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STEM SMART Biology Week 26 - Mutations

Mutations: Types & Effects

Subject & topics

Biology | Genetics | Genes & Alleles

Status

Not started

Stage & difficulty

A Level Practice 2

Part A

Point mutations

Point mutations are changes that happen to a single pair of nucleotides. They are most likely to occur during DNA replication (i.e. during S phase of the cell cycle).

What is the name given to the point mutation in which the correct nucleotide is replaced by an incorrect nucleotide?

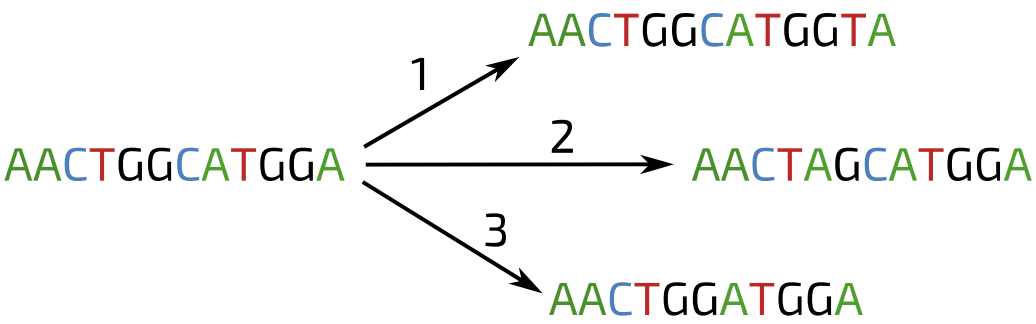
What is the name given to the point mutation in which a nucleotide pair is added into the sequence?

What is the name given to the point mutation in which a nucleotide pair is lost from the sequence?

Part B

Mutation identification

The image below shows a DNA sequence undergoing three different types of point mutation.



Match the mutation to the type.

- Mutation 1: ____
- Mutation 2: ____
- Mutation 3: ____

Items:

inversion | insertion | duplication | translocation | deletion | substitution

Part C

Coding sequence mutations

Within a coding region of a gene, a single-nucleotide [ ] is likely to have a large effect on the amino acid sequence, because this type of mutation will change the reading frame (the grouping of the sequence into codons). Therefore, this type of mutation (which can also be called a [ ] mutation) will not only likely change the amino acid at that site, but all subsequent amino acids in the sequence.

Within a coding region of a gene, a single-nucleotide [ ] will only change the amino acid at that site (and it may not change the amino acid, due to the redundancy of the genetic code). However, this type of mutation could produce a premature [ ] in the sequence, which would remove all subsequent amino acids from the amino acid sequence.

A substitution which changes the amino acid is called a [ ] substitution.

A substitution which does not change the amino acid is called a [ ] substitution.

Items:

nonsynonymous **substitution** **frameshift** **stop codon** **insertion/deletion** **synonymous**

Part D

Chromosomal mutations

Chromosomal mutations are changes that happen to entire regions of a chromosome, and may therefore cause changes to multiple genes. They are most likely to occur during meiosis, as a result of homologous chromosomes not aligning correctly.

What is the name given to the chromosomal mutation in which a region of the chromosome is removed and lost?

What is the name given to the chromosomal mutation in which a region of the chromosome becomes repeated within the same chromosome?

What is the name given to the chromosomal mutation in which a region of the chromosome is removed and becomes part of another chromosome?

What is the name given to the chromosomal mutation in which a region of the chromosome is removed and reinserted in the opposite direction within the same chromosome?

Part E

Mutation consequences

Which of the following are possible consequences of a germ cell mutation? Select all that apply.

- [ ] a new advantageous allele of a gene is produced that may be passed on to offspring
- [ ] a new deleterious allele of a gene is produced that may be passed on to offspring
- [ ] a change in the amino acid sequence that the gene codes for
- [ ] no change in the amino acid sequence that the gene codes for
- [ ] a change in the expression of the gene (e.g. the gene is expressed in different contexts, or for different durations)
- [ ] a change to an essential cell process that causes the death of the cell

Which of the following are possible consequences of a somatic cell (e.g. liver cell) mutation? Select all that apply.

- [ ] a new advantageous allele of a gene is produced that may be passed on to offspring
- [ ] a new deleterious allele of a gene is produced that may be passed on to offspring
- [ ] a change in the amino acid sequence that the gene codes for
- [ ] no change in the amino acid sequence that the gene codes for
- [ ] a change in the expression of the gene (e.g. the gene is expressed in different contexts, or for different durations)
- [ ] a change to an essential cell process that causes the death of the cell

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STEM SMART Biology Week 26 - Mutations

Genotypes & Phenotypes

Subject & topics

Biology | Genetics | Genes & Alleles

Status

Not started

Stage & difficulty

A Level Practice 2

Part A

Gene definitions

Match the terms to the definitions in the table below.

Term	Definition
	a region of DNA that codes for a protein
	one of several (two or more) variants of a gene that exist in a population
	the allele(s) that an individual has
	an observable trait that an individual has
	containing two copies of the same allele
	containing two different alleles

Items:

dominant | gene | phenotype | genotype | allosome | homozygous | autosome | heterozygous | recessive | allele

Part B

Allelic interactions

Match the terms to the definitions in the table below.

Term	Definition
	an allele that will produce a phenotype regardless of the presence of other alleles for that gene
	an allele that will only produce a phenotype if it is the only allele present
	when two different alleles combine to produce a different phenotype from either homozygous phenotype
	when the phenotype produced by a genotype for one gene depends on the genotype for another gene

Items:

dominant **recessive** **epistasis** **heterozygous** **autosomal-linked** **homozygous** **codominance** **sex-linked**

Part C

Chromosomes & linkage

Each human [ ] cell contains [ ] pairs of chromosomes. One of these pairs is responsible for sex determination in the early embryo. These two chromosomes are called [ ] (also called "allosomes"). The other chromosomes (non-sex chromosomes) are called [ ].

Genes found on the sex chromosomes are described as "[ ]". This means that the inheritance patterns of these genes differs between males and females.

Genes that are located on the **same** non-sex chromosome are described as "[ ]". This means that the alleles for these genes will be inherited together (i.e. not recombine with alleles found on the homologous chromosome) unless crossing over occurs.

Items:

sex chromosomes **autosomes** **gametic** **sex-linked** 46 23 **somatic** **autosomal-linked**

Part D

Examples

Match the terms to the descriptions in the table below.

Description	Example of
Two plants are both homozygous for a gene involved in flower colour. One plant produces red flowers, and the other plant produces white flowers. When the two plants are bred together, all of the offspring produce pink flowers.	
Two flies are both heterozygous for two genes: one involved in body colour and one involved in wing length. When the two flies mate, almost all of the offspring either have brown bodies & long wings **or** black bodies & short wings. Very few offspring have brown bodies & short wings or black bodies & long wings.	
A female mouse is homozygous for a gene ("gene A"). A male mouse only has one copy of gene A, and contains a different allele to the female. When the two mice mate, none of the male offspring contain the father's allele for gene A.	
Two dogs are both heterozygous for two genes ("gene A" and "gene B"). Gene A codes for a pigment, and is therefore involved in body colour. When the two dogs mate, the offspring that have the same genotype for gene A are different colours because they have different genotypes for gene B.	

Items:

epistasis **codominance** **autosomal-linkage** **sex-linkage**

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STEM SMART Biology Week 26 - Mutations

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STEM SMART Biology Week 26 - Mutations

Genetic Differences

Subject & topics

Biology | Genetics | Genes & Alleles

Status

Not started

Stage & difficulty

A Level Practice 2

Part A

Gene sequences

A student analysed a gene sequence that had been identified in four different types of organism. The gene codes for a functional protein. A section of the gene's DNA is shown below. The rest of the DNA from this gene (not shown) is identical in all four different types of organism.

organism	DNA sequence					
human	ACG	CCT	CGT	CAC	GCT	AAA
oak tree	ACG	GAA	TAT	GTA	GCT	AAA
mushroom	ACG	GAA	CTC	TTA	GCT	AAA
E. coli bacterium	ACG	TAC	GAT	GGG	GCT	AAA

Which of the following statements are correct? Select all that apply.

- [ ] The human version of this gene only differs by three codons from the *E. coli* version of this gene.
- [ ] This gene may code for chlorophyll.
- [ ] This gene may code for a ribosomal protein.
- [ ] This gene may be found in the nucleus or in the cytoplasm.
- [ ] The protein that this gene codes for is likely to be more similar in plants and fungi than in other organisms.
- [ ] A single-base deletion in the first codon would only change the first amino acid of the protein.

Part B

Mitosis & mutation

A diploid plant cell divides by mitosis.

After mitosis of this cell, a mutation occurs that changes the genotype of **one** of the daughter cells. This mutant daughter cell produces a non-functional enzyme instead of the functional enzyme produced by the other daughter cell. This mutation has no effect on the phenotype of the plant or the number or length of chromosomes in the plant cell.

Which of the following statements are correct? Select all that apply.

- [ ] The mutation may have changed the amino acids in the active site of the enzyme.
- [ ] Both daughter cells will contain the same alleles for every gene.
- [ ] Both daughter cells are haploid.
- [ ] The sequence of bases along each allele will be the same in each daughter cell.
- [ ] The chromosomes in the nucleus of each daughter cell will contain the same genes.

Part C

Mutation origins

A child is affected by a dominant genetic condition. All of his cells have the same genotype. The allele associated with this condition is not present in either of his parents.

Which of the following statements are correct? Select all that apply.

- [ ] The mutation may have occurred in the child's DNA after he was born.
- [ ] Both of his grandmothers had the condition.
- [ ] The mutation may have occurred in the child's DNA during the zygote stage.
- [ ] All of the child's future offspring will have the condition.
- [ ] The mutation may have occurred during meiosis in his father.

Adapted with permission from NSAA 2018 Section 1 Q69 & NSAA 2018 Section 1 Q72 & NSAA 2019 Section 1 Q66

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Haemoglobin Mutations

Subject & topics

Biology | Genetics

Status

Not started

Stage & difficulty

A Level Challenge 1

Table 1 shows the first seven amino acids of the β chain of haemoglobin and the corresponding 21 base pairs of DNA that code for these seven amino acids. The leftmost base pair is referred to as base pair 1.

Table 1

Amino acid sequence	Val	His	Leu	Thr	Pro	Glu	Glu
DNA sequence	CAC	GTG	GAC	TGA	GGA	CTC	CTC
	GTG	CAC	CTG	ACT	CCT	GAG	GAG

Table 2 shows the corresponding mRNA codons for a selection of amino acids.

Table 2

Amino acid	**mRNA codon(s)**
cysteine (Cys)	UGC, UGU
glutamic acid (Glu)	GAA, GAG
histidine (His)	CAC, CAU
leucine (Leu)	CUA, CUC, CUG, CUU
proline (Pro)	CCA, CCC, CCG, CCU
threonine (Thr)	ACA, ACC, ACG, ACU
valine (Val)	GUA, GUC, GUG, GUU
no amino acid (STOP)	UAA, UAG, UGA

Part A

DNA strands

What is the name given to the **top** DNA strand (with the sequence "CAC GTG...") in **Table 1**?

What is the name given to the **bottom** DNA strand (with the sequence "GTG CAC...") in **Table 1**?

Part B

Deletion of base pair 6

A single base pair deletion occurs at base pair 6 in the β haemoglobin DNA sequence shown in **Table 1**.

Enter the new amino acid sequence for the first three DNA base pair triplets.

Enter the amino acid sequence from left to right, with the first amino acid on the left.

[ ] – [ ] – [ ]

Items:

Cys Glu His Leu Pro Thr Val STOP

Part C

Deletion of base pairs 7 to 9

In another cell, base pairs 7, 8, and 9 are deleted from the β haemoglobin DNA sequence shown in **Table 1**.

Enter the new amino acid sequence for the first four DNA base pair triplets.

Enter the amino acid sequence from left to right, with the first amino acid on the left.

[ ] – [ ] – [ ] – [ ]

Items:

Cys Glu His Leu Pro Thr Val STOP

Adapted with permission from CIE AS Level Biology, June 2017, Paper 2, Question 4b

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Question deck:

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STEM SMART Biology Week 26 - Mutations

Synonymous & Nonsynonymous Substitutions

Subject & topics

Biology | Genetics | Genes & Alleles

Status

Not started

Stage & difficulty

A Level Challenge 2

The figure below shows how each possible DNA codon is translated.

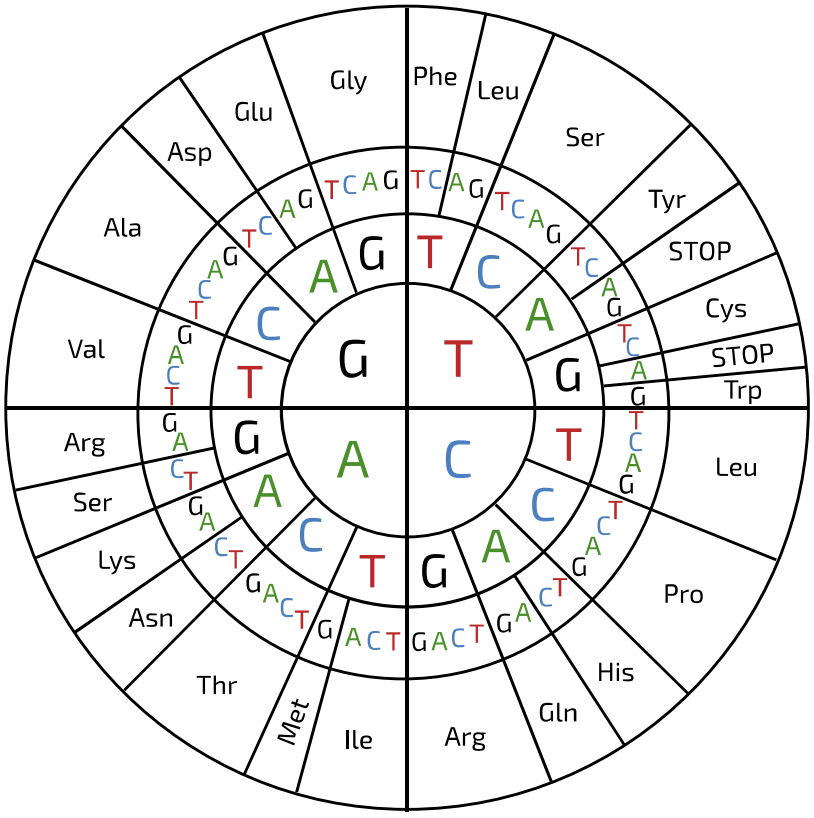

Figure 1: DNA codon wheel. The diagram is read from inside to out, with amino acid names given as abbreviations. For example, the base triplets CAT and CAC both code for the amino acid histidine (His).

The coding region of a particular gene contains the following sequence:

$$5' - \text{GCA CTG} - 3'$$

Part A

Translate the codons

Translate the DNA coding sequence above into an amino acid sequence. Assume that the sequence is "in-frame" i.e. that the first three bases correspond to a single codon.

Amino acid sequence: [ ] – [ ]

Part B

Percentage of synonymous substitutions

What percentage of single-base substitutions that could occur in this sequence are **synonymous** substitutions?

Give your answer to 2 sf.

Part C

Percentage of nonsynonymous substitutions

What percentage of single-base substitutions that could occur in this sequence are **nonsynonymous** substitutions?

Give your answer to 2 sf.

Part D

A sequence with no synonymous substitutions

Enter a six-base DNA sequence below that would have no possible synonymous substitutions. Your sequence should produce an amino acid sequence containing two unique amino acids.

Enter your answer with a space between the two codons e.g. GCA CTG

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Premature Stop Codons

Subject & topics

Biology | Genetics | Genes & Alleles

Status

Not started

Stage & difficulty

A Level Challenge 2

Of the 64 possible codons, there are 3 "stop codons". When a ribosome reaches a stop codon on the mRNA, the polypeptide chain is released and no further amino acids are added.

The three mRNA stop codons are:

- UAA
- UAG
- UGA

A mutation to a coding region of a gene can result in a "premature stop codon" by changing an existing codon that codes for an amino acid to one of the three stop codons above. This results in a shorter polypeptide chain, which may result in a non-functional protein.

Consider the DNA sequence below ("Sequence Z"). The sequence is found on the coding/sense strand of a gene, and is "in-frame" i.e. the first three bases correspond to a single codon. Each codon codes for a different amino acid.

Sequence Z:

$$5' - \mathrm{ATA\ AAC\ CAG} - 3'$$

Part A

Substitution

Enter a nine-base DNA coding sequence below that would be produced by a single-nucleotide **substitution** occurring in sequence Z to result in a premature stop codon.

Enter your answer with a space between each codon e.g. ATA AAC CAG. The first three bases should correspond to the first codon.

Part B

Insertion

Enter a ten-base DNA coding sequence below that would be produced by a single-nucleotide **insertion** occurring in sequence Z to result in a premature stop codon.

Enter your answer with a space between each codon (note that the last codon will be incomplete) e.g. ATA AAC CAG A. The first three bases should correspond to the first codon.

Part C

Deletion

Enter an eight-base DNA coding sequence below that would be produced by a single-nucleotide **deletion** occurring in sequence Z to result in a premature stop codon.

Enter your answer with a space between each codon (note that the last codon will be incomplete) e.g. ATA AAC CA. The first three bases should correspond to the first codon.

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Question deck:
STEM SMART Biology Week 26 - Mutations

STEM SMART Biology Week 26 - Mutations

Mutation Probabilities

Subject & topics

Biology | Genetics | Genes & Alleles

Status

Not started

Stage & difficulty

A Level Challenge 2

The figure below shows how each possible DNA codon is translated.

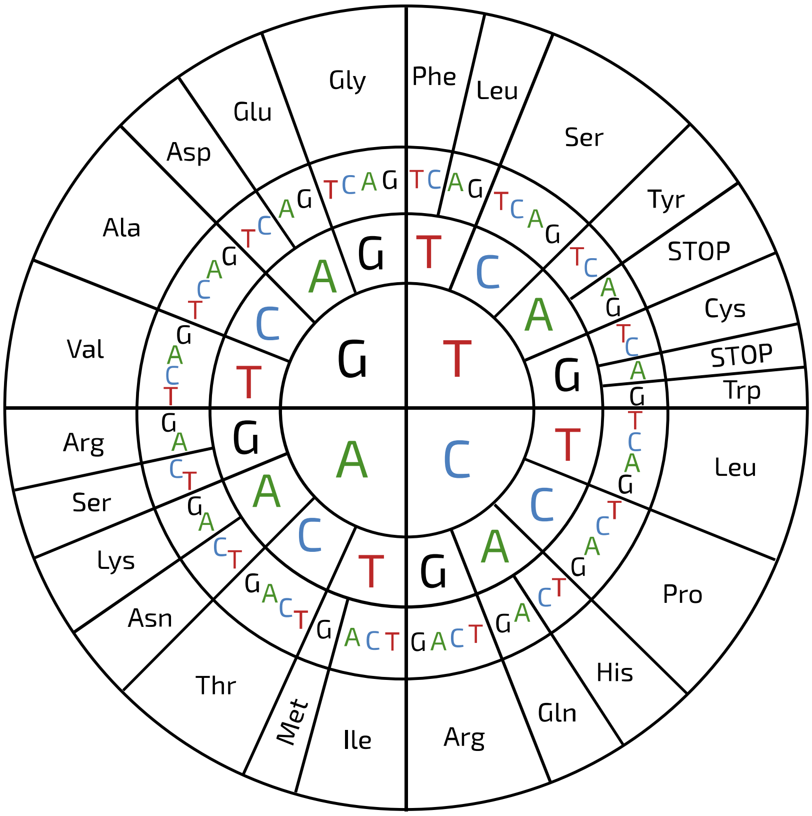

Figure 1: DNA codon wheel. The diagram is read from inside to out, with amino acid names given as abbreviations. For example, the base triplets CAT and CAC both code for the amino acid histidine (His).

In the questions below, assume that it is equally likely that mutations can change any base to any other base, and that the probability of this resulting in a change in any particular base during one cell division is 2.00×10^{-9}.

Part A

Methionine to proline

What is the probability that a triplet that codes for methionine (Met) changes to code for proline (Pro) in one round of division?

Give your answer to 3 significant figures.

Part B

Phenylalanine to glutamine

What is the probability that a triplet that codes for phenylalanine (Phe) changes to code for glutamine (Glu) in one round of division?

Give your answer to 3 significant figures.

Part C

Start to stop

What is the probability that a start codon (Met) changes to a stop codon (STOP) in one round of division?

Give your answer to 3 significant figures.

Part D

Redundancy

Based on **Figure 1**, which of the following amino acids display redundancy/degeneracy in how they are encoded? Select all that apply.

- [ ] glutamine (Glu)
- [ ] proline (Pro)
- [ ] phenylalanine (Phe)
- [ ] tryptophan (Trp)
- [ ] methionine (Met)

Adapted with permission from NSAA 2020 Section 2 Q60