In Class Assignment KEY

June 8, 2021 9:58 PM

Science 10: *Introduction to Mutations*

Name: _____

Date: _____

When you're tuning a guitar, the smallest of tweaks can make a big difference to the way the instrument sounds. And in books, small changes in the way the words are put together can change how the story unfolds – little changes can have big effects.

The same applies to DNA – the molecule that carries the genetic information you inherited from your parents. Your DNA directly affects how your body is built and functions, and small changes in the information it contains – *mutations* – can have a big impact.

Often these mutations occur because the processes used to copy DNA are imperfect. Very occasionally the misspellings originate in your own body, but most often they are inherited from parents, who in turn likely inherited them from their parents. The mutations can alter the construction of proteins vital to our bodies, with significant harmful results. On the other hand, many mutations have no noticeable effects at all.

BRCA2 is a very well studied gene because it has a mutation that increases a woman's chance of breast cancer five to ten times. In fact, the gene's name stands for *Breast Cancer 2*. But recently scientists found a new mutation on the same gene. It also increases the risk of breast cancer but is dangerous in a different way as well – it significantly increases the risk of lung cancer in smokers. Which goes to show how two small tweaks in the same gene can carry two very different – and dangerous – tunes.

Just to warm you up, here are some questions to find out what you already know about genes. Perhaps you can discuss them in class before answering.

Question 1

If you look more like one parent then you have inherited more of that parent's genes.

- ○ True
- ● False

Question 2

Boys get all their traits from their fathers and girls get their traits from their mothers.

- ○ True
- ● False

Question 3

The genetic information passed on to you from your parents came from their sex cells – that is, from your father's sperm and your mother's egg cells.

- ● True
- ○ False

Question 4

Your genes determine all your characteristics.

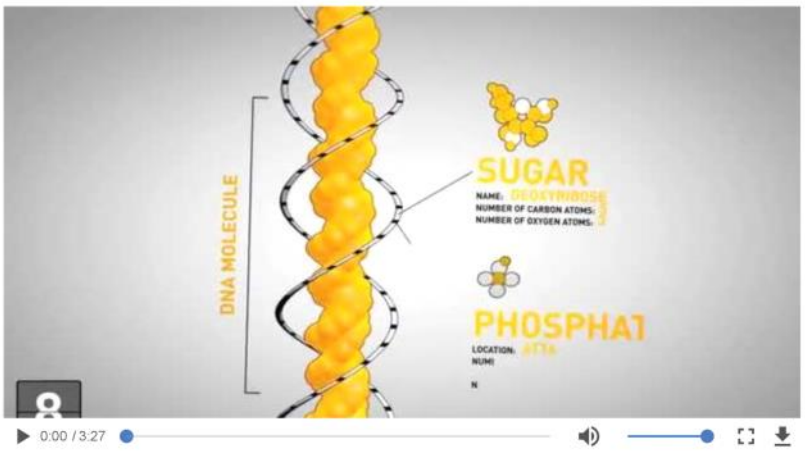
- ○ True
- ● False

Left: DNA samples are often taken by lightly scraping the inside of the mouth. Some of our cells, containing DNA, are in the saliva. Right: Testing DNA in a lab.

GATHER: DNA, chromosomes and genes

Gregor Mendel showed in 1865 that both parents pass traits on to offspring. But no-one knew how. It wasn't until 1944 that the chemical substance that carried these traits was discovered. It was a molecule called deoxyribonucleic acid, or **DNA**.

Watch the Video: 18 Things You Should Know About Genetics by Genome BC (YouTube)
(there is also a direct link on our class website)



Question 1

Notes: Use this space to take notes for the video.

Note: This is not a question and is optional, but we recommend taking notes – they will help you remember the main points of the video and also help if you need to come back to answer a question or review the lesson.

answers will vary

Question 2

Match: Which of the following statements apply best to *DNA*, *gene* or *chromosome*.

Statement	applies to...
Shaped like a twisted ladder called a double helix	DNA
Contains many genes	chromosome
Unit of instruction inherited from parents	gene
Constructed from sugar and phosphate molecules and four bases	DNA
Long DNA molecule packaged tightly into a coil	chromosome
Provides a particular cellular instruction (to make a protein)	gene

2

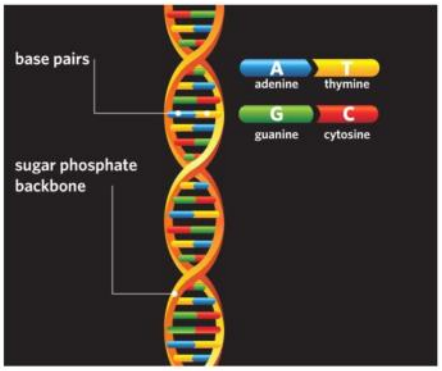
Each DNA molecule is a *double helix*. You can easily see the=two strands in the diagram (right) where the *double* comes=from, and *helix* just means that the strands are twisted=around each other.

If you straighten out the molecule, DNA looks like a ladder.=Each "step" is made up of two **bases –** also called=*nucleobases*. These link together, joining the two strands of=the molecule together.

There are four types of base and each one links to only=one other type, forming **base pairs**:

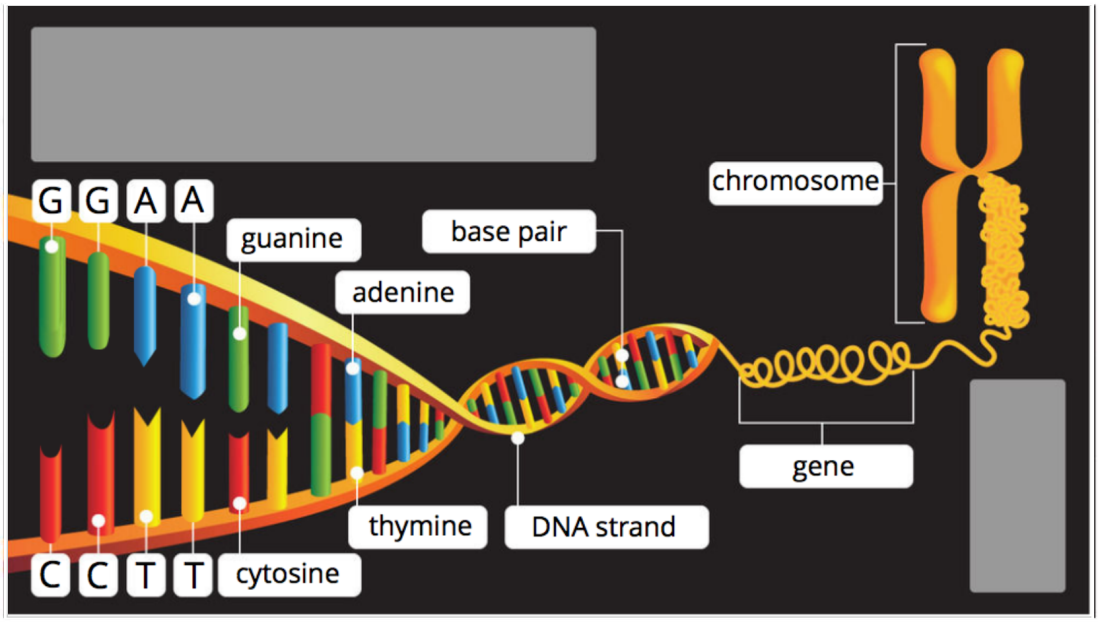
- adenine (A) and thymine (T) link together – **A-T**
- guanine (G) and cytosine (C) link together – **G-C**

When all the bases on two DNA strands align in their A-T= and G-C pairings a stable DNA molecule forms.



Question 3

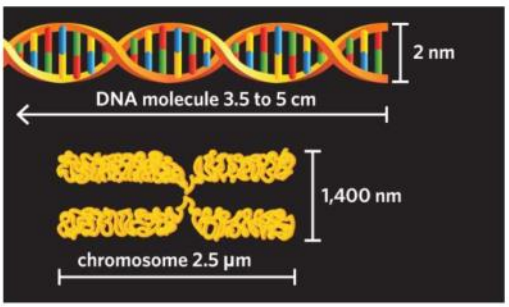
Label: In the diagram below



Did you know?

In humans, each DNA molecule is from 3.5 cm to 5 cm long. Put all the DNA from the 46 chromosomes – in just one cell – end-to-end, and it comes to 1.8 m. That's probably taller than you!

Sometimes, for example when a cell is dividing, the DNA molecules coil themselves up to form the H-shaped chromosomes you sometimes see pictures of. They end up about 2.5 µm long – 1/16,000th of their original length.



3

Codons, amino acids and proteins

The sequence of bases in each gene provides instructions for building **proteins**.

Proteins are molecules. They are often very large, made up of many thousands of atoms, and they are critical to all living organisms. They make up most of the tissue in the organisms' bodies and help control all of the chemical reactions that occur in the organisms.

When the machinery in a cell reads the bases that make up a gene it interprets them in groups of three. Each group of three is called a **codon**. Codons "code" for **amino acids** – the building blocks of proteins.

So, to summarize:

- genes are made up of sequences of bases,
- bases are grouped into codons,
- codons code for amino acids,
- amino acids make up proteins.

With the essential roles that proteins play, you can see how important our genes are!

Human proteins are made from 20 amino acids. Your answer above should have told you that there are more codon= permutations than that, so some amino acids are coded for by more than one codon.

Which codons code for which amino acids can be shown in a codon wheel, like the one below.

To use it, fid the fist letter of your codon in the inner circle and work outwards to the second letter and fially to the third=letter. The outermost ring shows the amino acid coded for.

Hint: To double-check your answer for the number of codon permutations above, you can count the number of sections in the= third ring of the codon wheel.

Here are some examples using the wheel:

1. ATG = methionine – this amino acid is found at the start of every gene
2. GTA = valine
3. TGA, TAA and TAG do not code for amino acids. They each stop protein creation.

Note: A larger, clearer version is provided on the next page

A – alanine
C – cysteine
D – aspartic acid
E – glutamic acid
F – phenylalanine
G – glycine
H – histidine
I – isoleucine
K – lysine
L – leucine
M – methionine
N – asparagine
P – proline
Q – glutamine
R – arginine
S – serine
T – threonine
V – valine
W – tryptophan
Y – tyrosine

Question 7

Identify: Identify one amino acid that is coded for by more than one codon. Write two 3-letter codon sequences associated= with your chosen amino acid.

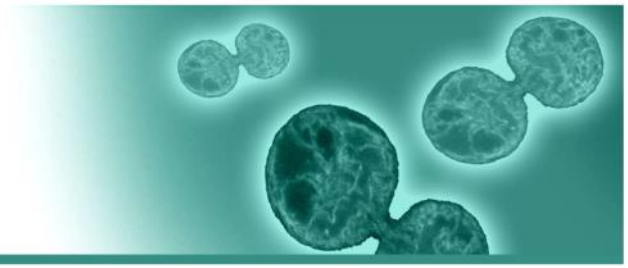
All amino acids except for tryptophan and methionine are encoded by more than one codon so many different answers are possible.

For example: arginine (R) = CGA, CGC, CGG, CGT, AGA, AGG

4

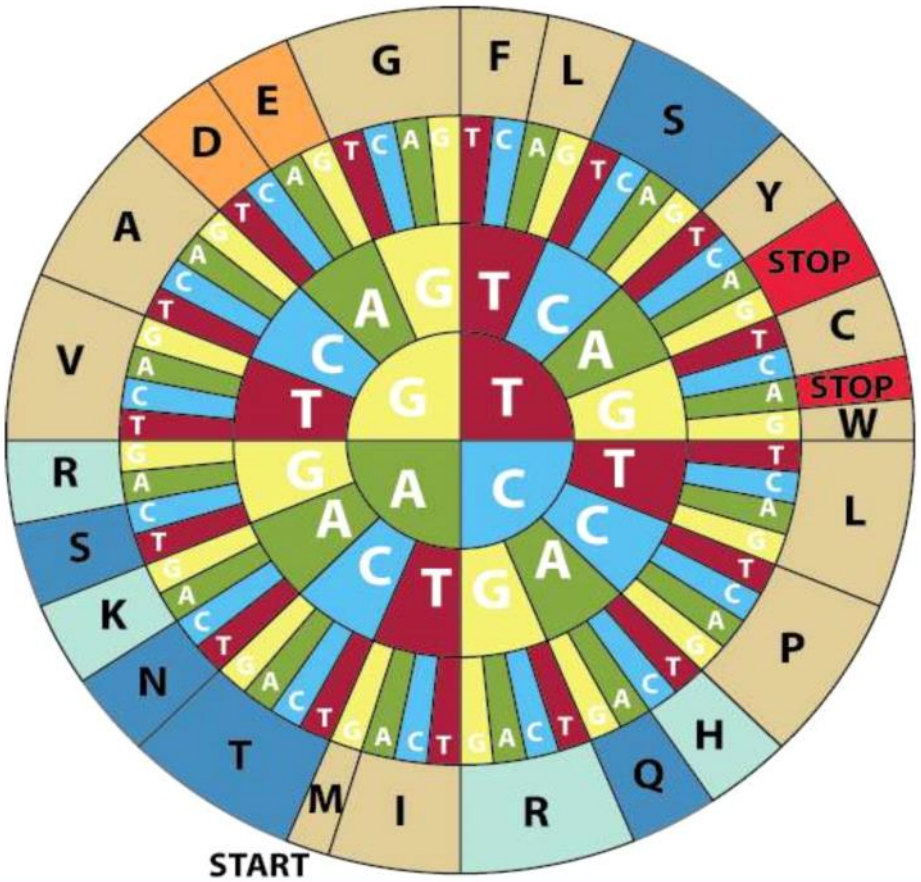
Codon Wheel

Decoding DNA



Use the codon wheel to translate DNA codons into amino acids.

To decode a codon find the first letter of your sequence in the inner circle and work outwards to see the corresponding amino acid. For example: CAT codes for H (Hisitidine).
*Please note that this wheel uses the sense DNA codons (5' to 3').



Amino acid code

A - Alanine	G - Glycine	M - Methionine	S - Serine
C - Cysteine	H - Hisitidine	N - Asparagine	T - Threonine
D - Aspartic Acid	I - Isoleucine	P - Proline	V - Valine
E - Glutamic acid	K - Lysine	Q - Glutamine	W - Tryptophan
F - Phenylalanine	L - Leucine	R - Arginine	Y - Tyrosine

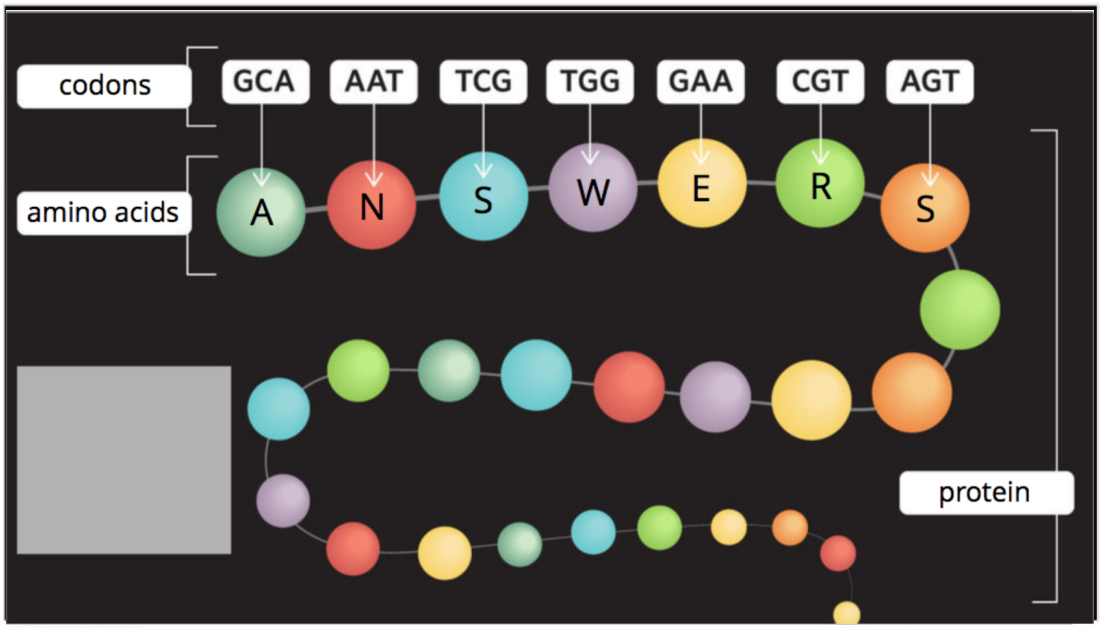
Image: C. Brooksbank, European Bioinformatics Institute

yourgenome.org

wellcome trust sanger institute

Question 8

Interpret: Using the Word Bank provided, label the diagram shown below:



Question 9

Determine: Two of the following DNA sequences code for the same small protein. Determine the amino acid sequence of that protein.

1. ATG TTA ACG AGA AGT

1 & 3
M, L, T, R, S

Process: Genetics

Point mutations

The *Introduction* tells how small changes in genes can have big consequences. Scientists use the term **mutation** to refer to= any such changes that persist, passing down to new generations. There are many different types of mutations, but some= involve a change to just a single base. These are called **point mutations**.

Watch the Video:
Mutations in DNA by Teacher's Pet (YouTube).

(there is also a link on our class website)

Question 1

Notes: Use this space to take notes for the video.

Note: This is not a question and is optional.

answers will vary

Question 2

Define: A point mutation is a change in a single

- [ ] amino acid
- [ ] protein
- [x] base
- [ ] gene

Question 3

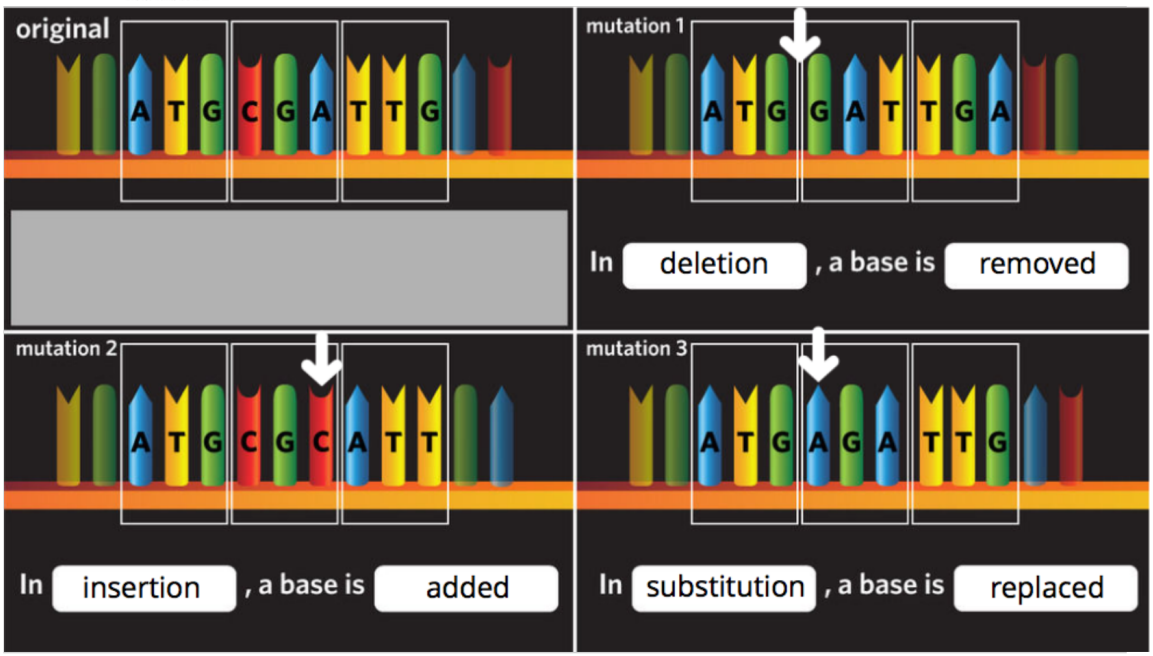
Consider: If a point mutation causes a frameshift, the resulting protein is likely nonfunctional. This is because:

- [ ] frameshifts always replace many bases, changing the sequence of amino acids in the protein.
- [ ] frameshifts only change a single amino acid, but that is enough to disable the resulting protein.
- [x] all the amino acids after the changed base are likely to be different.

Question 4

Identify: The diagram below shows an original base sequence and three mutations of it, each one caused by a different point mutation.

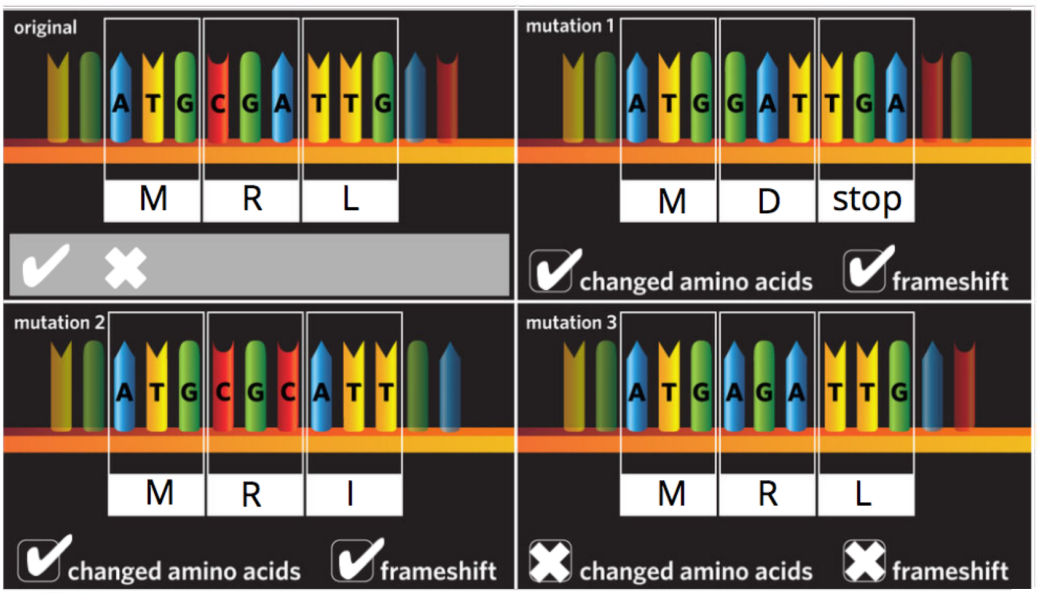
1. Using the word bank provided, label the boxes to identify and explain each type of point mutation.
2. Draw the arrows to show which base changed in each mutation, or where a base was removed.



Question 5

Deduce: The diagram below shows the same original sequence and point mutations as the question above. This time:

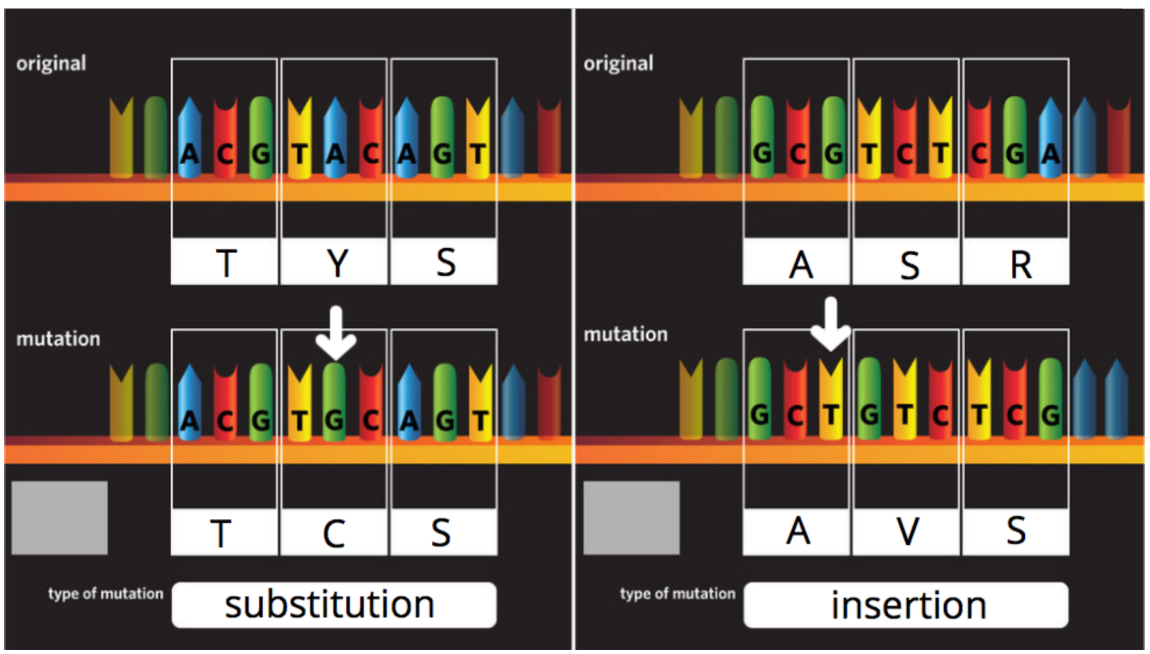
1. Use the codon wheel on page 5 to work out the amino acids produced in each case, and write them in. *Double-click or tap the text boxes to enter text.*
2. Put ticks or crosses in the appropriate boxes to indicate if the point mutations have changed the amino acid sequences, and if there has been a frameshift.



Question 6

Summarize: For each of the two point mutations below:

- Draw an arrow to identify where the mutation occurred
- Use the DNA codon wheel on page 5 to find the mutated sequence
- Name the type of point mutation



Question 7

Reason: A single-base substitution can occur in the first, second or third position of a codon. In which position is the point mutation most likely to go unnoticed?

Why? Use examples from the DNA codon wheel to support your answer.

A substitution is most likely to go unnoticed in the third position. This is because for many codons the first two positions determine the amino acid, and the 3rd position can have any of the 4 bases. Examples are:

- any codon that starts with CC codes for proline; CCA, CCC, CCG, and CCT all code for proline

In many other cases, with the first two positions fixed, 2 bases in the 3rd position code for the same amino acid. For example:

- AGG and AGA both code for arginine, and AGC and AGT both code for serine.

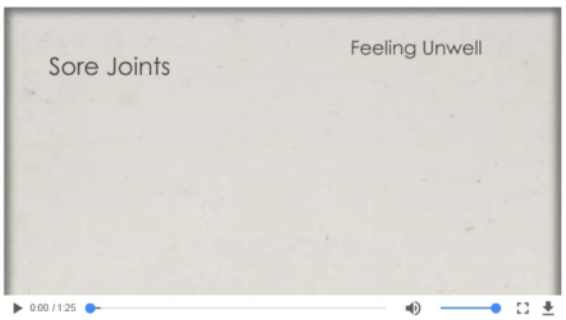
In these cases changing the base in the 3rd position still leaves a 33% chance that the amino acid will not change.

Haemochromatosis

Haemochromatosis, or inherited iron overload, is a treatable metabolic disorder caused by DNA mutations.

Watch the Video: Haemochromatosis - Animation (YouTube).

(a direct link can be found on the class website)



Question 8

Notes: Use this space to take notes for the video.

Note: This is not a question and is optional.

answers will vary

Question 9

Relate: People faced with haemochromatosis, either because they have or carry the condition or someone in their family does, can meet with a genetic counsellor to discuss the disorder and seek advice. You can find some of the information they might be given in this booklet.

Working with a partner, jot down three or four questions that someone with haemochromatosis in their family might want to ask, and the answers. Work these into a short dialogue between the person and a counsellor.

Act out your dialogue in front of your class peers. If you have time, switch roles.

[There are many possible questions and answers... some examples are provided below.]

Client: Neither of my parents have haemochromatosis; does that mean that I am safe?

Counsellor: Not necessarily, it could be that both of your parents are carriers. In that case there is about a 25% chance that your genes have two copies of the mutation that predisposes you to having the disorder.

Client: So if I have those genes I'll definitely get the disorder?

Counsellor: No, even if you have two copies of the mutated gene there is a chance that you won't develop the disorder. 20% of men with two mutated genes and 40% of women don't develop it.

Client: How would I find out if I or my children had haemochromatosis?

Counsellor: First you might notice some symptoms, like tiredness, weakness, weight loss or stomach pains. People are very different in the symptoms they show. The first step to be sure is to take some blood and test the amount of iron in it. If it is very high then we would take more blood and test for the genetic mutations associated with the disorder.

Client: What would the treatment be?

Counsellor: The goal of treatment is to stop blood iron levels getting too high. The main way that this is done is by "venesection", which just means taking blood with a needle, just the same as when people donate blood.

Client: How often?

Counsellor: That differs a lot, but men usually need 3 or 4 venesections a year.

Note: There may be more than one correct answer to the multiple-choice questions below.

Question 1 (1 mark)

Genes:

- [ ] produce DNA
- [ ] make chromosomes coil up
- [x] provide instructions for cells to make proteins
- [ ] fight disease in the body
- [x] are inherited from parents
- [ ] I'm not sure

Question 2 (1 mark)

On one _____ there are many _____ .

- [ ] gene; chromosomes
- [ ] base; genes
- [ ] chromosome; proteins
- [x] chromosome; genes
- [ ] I'm not sure

Question 3 (1 mark)

Which of the following are true about DNA?

- [x] It is a molecule
- [x] A single strand in a cell is a chromosome
- [ ] Each individual piece has just three base pairs
- [x] It is shaped like a twisted ladder
- [ ] It is made up of amino acids
- [ ] I'm not sure

Question 4 (1 mark)

Which of the following are true about bases? A-T-C-G

- [ ] Each base pair constitutes a gene
- [x] They are part of DNA
- [x] Humans have four types of base
- [ ] Each base type can link with each other base type
- [ ] I'm not sure

Question 5 (1 mark)

One _____ has many _____ .

Hint: There are two correct answers.

- [ ] amino acid; base pairs
- [x] gene; base pairs
- [x] chromosome; base pairs
- [ ] base pair; genes
- [ ] I'm not sure

Question 6 (1 mark)

Which of the following base pair linkages are possible?

- [x] A-T (adenine links to thymine)
- [ ] A-C (adenine links to cytosine)
- [x] G-C (guanine links to cytosine)
- [ ] G-T (guanine links to thymine)
- [ ] I'm not sure

1 1

Question 7 (1 mark)

Three _____ in sequence are called a/an _____ and code for a/an _____ .

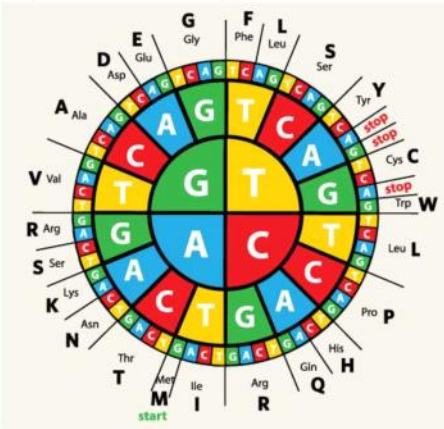
- [ ] bases; amino acid; protein
- [ ] amino acids; codon; protein
- [ ] genes; amino acid; protein
- [x] bases; codon; amino acid
- [ ] I'm not sure

Question 8 (1 mark)

Which of the following are true about proteins?

- [x] They control chemical reactions in the body
- [ ] They are inherited directly from parents
- [x] They are molecules
- [ ] Most of the tissue in our bodies is made from them
- [x] They have complex branching structures made up of amino acids
- [ ] I'm not sure

Use the diagram below to answer **Questions 9** and **10**.



A - alanine
C - cysteine
D - aspartic acid
E - glutamic acid
F - phenylalanine
G - glycine
H - histidine
I - isoleucine
K - lysine
L - leucine
M - methionine
N - asparagine
P - proline
Q - glutamine
R - arginine
S - serine
T - threonine
V - valine
W - tryptophan
Y - tyrosine

Question 9 (1 mark)

Use the diagram to identify which of the claims below are true.

- [x] The sequence CAT codes for histidine (H)
- [x] Sequences CGC and AGA both code for the same amino acid
- [ ] The sequence TAT codes for threonine (T)
- [x] Any sequence beginning TC codes for serine (S)
- [ ] I'm not sure

Question 10 (1 mark)

What do the "stop"s in the diagram indicate?

- [x] A sequence marking the end of a gene, where protein production stops
- [ ] A sequence marking the end of a chromosome
- [ ] A sequence to produce a particular type of amino acid called a "stop" acid
- [ ] That these base sequences never occur in nature
- [ ] I'm not sure

Question 11 (1 mark)

A point mutation is:

Choose the best answer.

- [ ] a change to a single gene
- [ ] a change to a single codon
- [x] a change to a single base
- [ ] a change that results in the production of pointy proteins
- [ ] any change that has no impact on protein production
- [ ] I'm not sure

Question 12 (1 mark)

A *substitution* point mutation always results in a different amino acid being coded for.

- [ ] True
- [x] False
- [ ] I'm not sure

Question 13 (1 mark)

Which of the following are point mutations that can cause a frameshift?

- [x] Insertion
- [ ] Mutation
- [x] Deletion
- [ ] Substitution
- [ ] Adaptation
- [ ] I'm not sure

Question 14 (1 mark)

A sequence of bases in a particular gene is normally:

A A T G T A C C C

but a point mutation changes this to:

A T G T A C C C G

This is an example of:

- [ ] Insertion
- [ ] Mutation
- [x] Deletion
- [ ] Substitution
- [ ] Adaptation
- [ ] I'm not sure

Question 15 (1 mark)

Compared to point mutations that don't result in frameshifts, ones that do are:

- [x] more likely to result in nonfunctional proteins, because many amino acids may change
- [ ] more likely to result in nonfunctional proteins, because the order of amino acids in the protein is reversed
- [ ] less likely to result in nonfunctional proteins, because most of the bases are not affected
- [ ] I'm not sure

Question 16 (1 mark)

Point mutations:

- [ ] are always beneficial
- [ ] are always harmful
- [ ] never impact protein production
- [x] can be beneficial, harmful or have no effect
- [ ] I'm not sure

Question 17 (1 mark)

Learning goal 1: DNA, chromosomes, bases and genes are all closely related. Briefly explain what they are.

Bases, such as A-T-C-G, make up the structure of DNA along with a phosphate and sugar molecule. During cell division, DNA condenses into Chromosomes *(the X shape)*. Genes are sections of chromosomes which code for a specific trait or function in the body.

Question 18 (1 mark)

Learning goal 2: In outline, describe how the bases in DNA produce proteins.=*Note: Include the words* gene *and* codon.

The specific sequences of bases along the DNA strand within a gene will code for a certain protein. A 3-base sequence, called *a codon*, codes for the production of 1 amino acid. A string of many amino acids together will combine to build the protein, to perform the function/trait in the organism

Question 19 (1 mark)

Learning goal 3: Briefly describe the three types of point mutation.

Point mutations in the DNA code refer to a single section (a single base) that has been altered. In some cases *(insertion or deletion)* this may cause a frameshift, meaning the remaining strand *(all the other bases in the code)* are shifted to the left *(in a deletion)* or the right *(in an insertion)*. A frameshift will most likely result in all remaining amino acids being coded incorrectly.

Substitution: when one DNA base pair is replaced by another. This causes a *frameshift* to the right. For example the C has been replaced by a T *(diagram)*

Insertion: an additional/extra base has been added to the DNA strand. For example an extra C base has been added *(in diagram)*

Deletion: when a base has been removed form the DNA strand. This causes a *frameshift* to the left. For example the A base has been removed *(diagram)*