

Genetic Disease

It only takes one gene!

Mutations



Mutation

- A change in the order of bases in DNA
- Can cause a harmful effect (cancer, asthma), beneficial effect (immunity to malaria, evolution), or a neutral effect (not noticeable)
- Caused by mutagen (UV radiation, chemical) or random mistake when DNA is copied



Mutations

Types of Mutations

Normal gene

AS THE MAN SAW THE DOG HIT THE CAN END IT IS

Point mutation

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Deletion

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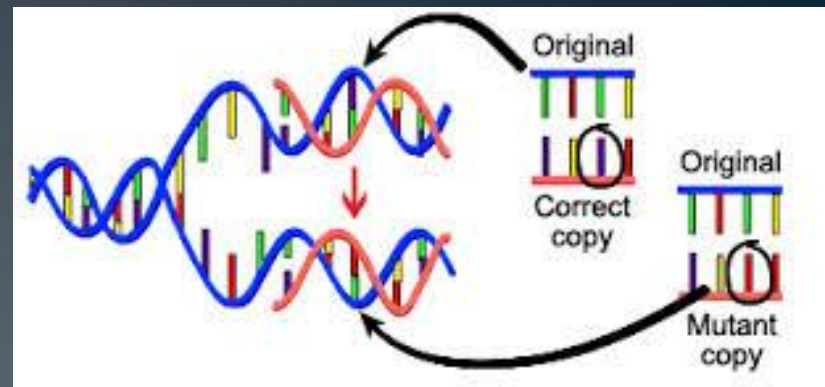
Insertion

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Frame Shift

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Genetic Disease



- A disorder caused by a mutation in an organism's DNA
- May be inherited from a parent
- May not be inherited and results from a new mutation during a person's lifetime
- Most are triggered or worsened by environmental factors (smoking, poor diet, lack of exercise, exposure to dangerous chemicals)

Cancer



- Cells' DNA is mutated causing them to grow and divide abnormally
- Cells do not die normally and begin to grow where not needed (tumor)
- Cancer cells can invade other parts of the body
- Over 100 types of cancer
- 580,350 people died of cancer in the U.S. last year

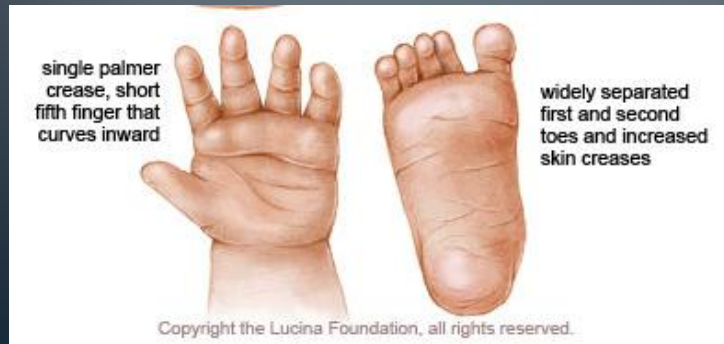
Hemophilia

- Bleeding disorder where your blood does not clot (form a scab)
- People will bleed for longer periods of time and could potentially bleed to death
- More common in males than females because it is a sex-linked trait (X-linked recessive)



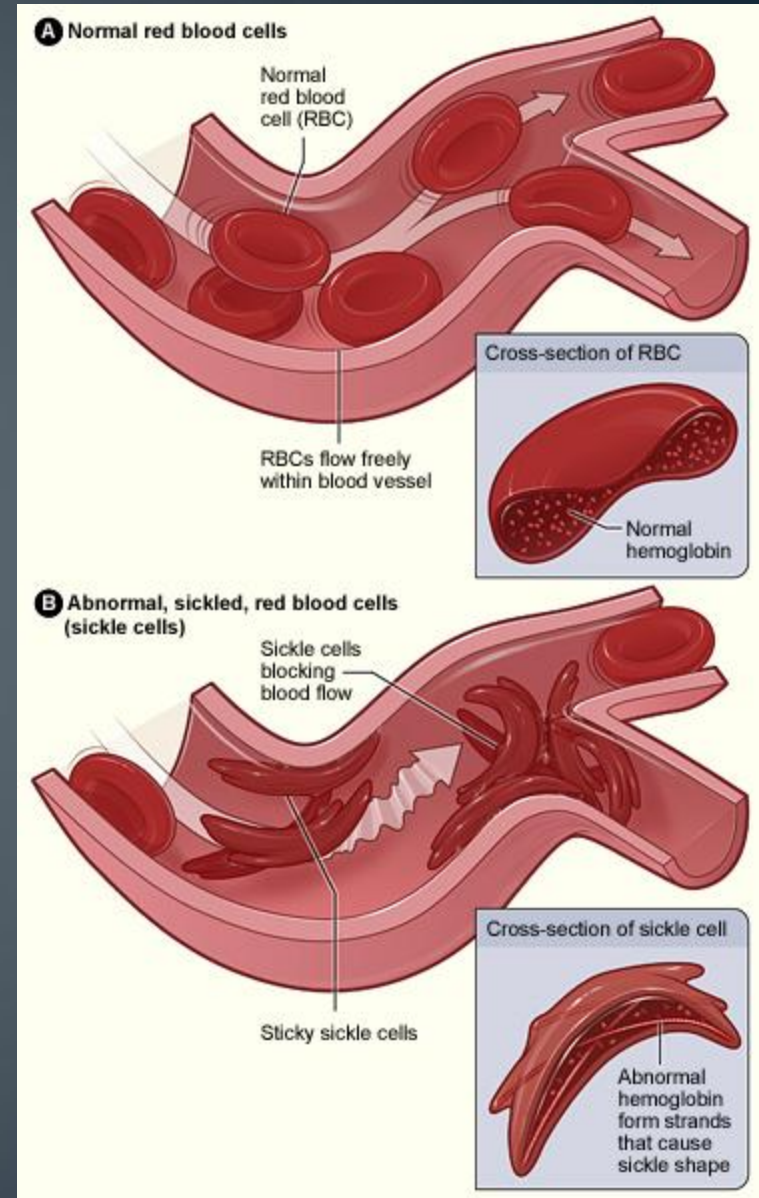
Down Syndrome (Trisomy 21)

- Caused by an extra copy of the 21st chromosome when sex cells are passed to offspring
- Causes significant developmental problems (speech, health, learning)
- Noticeable physical traits (short stature, large tongue, low muscle tone, slanted eyes)



Sickle Cell Anemia

- Serious disorder where body makes sickle-shaped red blood cells. (“Sickle-shaped” = crescent)
- Sickle cell anemia is an inherited, lifelong disease. People who have the disease are born with it. They inherit two recessive genes for sickle hemoglobin—one from each parent.
- Sickle cells are stiff and sticky. They tend to block blood flow in the blood vessels. Blocked blood flow can cause pain, organ damage and raise the risk for infection.



Spina Bifida

- Spina bifida is a birth defect resulting from the bones in the backbone not forming around the spine correctly
- It can be mild or severe
- The cause is unknown but thought to be an environmental factor that causes the genes to not create the cells properly

