Sample Diploma Question

Use the following information to answer question 23.

Gene segments may initiate, terminate or code for a particular amino acid sequence.

Gene Segment

A G C T A G

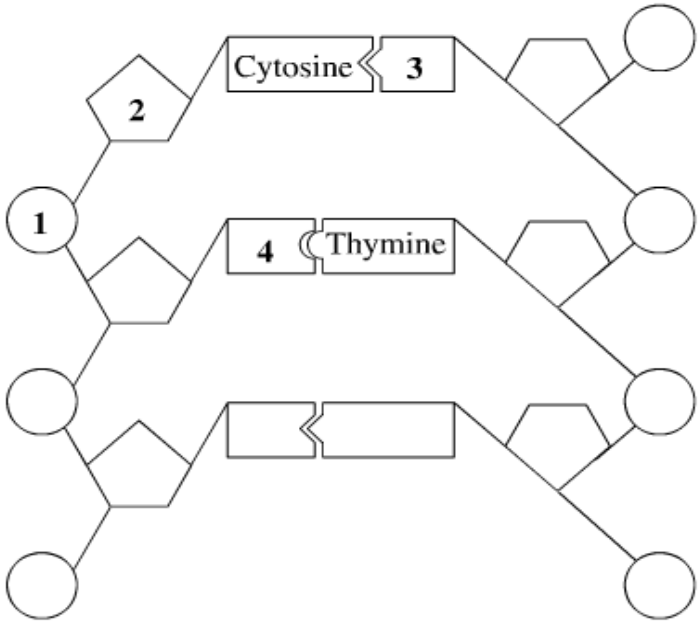
TAG is stop (not an amino acid)

23. The number of amino acids that the gene segment shown above codes for is

A. 1
B. 2
C. 3
D. 6

Use the following information to answer numerical-response question 5.

The structures that make up a section of a DNA molecule are represented in the diagram below.



Numerical Response

5. Match each of the structures numbered in the diagram above with its name given below. (Use each number only once.)

Structure:	4	2	1	3
Name:	**Adenine**	**Sugar**	**Phosphate**	**Guanine**

(Record all **four digits** of your answer in the response boxes at the bottom of the screen.)

Use the following information to answer question 26.

In humans, the *WRN* gene on chromosome 8 produces an enzyme responsible for unwinding and separating the two strands of DNA during replication.

26. Two components of the DNA molecule that the enzyme described above would be responsible for separating are

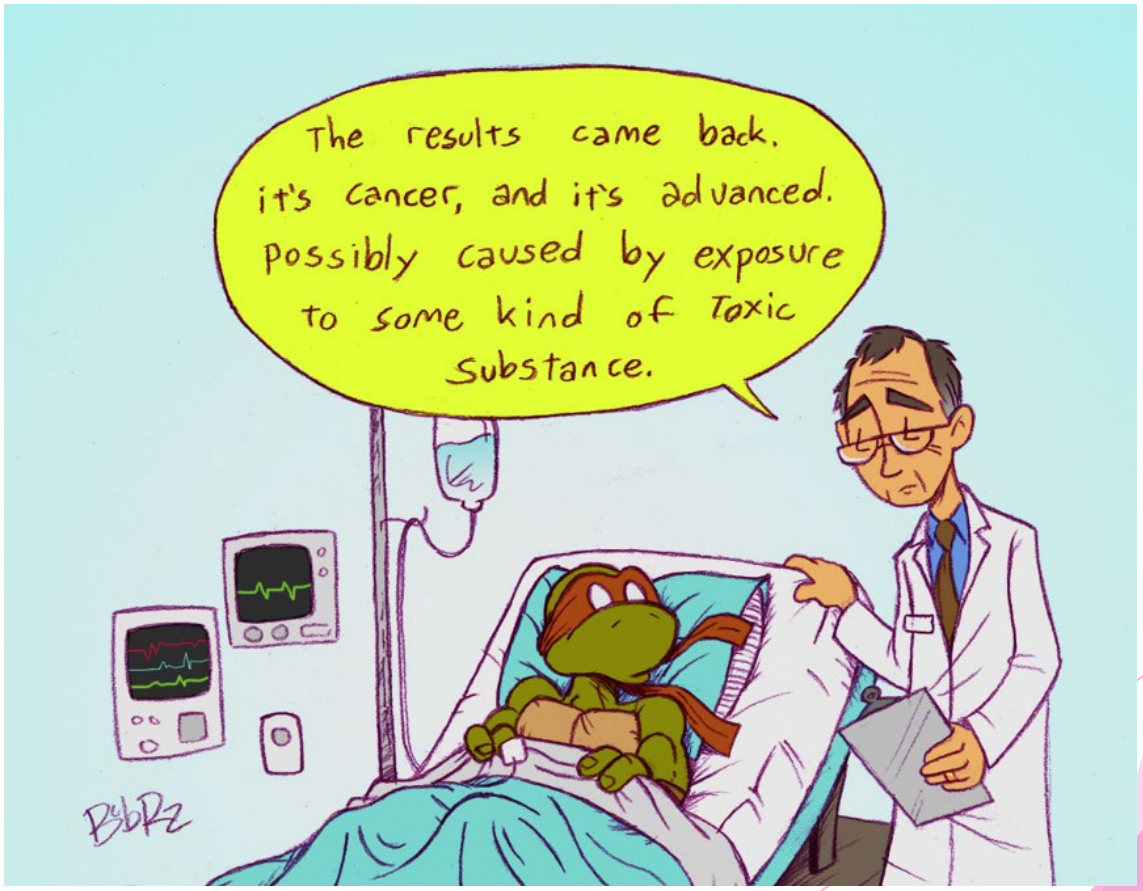
A. adenine and thymine

B. adenine and phosphate

C. deoxyribose and thymine

D. deoxyribose and phosphate

Mutations and Genetic Diseases



Curriculum

- describe how mutations in DNA affect the proteins produced resulting in human diseases
- describe the development of resistance in bacteria and viruses, based on the concepts of mutation, plasmid transfer, transformation and natural selection.
- investigate, with the aid of a pedigree chart, the familial inheritance of a specific trait that is controlled by a single pair of genes
- interpret patterns and trends in data associated with autosomal and sex-linked inheritance

Mutations

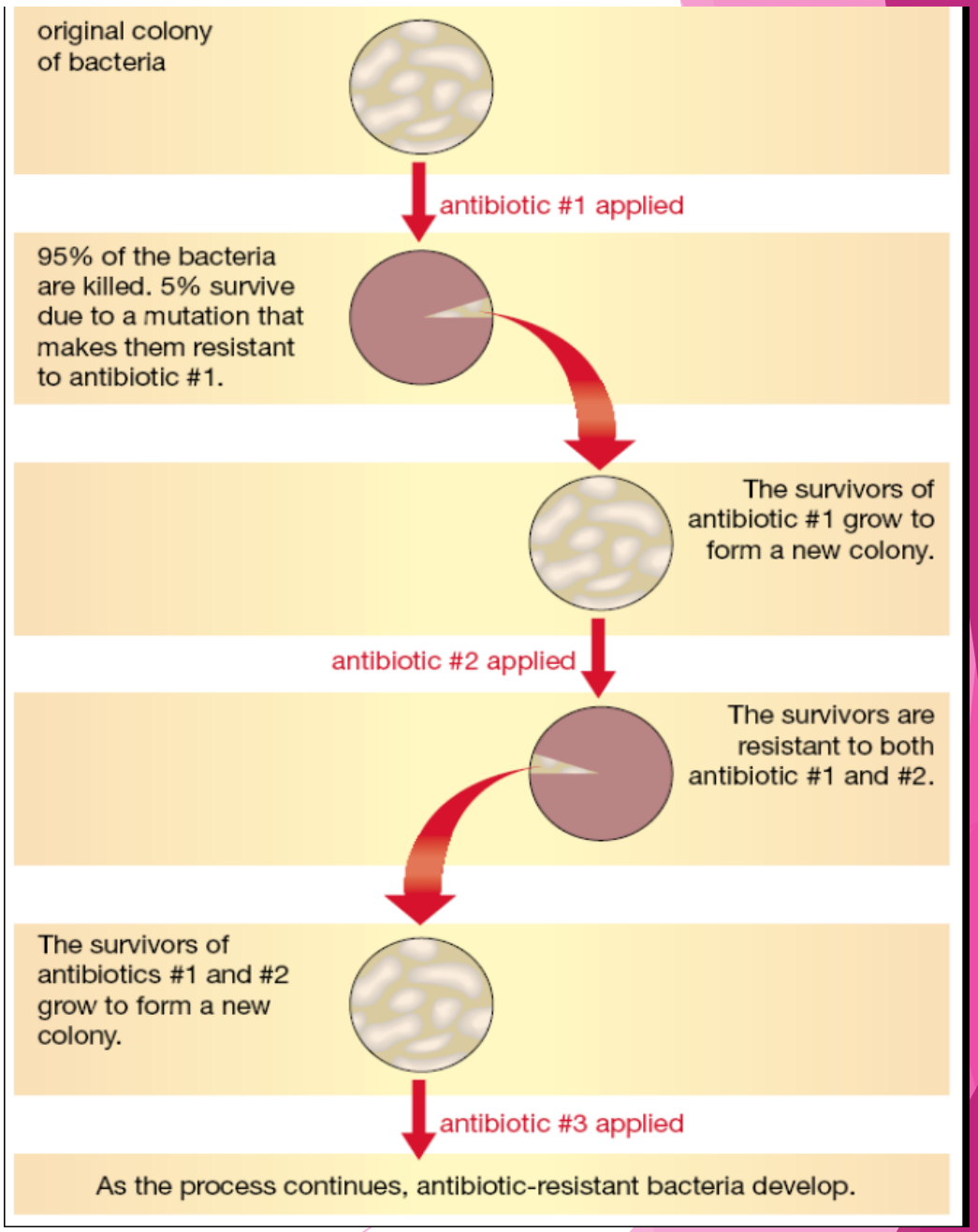
- A **mutation** is a change in the sequence of bases along DNA
- Most occur naturally or due to radiation exposure or chemicals or carcinogens
- Each change is carried to next generation and inherited by offspring
- Mutations increase **variability** and lead to adaptations (beneficial and harmful)

Mechanisms of Mutation

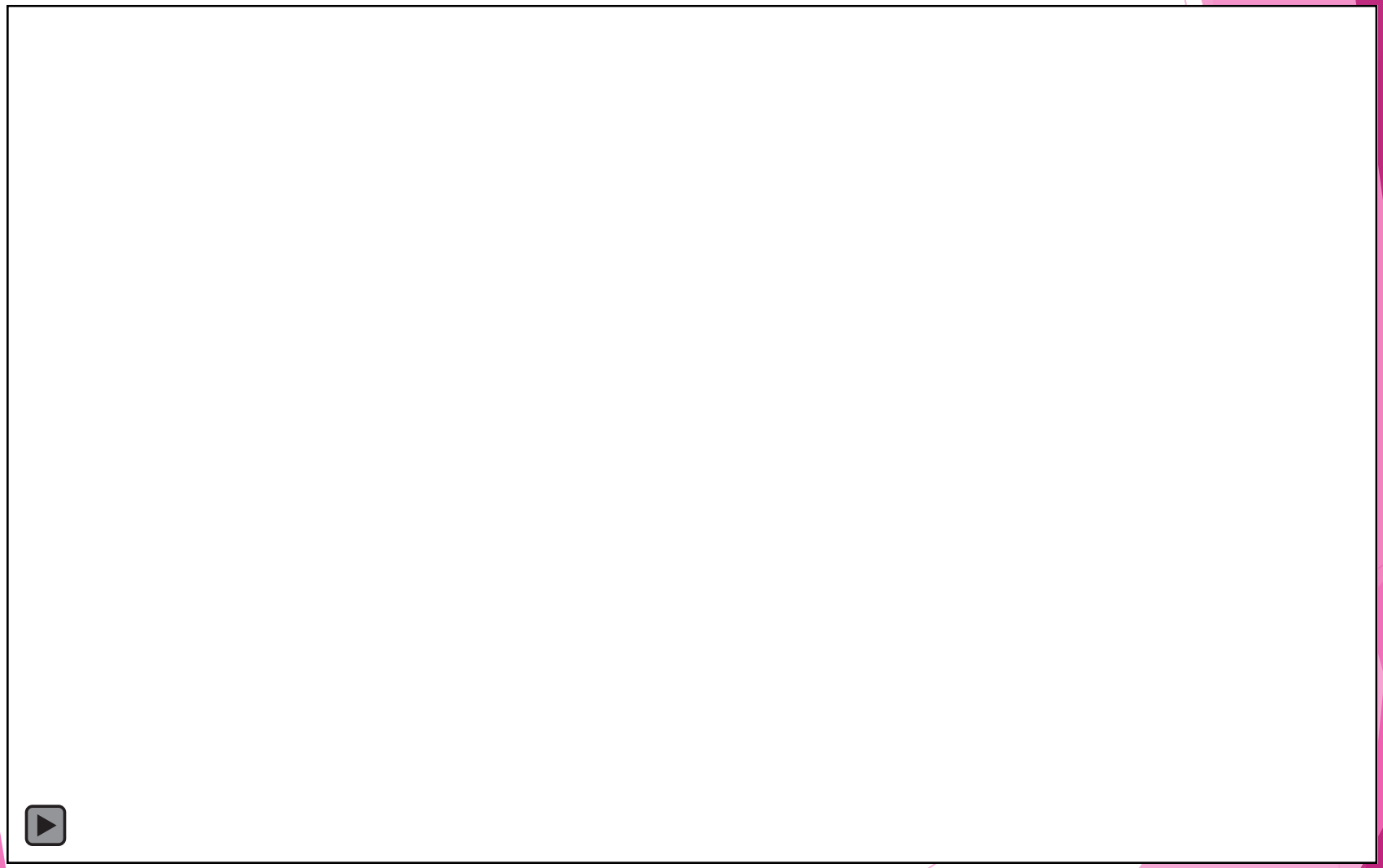
- **Mutagens** are agents that cause mutations
- Mutations are like typos in DNA

- If I spel somthign rong you can stil reed it
- Most mutations are the same way, little mutations have little effect on the product produced
- Mutations usually occur in chromosomal regions where they do not affect the protein; body can repair some mutations
- Some mutations cause gene to stop working or alter function

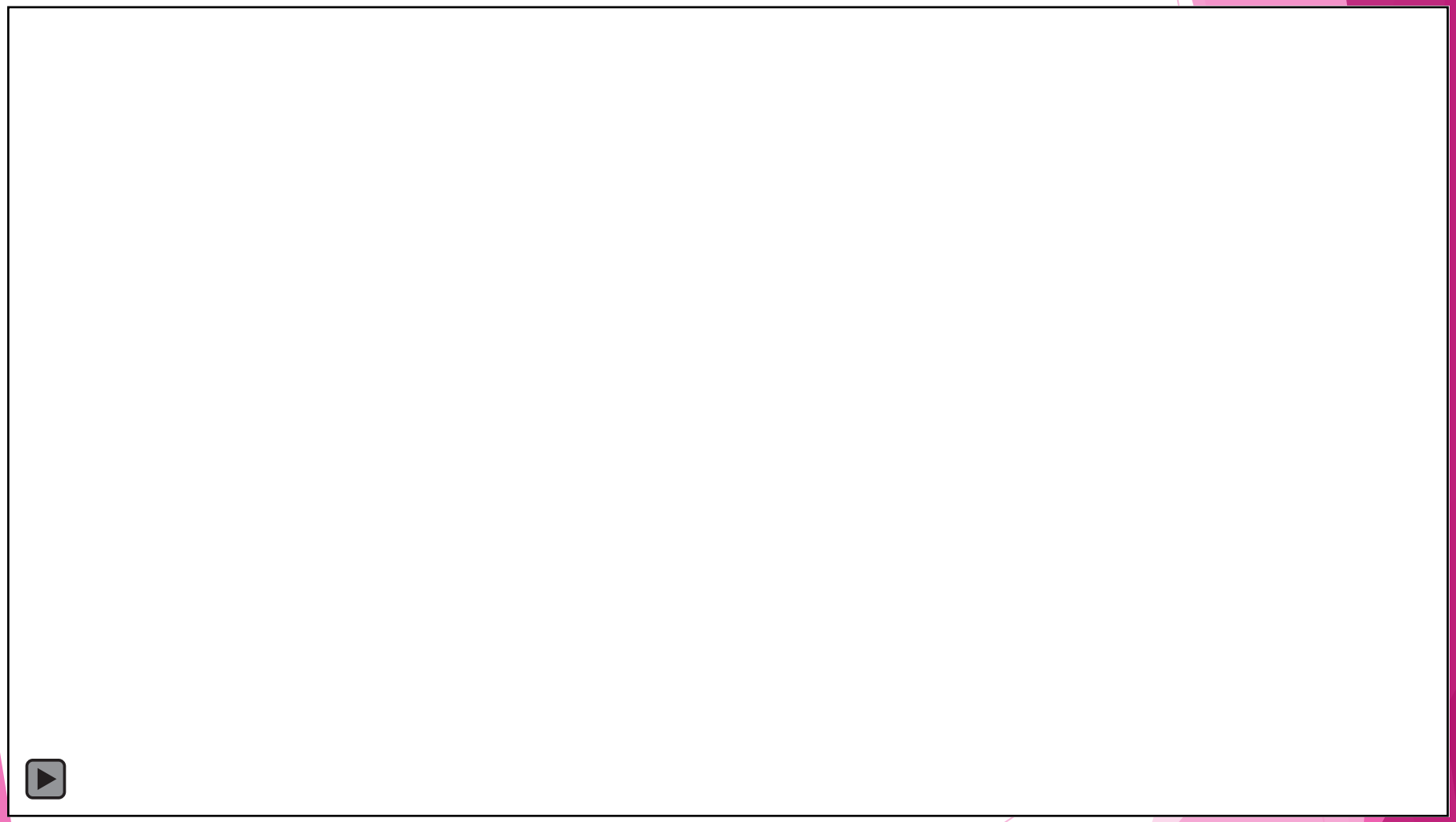
Bacteria evolve to become resistant to antibiotics because of variations caused by mutations



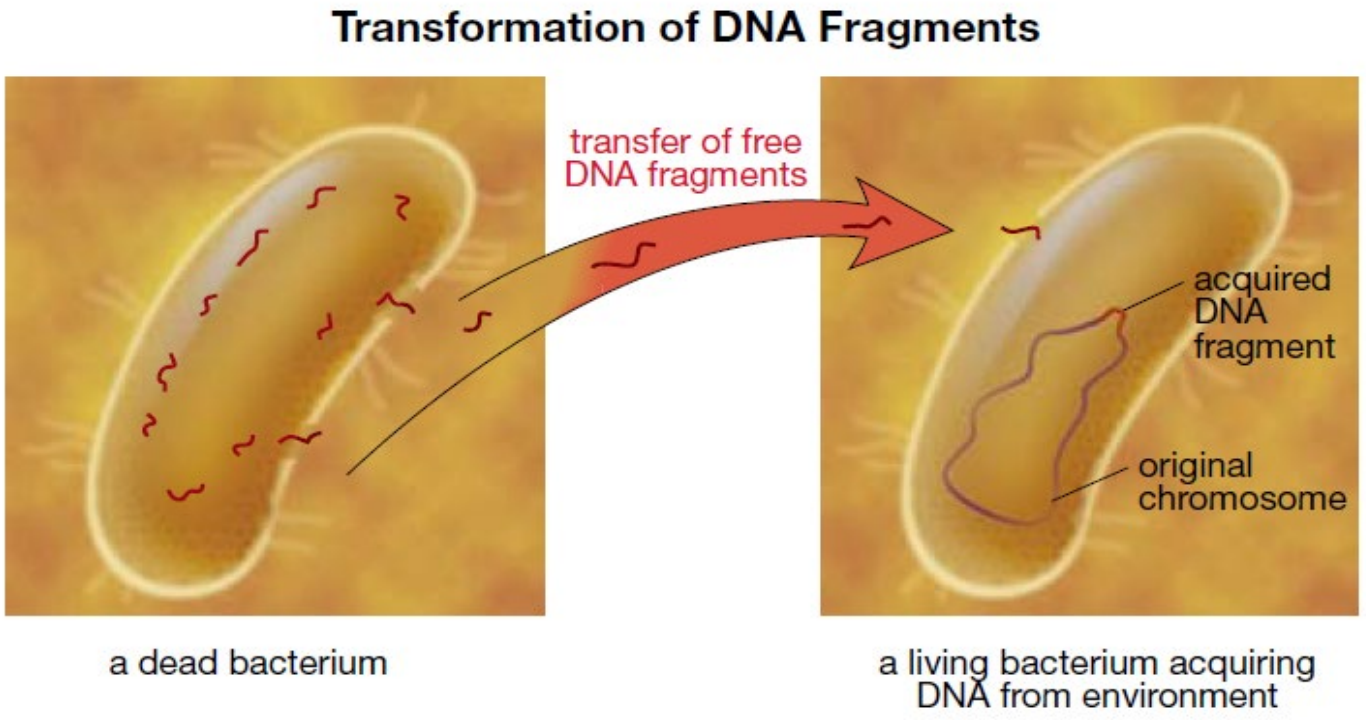
Beneficial Mutations (9 minutes)



Antibiotic Resistance (5 min)



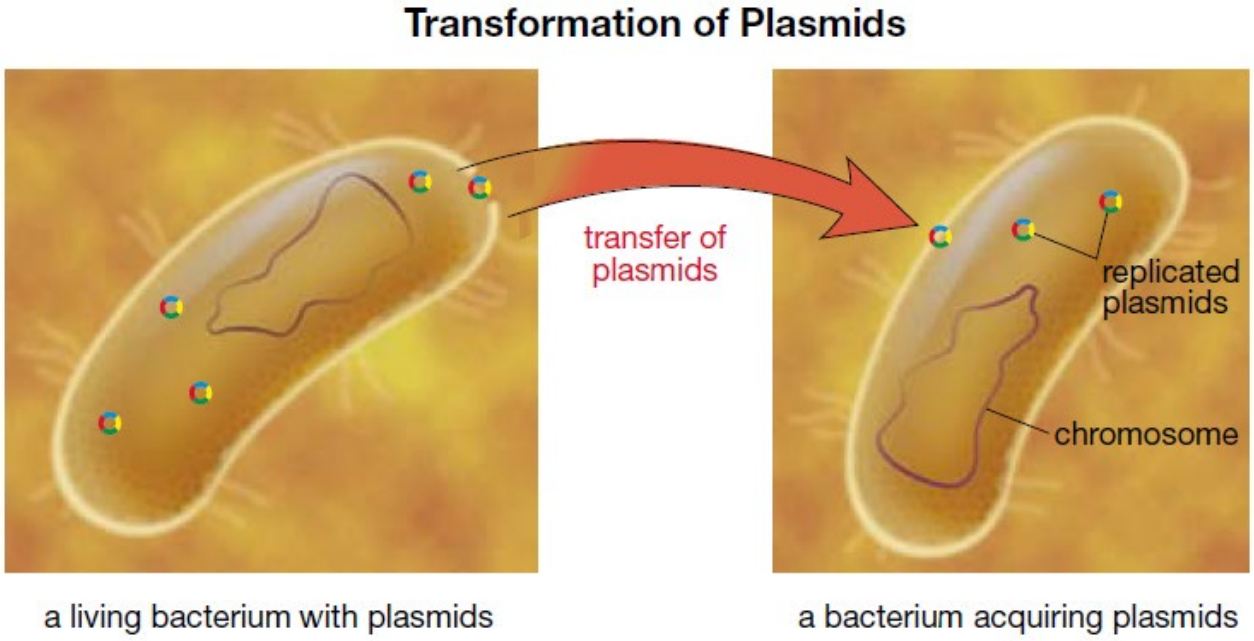
Transformation



- **Transformations** is free DNA being incorporated into bacteria cell

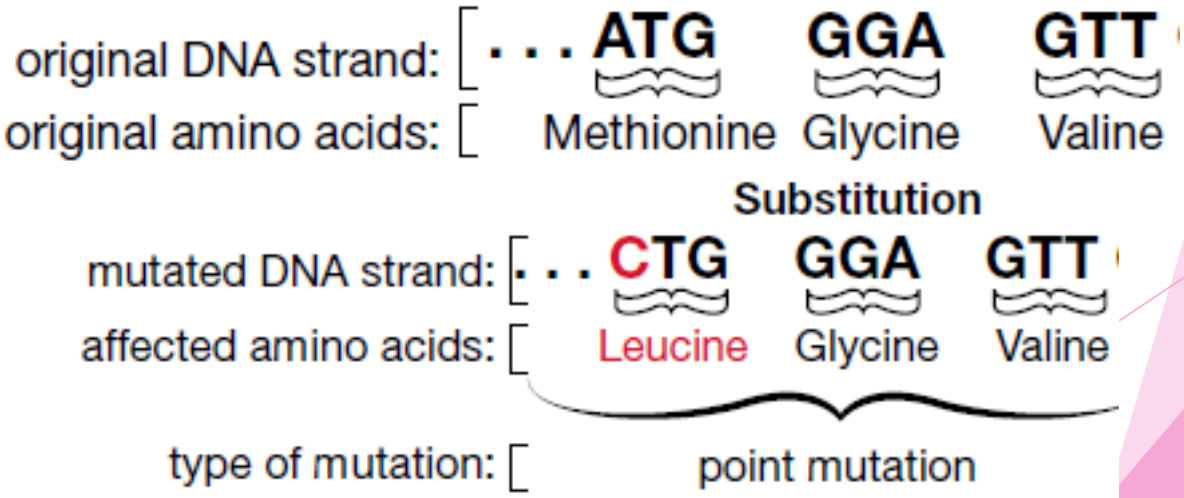
Plasmids

- Bacteria possess a circular DNA molecule called a **plasmid**
- This allows them to transfer DNA between living bacteria and sharing genes



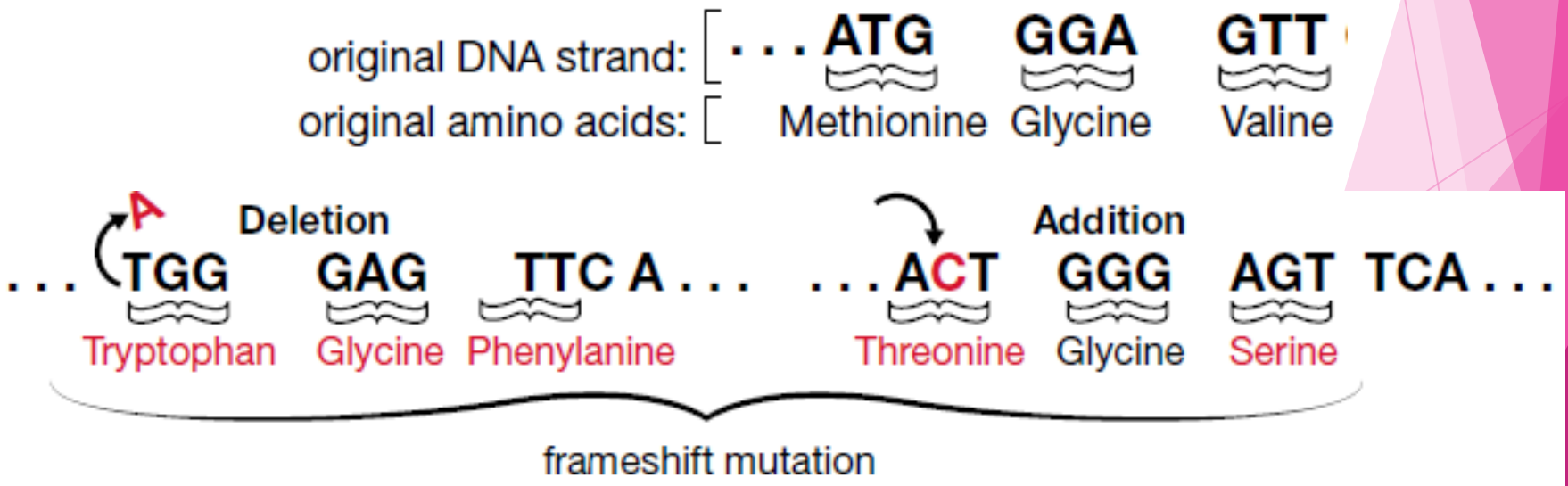
Point Mutation

- **Point mutation** is where one letter of a base is switched with another (like a typo)
- This is similar to changing the phrase “the fox can run” to “the box can run” it changes the meaning

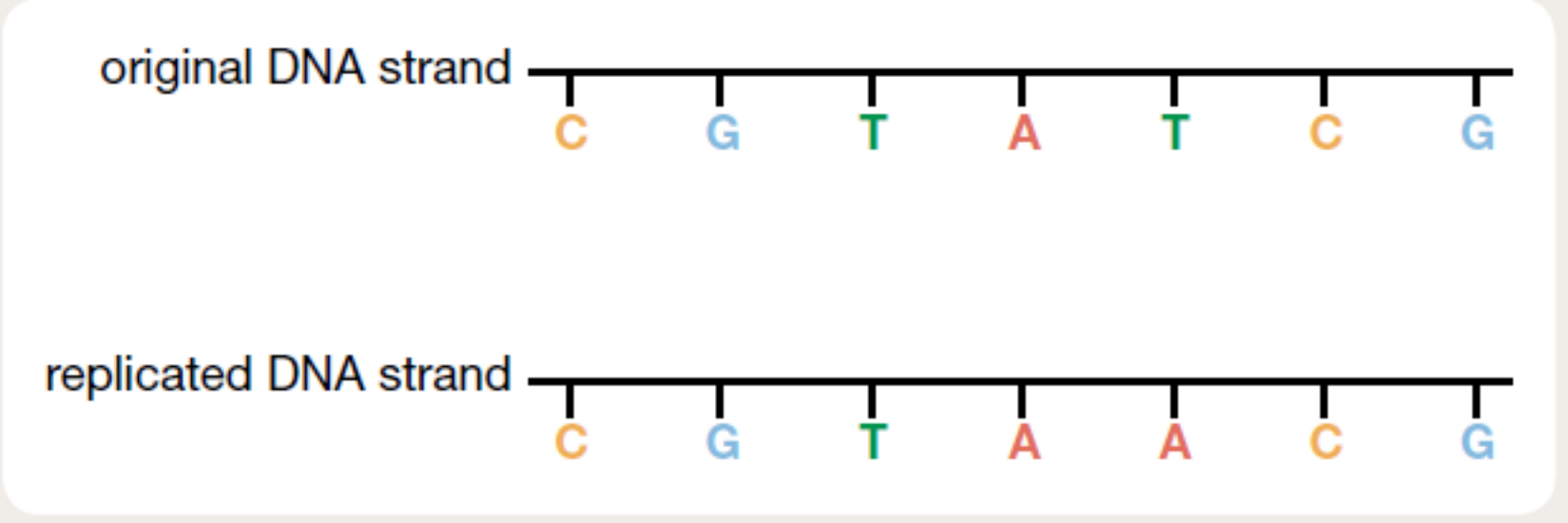


Frameshift Mutation

- Addition or deletion of a base can affect the whole sequence of DNA and is called a **frameshift mutation**
- If the phrase “the fox can run” had an extra letter is could be read as “ath efo xca nru n” which doesn’t make sense



Board Question



- What type of mutation is this? Point mutation, 1 letter changed
- What amino acid corresponds to the TAT in the original? ATA
- What amino acid corresponds to the replicated DNA? ATT

Board Question

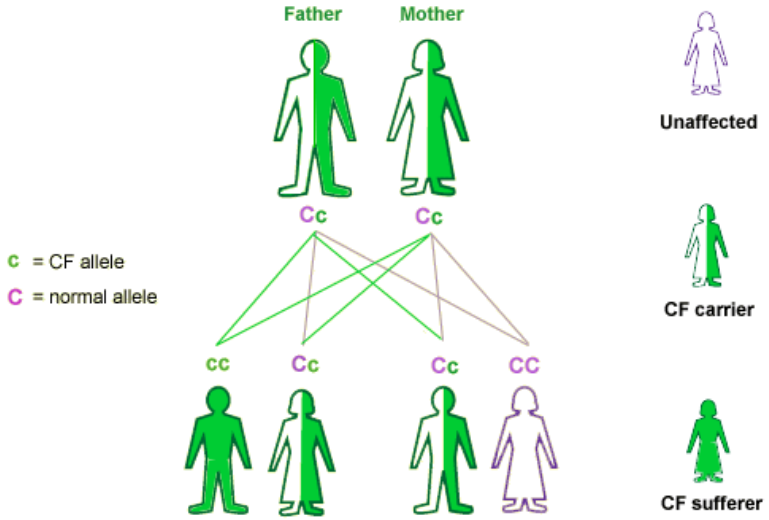
- A gene sequence reads GGATTAGAG. A mutation occurs and the sequence now appears as GGGATTAGAG
  - What type of mutation is this? Frame shift ‘g’ is added in extra
  - Use your table of DNA triplet codes to list the amino acid sequence produced by the original DNA strand
  - Use your table of DNA triplet codes to list the amino acid sequence produced by the new, mutated strand.

Passing on Mutations

- If a mutation is capable of being passed down to offspring it called a **genetic disease**
  - Ex Cystic fibrosis has a mutated chromosome #7 which results in it
- When an offspring receives mutated genes, every cell in their body has this mutation which can lead to early death in many cases
- Having two sets of chromosome help because one parent can supply a non mutated gene with no symptoms
- They are a **carrier** and can pass this disease to their offspring

Mechanism of Inheritance

- If a genetic disease is caused by a recessive allele, then the genotype needed to develop the diseases is homozygous recessive
- If a genetic disease is caused by the dominant allele, then the genotype heterozygous or homozygous dominant is required



Genetic Disease	Symptoms	Location of Gene	Mechanism of Inheritance	Prevalence
cystic fibrosis (CF)	People with CF produce thick sticky mucus that builds up in their lungs and digestive tract. This makes it difficult to properly breathe and digest food. People with CF are also prone to lung infections because they cannot easily clear bacteria from their lungs.	chromosome 7	autosomal recessive	Approximately 1 in 2500 children born in Canada has CF and 1 in 25 Canadians is a carrier of the defective allele that causes cystic fibrosis.
Huntington disease (once called Huntington's chorea)	Huntington disease causes brain cells to die in particular regions. This results in a continual reduction in the ability to control movements, remember events, make decisions, and control emotions. Symptoms usually appear between the ages of 30 and 45.	chromosome 4	autosomal dominant	Approximately 1 in 10 000 Canadians has Huntington disease.
hemophilia	There are two forms of this disease: hemophilia A and hemophilia B. Both forms are caused by a mutation of one of the genes that produces blood-clotting proteins. Both of the genes involved with producing the proteins for blood clotting are found on the *X* chromosome. A defective allele for either of these two *X*-chromosome genes can result in impaired blood-clotting ability. People with hemophilia bleed for a longer time period than people without this condition. Internal bleeding, or hemorrhaging, is a common risk associated with this dangerous condition.	*X* chromosome	sex-linked recessive	Hemophilia A affects about 1 in 10 000 people in Canada and hemophilia B affects as few as 1 in 50 000 people. Because of the sex-linked nature of the disease, males develop the disease more than females do.

Example

- A couple discover that they both have a family history of cystic fibrosis. They are thinking of having a child, and they ask for a genetic test to be done. Both the man and the woman discover that they are carriers of the recessive cystic fibrosis allele
  - Build a Punnett square to describe this cross
  - What is the percentage probability that their child will develop cystic fibrosis?
  - What is the percentage probability that their child will be a carrier of the cystic fibrosis allele?
  - What is the percentage probability that their child will not inherit the cystic fibrosis allele?

Board Question

- A man is heterozygous for the dominant Huntington allele, and he has a child with a woman who does not have a Huntington allele.
  - Write out the genotype for the man and the woman
  - Build a Punnett square to describe this cross.
  - What is the percentage probability that their child will not develop Huntington disease?
  - What is the percentage probability that their child will develop Huntington disease?

Board Question

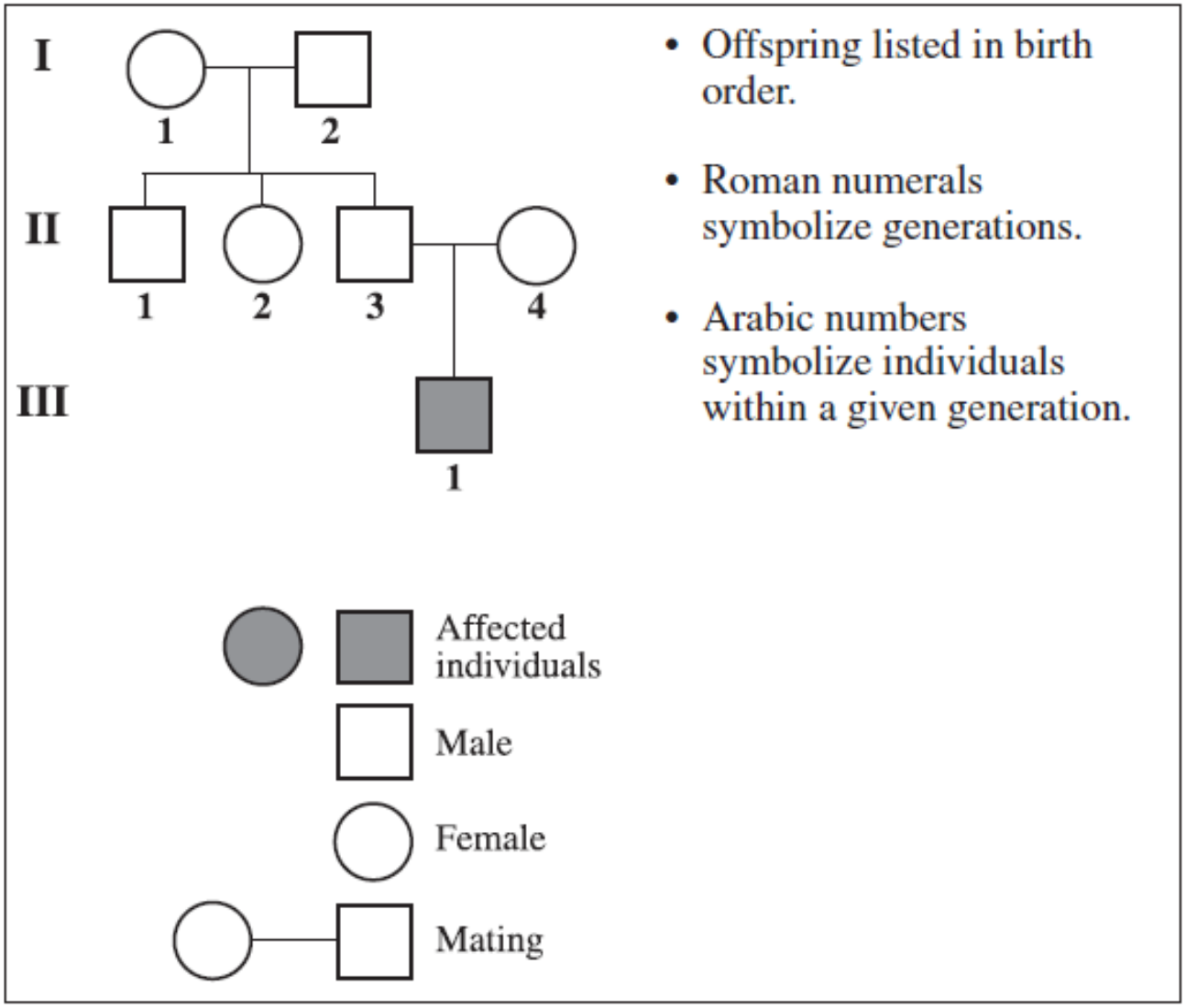
- A woman carries one of the defective recessive alleles on her X chromosome that causes hemophilia. She has a child with a man who does not possess the hemophilia allele.
  - Write out the genotype for the man and the woman
  - Build a Punnett square to describe this cross.
  - What is the percentage probability that she will have a child with hemophilia?
  - What percentage of females born from this cross are likely to have hemophilia?
  - What percentage of males born from this cross are likely to have hemophilia?

Pedigree Charts

- Being able to roll your tongue is a caused by a dominant allele
- If a person can roll their tongue, is it possible to tell if what their genotype is Rr, RR?
- But if you look at family history and see if two parents who can roll their tongues, have a child who can't, both parents must be Rr
- Geneticists use a **Pedigree Chart** to trace family histories

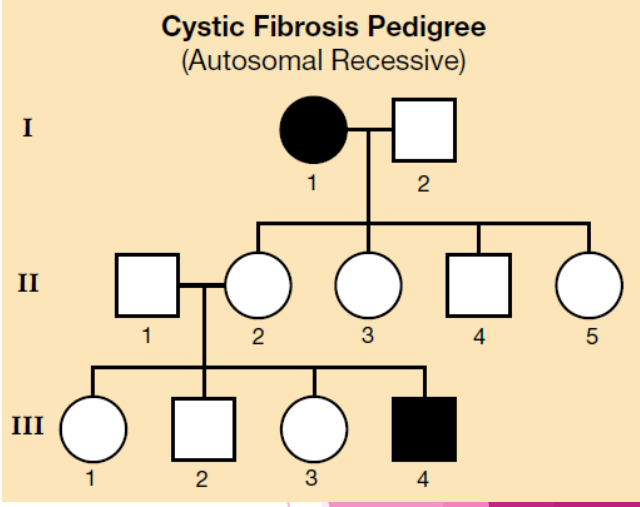
Page 13 of Data Booklet

Pedigree Chart



- Offspring listed in birth order.
- Roman numerals symbolize generations.
- Arabic numbers symbolize individuals within a given generation.

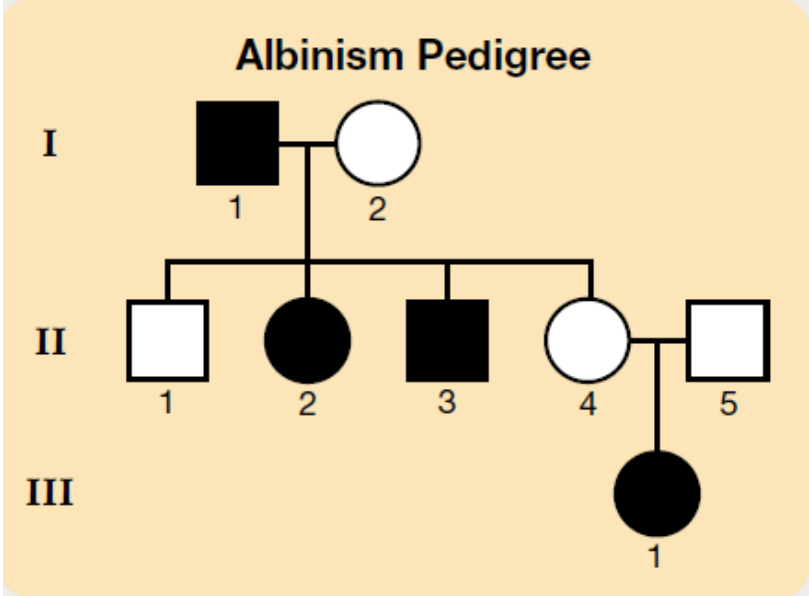
Example



- What do we know about generation I?
- Explain why none of their children have CF.
- How many boys and how many girls are in the second generation?
- Explain why CF showed up again in generation III.

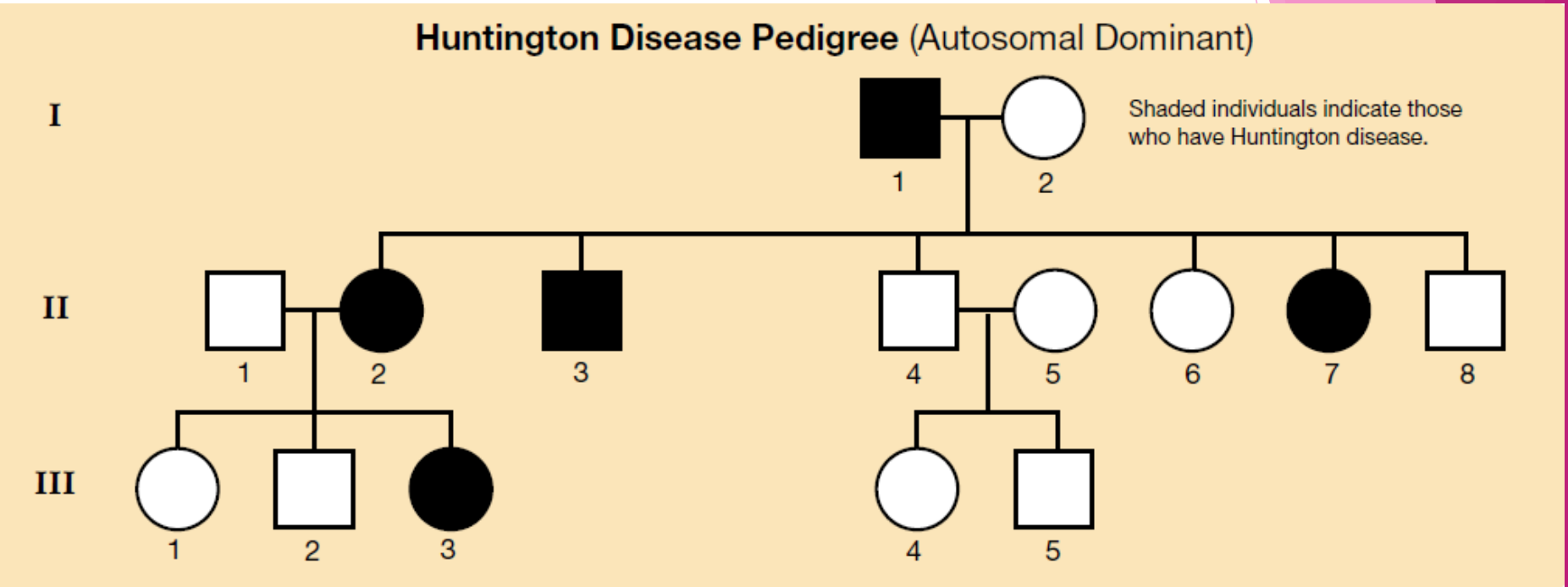
Example

- Albinism is a genetic condition that causes an absence of pigmentation in skin, hair, and eyes. In humans, the most severe form of albinism—called oculocutaneous albinism—is an autosomal recessive genetic disease.



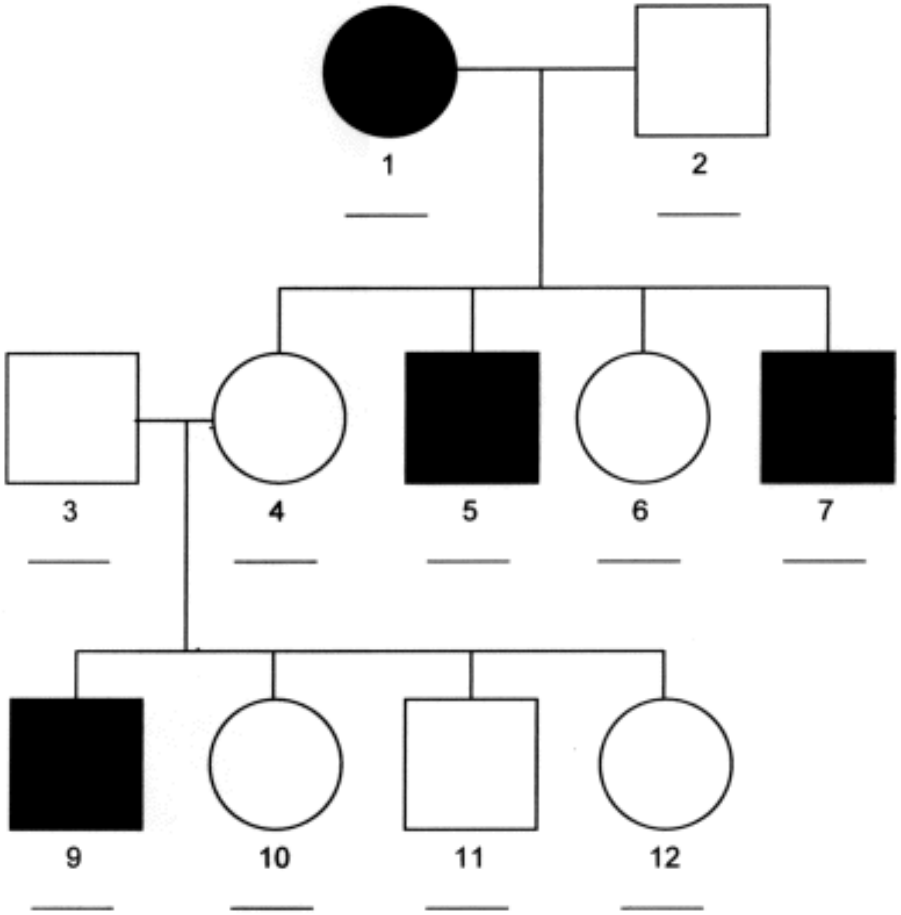
- What are the genotypes for the first generation parents?
- Determine the genotype for all of the offspring

Board Question



- Knowing the Huntington is autosomal dominant, what are the genotypes of the parents in gen I?
- What would be the genotypes for all the generation II offspring?

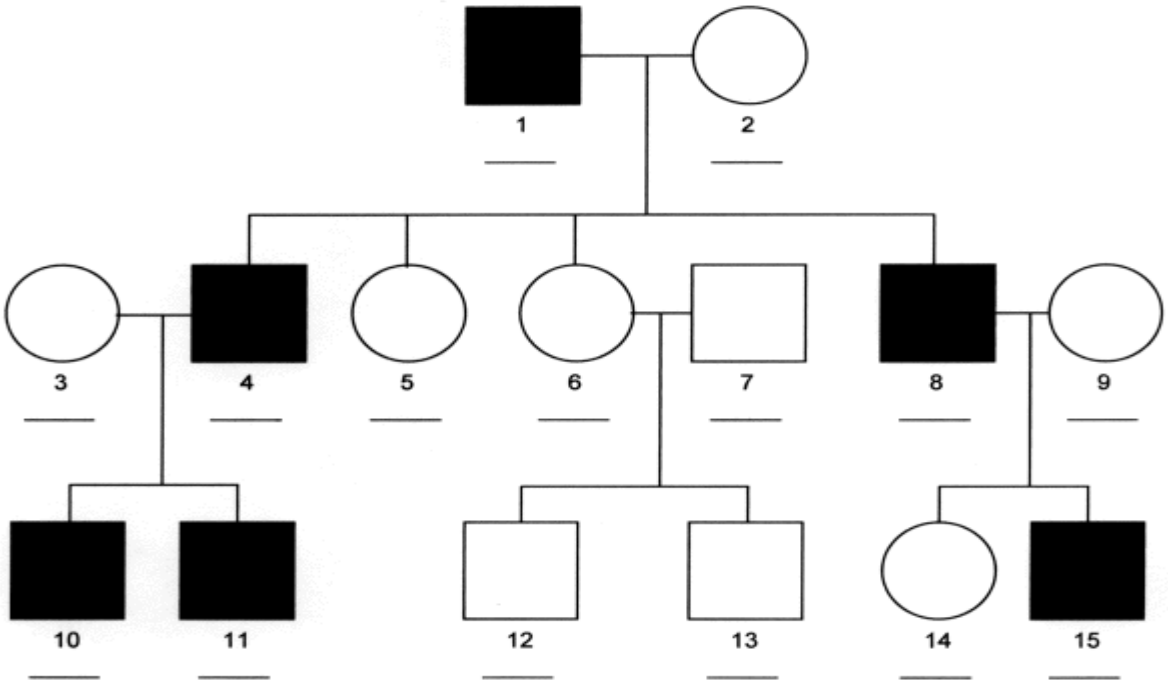
Example



- What do you notice about the offspring who are affected?
- Explain why this is happening

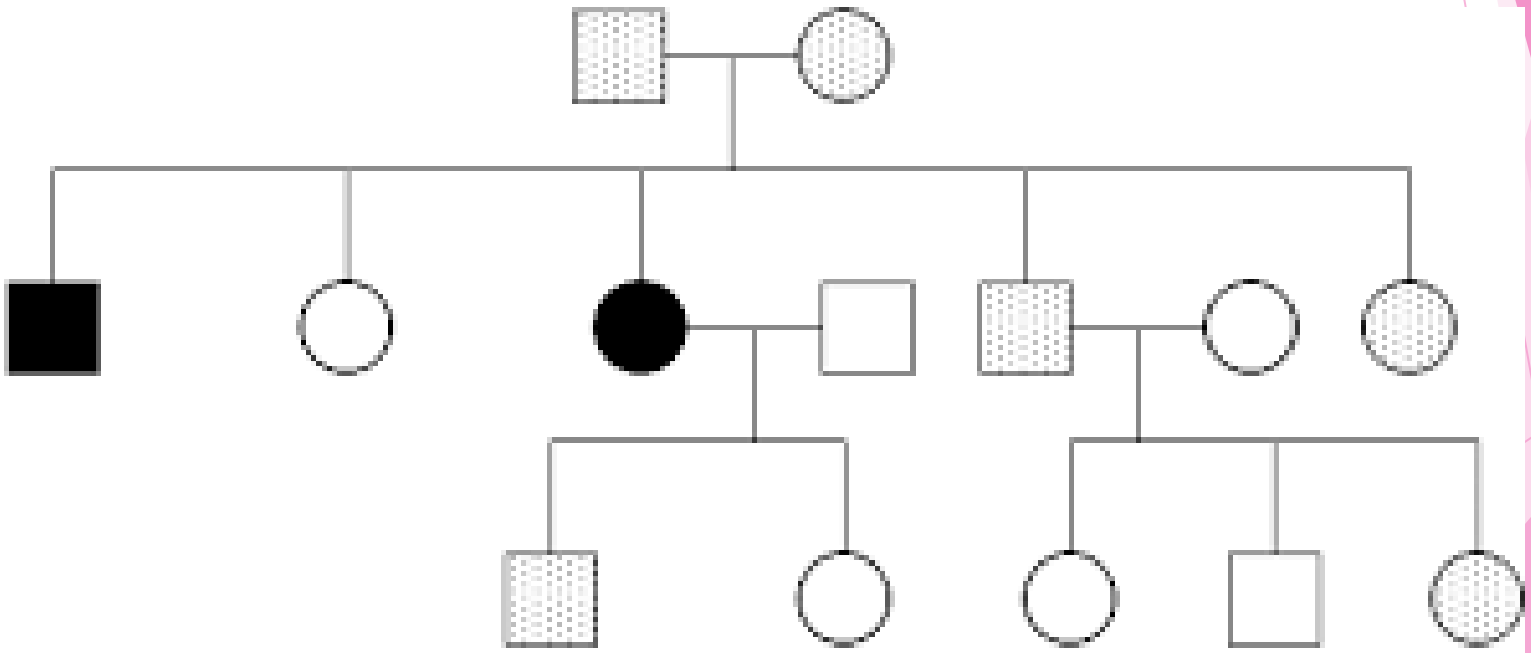
Board Question

- What type of disorder is this: Autosomal dominant, Autosomal recessive, Sex linked Y, Sex linked X



Board Question

- What type of disorder is this: Autosomal dominant, Autosomal recessive, Sex linked Y, Sex linked X



Board Question

- What type of disorder is this: Autosomal dominant, Autosomal recessive, Sex linked Y, Sex linked