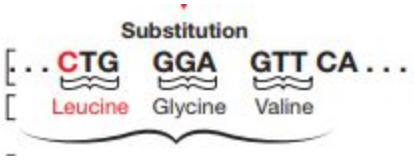
Science 30	Unit A: Biology
Lesson 13 - Mutations and Genetic Diseases	84 mins

Mutations

- The DNA strand changing, making a new different protein

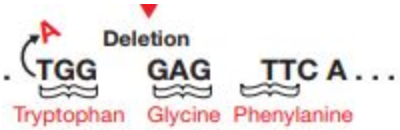
Point Mutations

- A nucleotide is changed for another
  - Substitution



Frameshift Mutation

- A nucleotide is completely removed
  - Deletion



- A nucleotide is added that was never there before
  - Addition

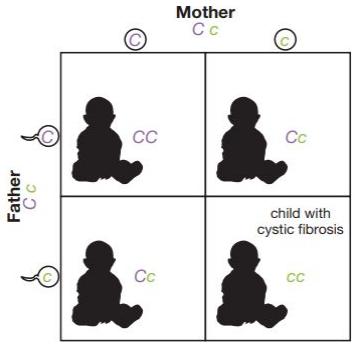
Passing on Mutations

Genetic Disease - a disease caused by a mutation of one or more genes that can be inherited by future generations

cystic fibrosis, colour blindness, sickle cell anemia, Huntington disease

Carrier - an individual who possesses a form of a gene (allele) that results in a disease but does not demonstrate, exhibit, show, or have the symptoms of that disease

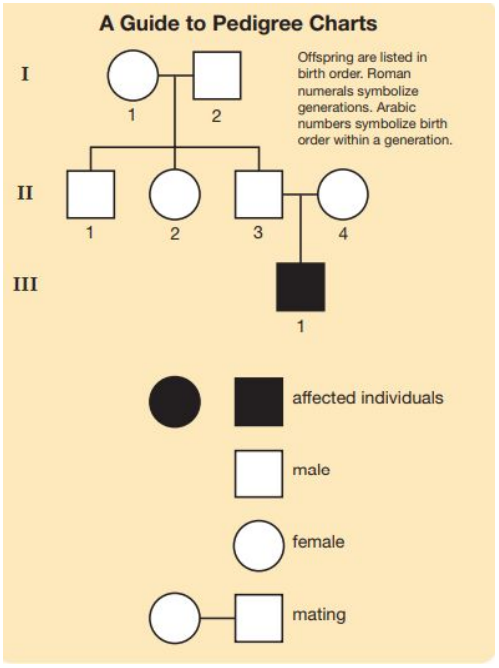
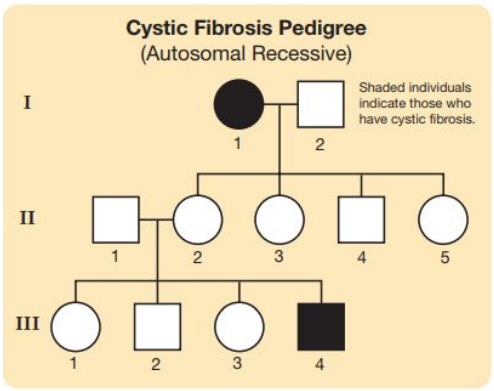
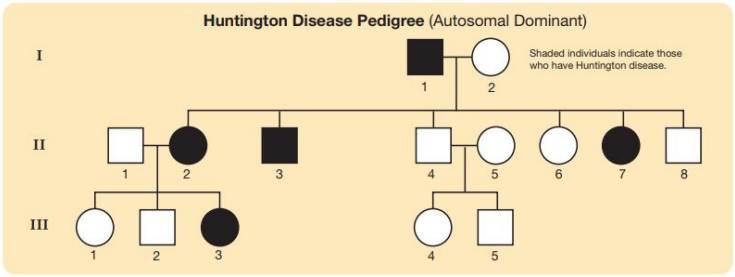
Parents (generally carriers) pass on their genes to the next generation



Factors That Increase Mutations

Mutagens - any agent that increases the likelihood of a mutation **Carcinogen**: any agent that causes the likelihood of cancer to increase	X-rays, UV radiation (tanning), smoking, anything that causes cancer...

Pedigree Charts

- a set of standard symbols used as a tool for geneticists to trace a particular trait 	It is like a genetic family tree.  

Beneficial Mutations

- Mutations in animals and insects for camouflage.	White rabbits in winter, all green insects… bacteria resistance….

Bacteria Resistance

- Bacteria that are not affected by antibiotics	- Occurs when bacteria are not fully treated.. DRAW!!
Transformation in Bacteria - Bacteria gaining DNA - Dead Bacteria	- DNA fragments are taken in and placed in their DNA (from dead bacteria)
- Plasmids	- Plasmids are self replicating pieces of DNA that can be passed between living bacteria… like a virus… but beneficial...

Science 30 - Lesson 13 - Mutations and Genetic Diseases

Name: _____

1) Explain why a doctor may ask questions about the history of certain traits or illnesses in your family

_____

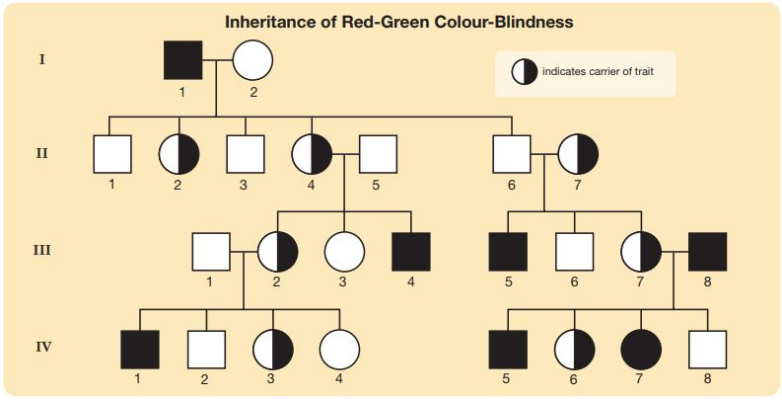
_____

2) Explain how people who are carriers of the allele for cystic fibrosis do not have disease symptoms

_____

_____

3) Red-green colour-blindness is a sex-linked trait. The pedigree in “Inheritance of Red-Green Colour-Blindness” shows the occurrence of the colour-blindness disorder for one family. Study the diagram and answer the following questions.



a) In the first generation, is the father or the mother colour-blind? _____

b) Determine the number of males and the number of females produced by the father and mother of the first generation. _____

c) State the number of individuals in this pedigree who are carriers for colour-blindness. _____

d) How many males and how many females have colour-blindness in this pedigree? _____

4) List two similarities and two differences between Punnett squares and pedigree charts

_____

_____

5) Despite new therapies and other medical breakthroughs, cystic fibrosis and other genetic diseases can still cause death before adulthood. Identify the significance of the symptoms of Huntington's disease not usually appearing until later in a person’s life.

_____

_____

6) Explain why your reproductive organs are usually shielded with lead sheets during an X-ray

_____

_____

Sickle cell anemia is an autosomal recessive genetic disease. The impaired gene causes red blood cells to be produced that are shrunken sickle shapes rather than the normal round disc shapes. These deformed red blood cells can block narrow blood vessels. People with two recessive sickle cell alleles become very ill and often die while they are very young. Most people who suffer from sickle cell anemia or carry the recessive gene tend to be from areas affected by malaria, which is a deadly disease carried by mosquitoes. The malaria parasite reproduces inside a person's red blood cells. People who are carriers of one of the mutated sickle cell anemia alleles actually have an advantage over non-carriers—they are resistant to malaria. This accounts for the fact that this allele is more common in people from areas affected by malaria.

7) Answer the following questions about the information above
   a) Use a Punnett square to show the probable results of the cross between two people who are heterozygous for the sickle cell trait (Ss x Ss) and are malaria resistant.

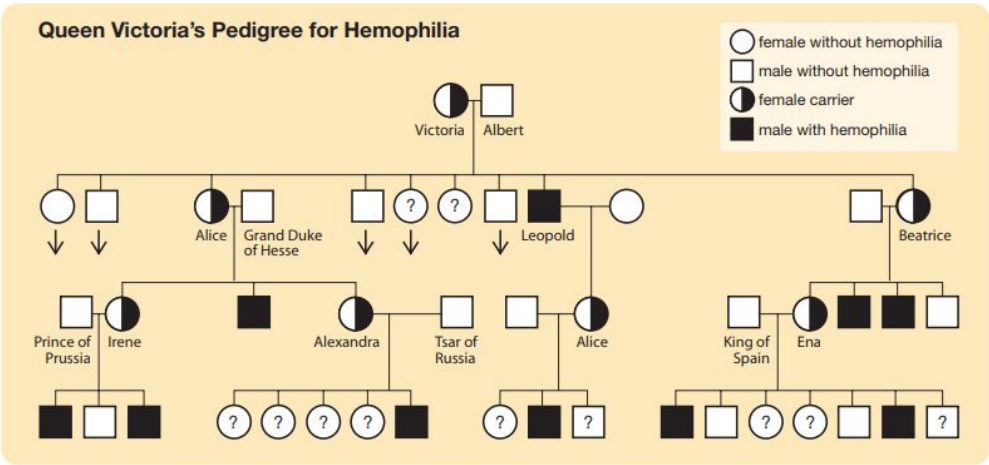
   b) List the possible genotypes of the children from this couple.

   c) State the probability of a child of this couple being resistant to malaria.

   d) State the probability of a child of this couple developing sickle cell anemia.

   e) Explain why the sickle cell anemia allele is more common in areas infected with malaria.

Queen Victoria of England was a carrier of the sex-linked genetic disease called hemophilia. Victoria had many children, but only one of them developed hemophilia. Several of her children married into other European royal families and passed on Victoria's hemophilia allele. The most famous case of hemophilia was Victoria's great-grandson Alexei, the heir to the Russian throne. The controversial historical figure Rasputin gained influence with the Russian royal family by claiming to be able to heal Alexis' hemophilia.



8) Answer the following questions about the information above
   a) The royal family's ancestry is well documented. There is no history of hemophilia in any of Queen Victoria's ancestors. Explain how you think the hemophilia defect appeared in her genes.

   b) Explain why the individuals with hemophilia in this pedigree are all males.

   c) Why are question marks written on some of Queen Victoria's female descendants?