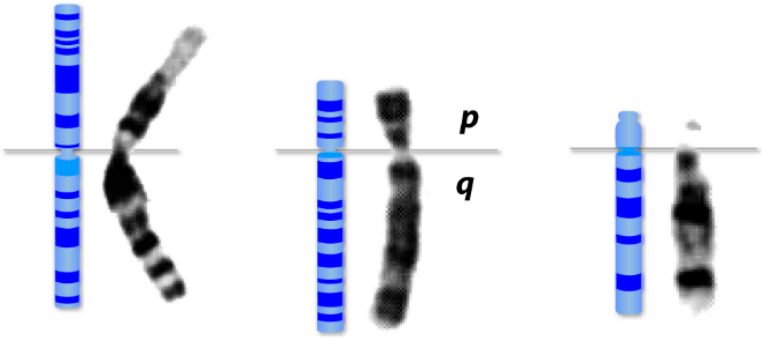
Centromere Positions

The position of the centromere relative to the ends helps scientists tell chromosomes apart. Centromere position can be described three ways: metacentric, submetacentric or acrocentric.

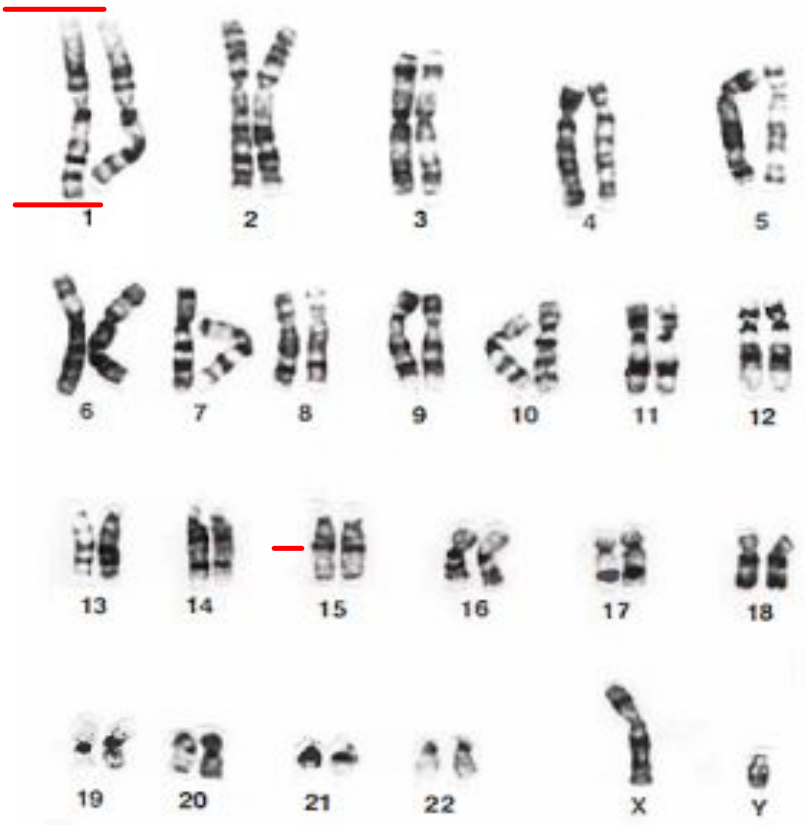
In **metacentric** (met-uh-CEN-trick) chromosomes, the centromere lies near the center of the chromosome.

Submetacentric (SUB-met-uh-CEN-trick) chromosomes have a centromere that is off-center, so that one chromosome arm is longer than the other. The short arm is designated "p" (for petite), and the long arm is designated "q" (because it follows the letter "p").

In **acrocentric** (ACK-ro-CEN-trick) chromosomes, the centromere is very near one end.



Karyotype for an individual with no abnormalities:



Although a karyotype cannot show errors in individual genes, it can show if a person has too many or too few chromosomes, or if any are broken.

An example of a genetic disorder diagnosed using a karyotype includes Down syndrome.

Make a Karyotype

GENETIC DISORDERS AND HUMAN KARYOTYPES –lets explore

Link

Task - Ethical and Moral Decisions