Big Idea 3:

Living systems store, retrieve, transmit and respond to information essential to life processes.

Big Idea 3: Part C

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Gene Expression: Overview

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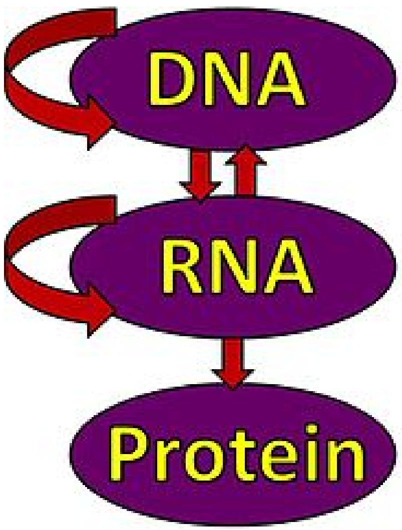
Gene Expression

Gene expression is the molecular process of reading the order of nucleic acids in a DNA molecule and making the product it codes for. This product is usually a protein but RNA is also coded for in genes.



Gene expression occurs whenever a specific protein or RNA moleculeis needed by the cell.

DNA to RNA to Protein

Expressing the information stored on a gene into a protein requires :

· translating from the 4 letter language of DNA to RNA

· Then from the 4 letter language of RNA, to the 20 letter language of proteins (their amino acid sequence).

RNA

RNA is essential for bringing the genetic information stored in the DNA to where it can be used in the cell.

Recall that RNA is made up of a sugar molecule and phosphate group "backbone" and a sequence of nitrogen bases:

Adenine (A)
Uracil (U)
Guanine (G)
Citosine (C)

These bases hydrogen bond in pairs: A bonds to U and G bonds to C.

RNA

A strand with bases in the sequence:

ACUAGGUACAUG

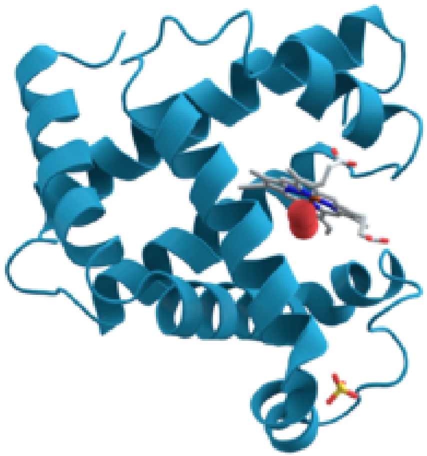
has a different shape, and functions differently, than a strand with the sequence:

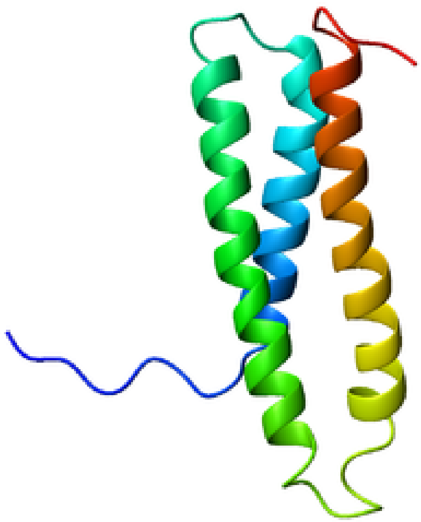
CUAGAACAGUCAA

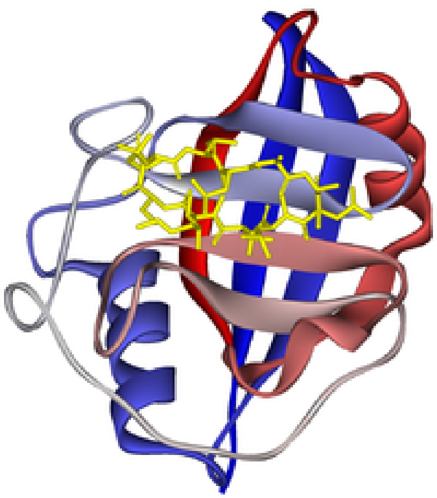
Letter changes result in a new shape, and new functions.

Proteins

Proteins are large biological molecules consisting of one or more chains of amino acids. Proteins perform a vast array of functions within living organisms, including catalyzing metabolic reactions, replicating DNA, responding to stimuli, and transporting molecules from one location to another.

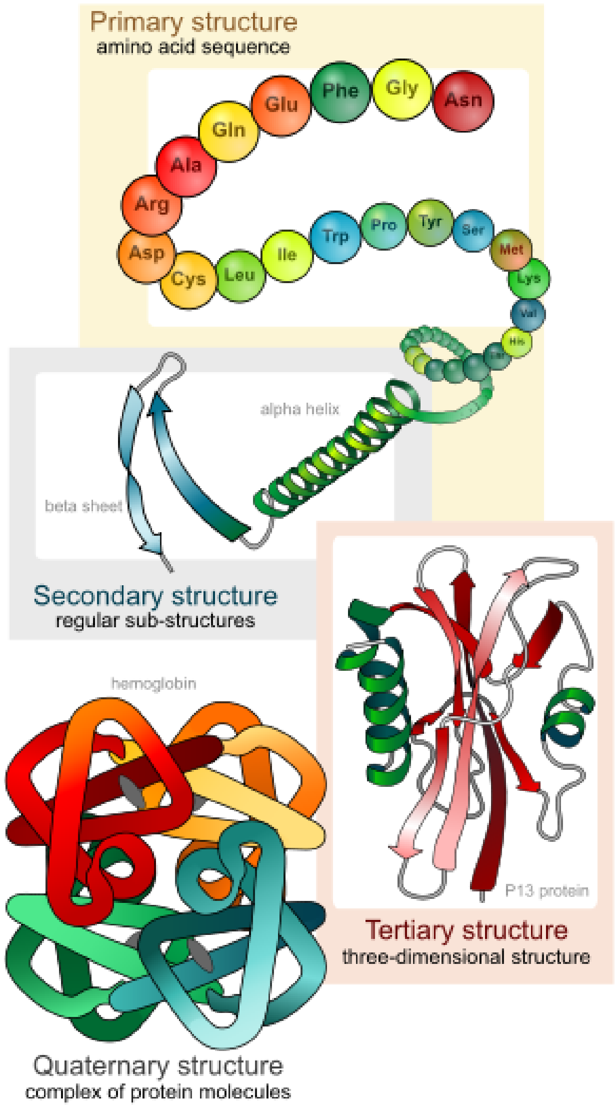






Proteins

Proteins differ from one another primarily in their sequence of amino acids, which is dictated by the nucleotide sequence of their genes, and which usually results in folding of the protein into a specific three-dimensional structure that determines its activity.



Codons

The mRNA "message" is read in 3-letter words called **codons.** Each codon codes for an amino acid or tells the process to stop.

There are 64 codons (4x4x4) but only 20 amino acids. So some codons code for the same amino acid.

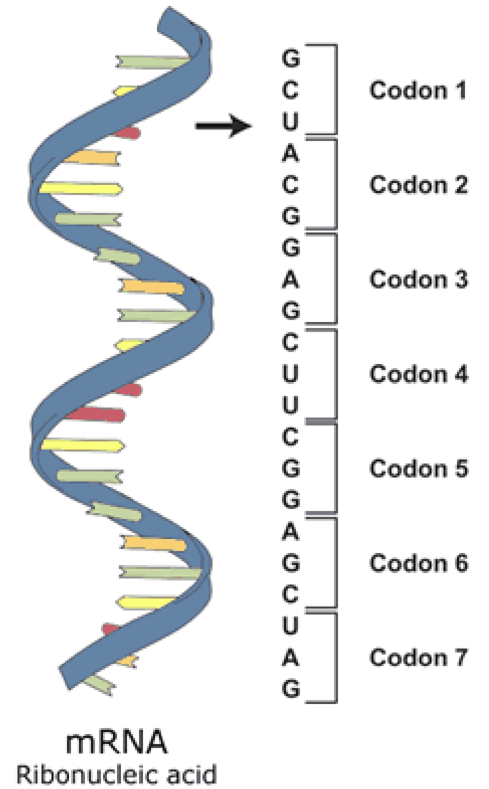


Image adapted from: National Human Genome Research Institute. Talking Glossary of Genetic Terms. Available at: www.genome.gov/ Pages/Hyperion/DIR//VIP/Glossary/Illustration/codon.shtml.

The Universal Genetic Code

- 61 of the codons code for an amino acid

- 3 of the remaining codons are "STOP" codons that do not code for an amino acid. They just signal that translation is over.

- 1 codon that codes for the amino acid "methionine" is also the "START" codon. Methionine is always the first amino acid in a protein.

Second letter

First letter	U	C	A	G	Third letter
U	UUU } Phe UUC } UUA } Leu UUG }	UCU } UCC } Ser UCA } UCG }	UAU } Tyr UAC } UAA Stop UAG Stop	UGU } Cys UGC } UGA Stop UGG Trp	U C A G
C	CUU } CUC } Leu CUA } CUG }	CCU } CCC } Pro CCA } CCG }	CAU } His CAC } CAA } Gln CAG }	CGU } CGC } Arg CGA } CGG }	U C A G
A	AUU } AUC } Ile AUA } AUG Met	ACU } ACC } Thr ACA } ACG }	AAU } Asn AAC } AAA } Lys AAG }	AGU } Ser AGC } AGA } Arg AGG }	U C A G
G	GUU } GUC } Val GUA } GUG }	GCU } GCC } Ala GCA } GCG }	GAU } Asp GAC } GAA } Glu GAG }	GGU } GGC } Gly GGA } GGG }	U C A G

The Universal Genetic Code

This is called a "universal" code because **ALL LIFE uses the same genetic code**... from the smallest bacteria or virus to the largest animal or tree.

This tells us that **this code goes back billions of years**, in the first cell...or even before that.

If there were alternative codes that could work, they would have appeared in nature.

There are very minor alterations, but they are rare and insignificant in their effect.

1 What is a codon?

- A a 3 base sequence on tRNA
- B a 3 base sequence on mRNA
- C a 3 base sequence on DNA
- D B and C

2 The codon UAA specifies:

- A Adenine
- B Glycine (Gly)
- C STOP
- D Arginine
- E Valine

Second position

<table>
<tr><th>First position</th><th>U</th><th>C</th><th>A</th><th>G</th><th>Third position</th></tr>
<tr><td rowspan="4">U</td><td rowspan="2">phenyl-alanine</td><td rowspan="4">serine</td><td rowspan="2">tyrosine</td><td rowspan="2">cysteine</td><td>U</td></tr>
<tr><td>C</td></tr>
<tr><td rowspan="2">leucine</td><td>stop</td><td>stop</td><td>A</td></tr>
<tr><td>stop</td><td>tryptophan</td><td>G</td></tr>
<tr><td rowspan="4">C</td><td rowspan="4">leucine</td><td rowspan="4">proline</td><td rowspan="2">histidine</td><td rowspan="4">arginine</td><td>U</td></tr>
<tr><td>C</td></tr>
<tr><td rowspan="2">glutamine</td><td>A</td></tr>
<tr><td>G</td></tr>
<tr><td rowspan="4">A</td><td rowspan="3">isoleucine</td><td rowspan="4">threonine</td><td rowspan="2">asparagine</td><td rowspan="2">serine</td><td>U</td></tr>
<tr><td>C</td></tr>
<tr><td rowspan="2">lysine</td><td rowspan="2">arginine</td><td>A</td></tr>
<tr><td>methionine</td><td>G</td></tr>
<tr><td rowspan="4">G</td><td rowspan="4">valine</td><td rowspan="4">alanine</td><td rowspan="2">aspartic acid</td><td rowspan="4">glycine</td><td>U</td></tr>
<tr><td>C</td></tr>
<tr><td rowspan="2">glutamic acid</td><td>A</td></tr>
<tr><td>G</td></tr>
</table>

answer

3 The codon GGG specifies:

- A Adenine
- B Glycine
- C STOP
- D Arginine
- E Valine

Second position

First position	U	C	A	G	Third position
U	phenyl-alanine	serine	tyrosine	cysteine	U
					C
	leucine		stop	stop	A
			stop	tryptophan	G
C	leucine	proline	histidine	arginine	U
					C
			glutamine		A
					G
A	isoleucine	threonine	asparagine	serine	U
					C
			lysine	arginine	A
	methionine				G
G	valine	alanine	aspartic acid	glycine	U
					C
			glutamic acid		A
					G

answer

4 The codon GAC specifies:

- A Adenine
- B Glycine
- C STOP
- D Arginine
- E Aspartic Acid

Second position

First position	Third position	U	C	A	G
U	U	phenyl-alanine	serine	tyrosine	cysteine
	C				
	A	leucine		stop	stop
	G			stop	tryptophan
C	U	leucine	proline	histidine	arginine
	C				
	A			glutamine	
	G				
A	U	isoleucine	threonine	asparagine	serine
	C				
	A			lysine	arginine
	G	methionine			
G	U	valine	alanine	aspartic acid	glycine
	C				
	A			glutamic acid	
	G				

answer

5 Why is Methionine the very first amino acid in all proteins?

- A because it is coded by the stop codon
- B because it is coded for by AUG which is the start codon
- C Methionine is coded for by more than one codon
- D none of the above

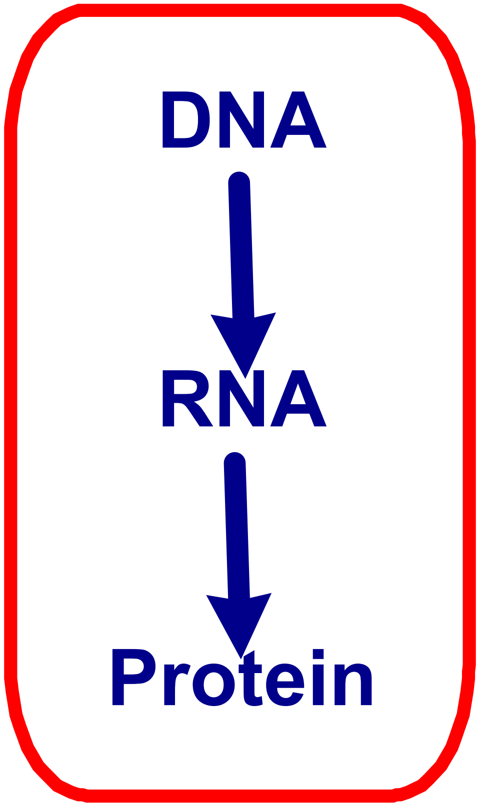
answer

Steps of Gene Expression

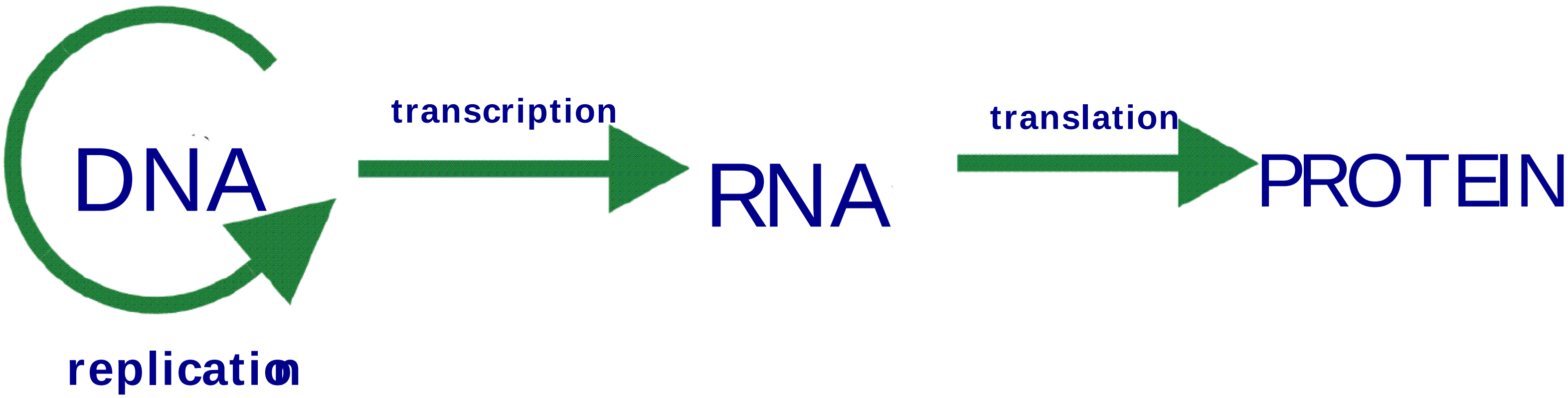
Gene expression occurs in two steps:

1. The gene is copied from DNA into RNA through a process called **transcription.**

2. The RNA build a protein in a process called **translation.**



The Central Dogma



The processes of replication, transcription and translation are so critical that they are called the **Central Dogma** of Biology.

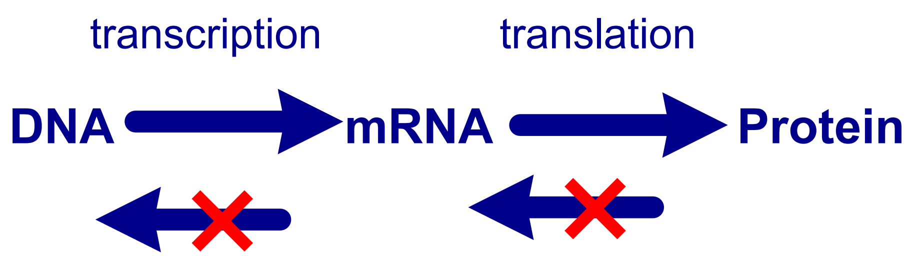
A "Dogma" is a postulate; an idea; a philosophy.

It is "Central" because it is what life is based on.

The Central Dogma

The Central Dogma is a one way process.

Changes in DNA affect mRNA and protein.



But changes in proteins or mRNA do not affect the DNA.

This will have important implications when we study genetics.

6 What is meant by "gene expression"?

- A making the protein or RNA coded in the nucleic acid
- B making amino acids so they can be made into protein
- C making tRNA only
- D folding of the protein

answer

7 Which one of the following sequences best describes the Central Dogma of biology?

- A RNA to DNA to RNA to Protein
- B DNA to RNA to Protein
- C Protein to RNA to DNA
- D DNA to Amino Acid to RNA to Protein

answer

Gene Expression: Transcription

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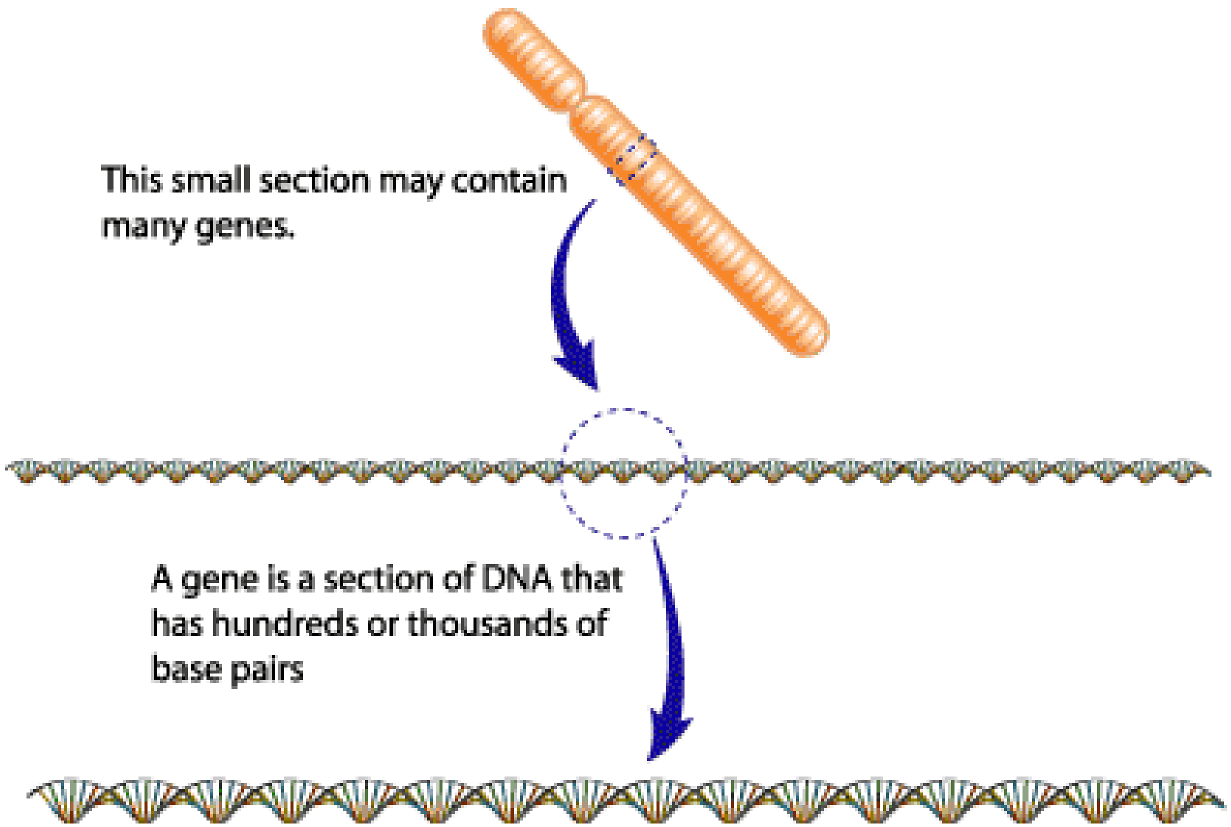
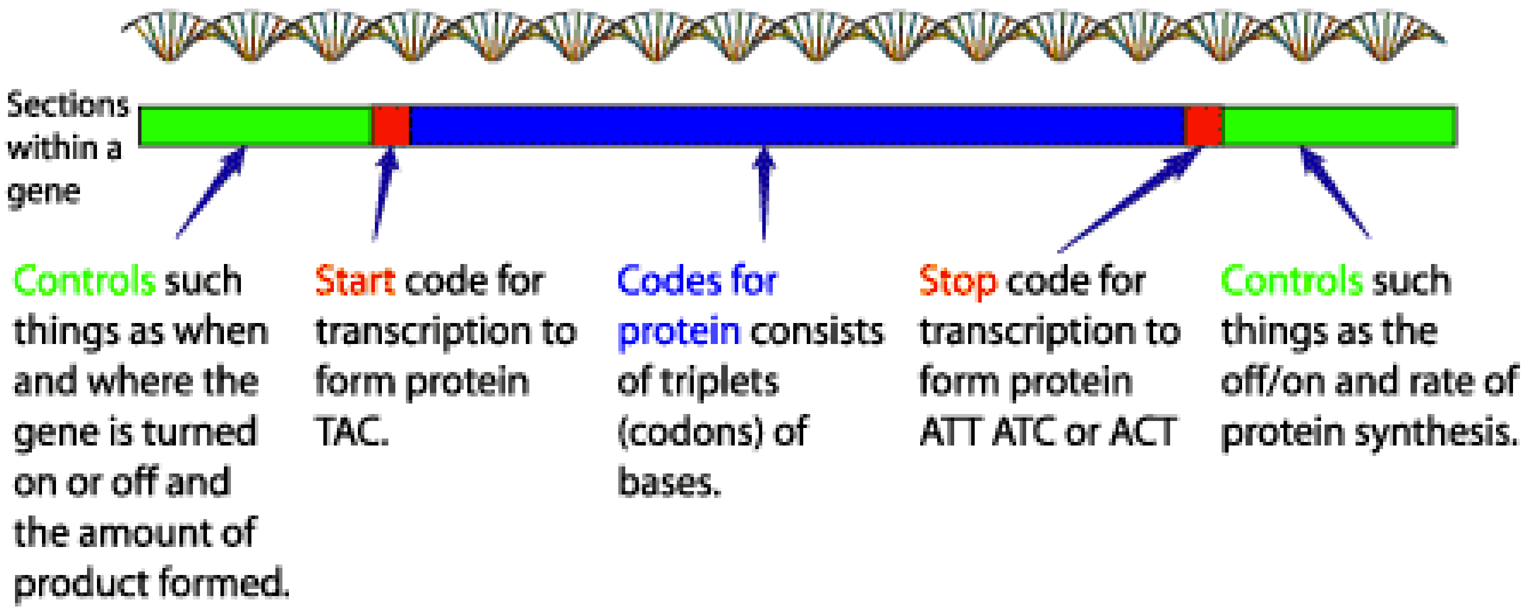
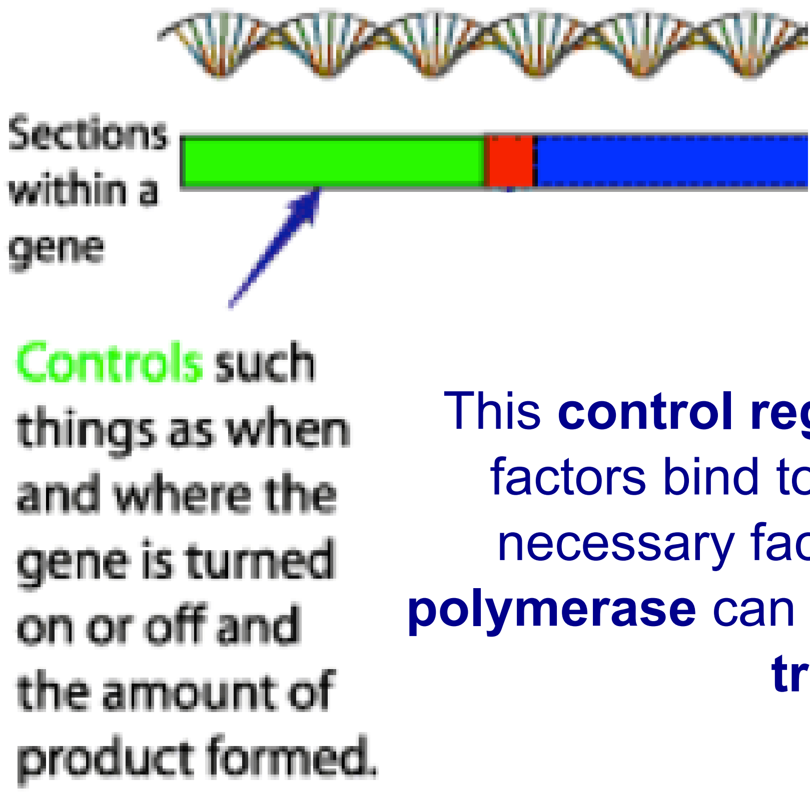
Gene Anatomy



Gene Anatomy



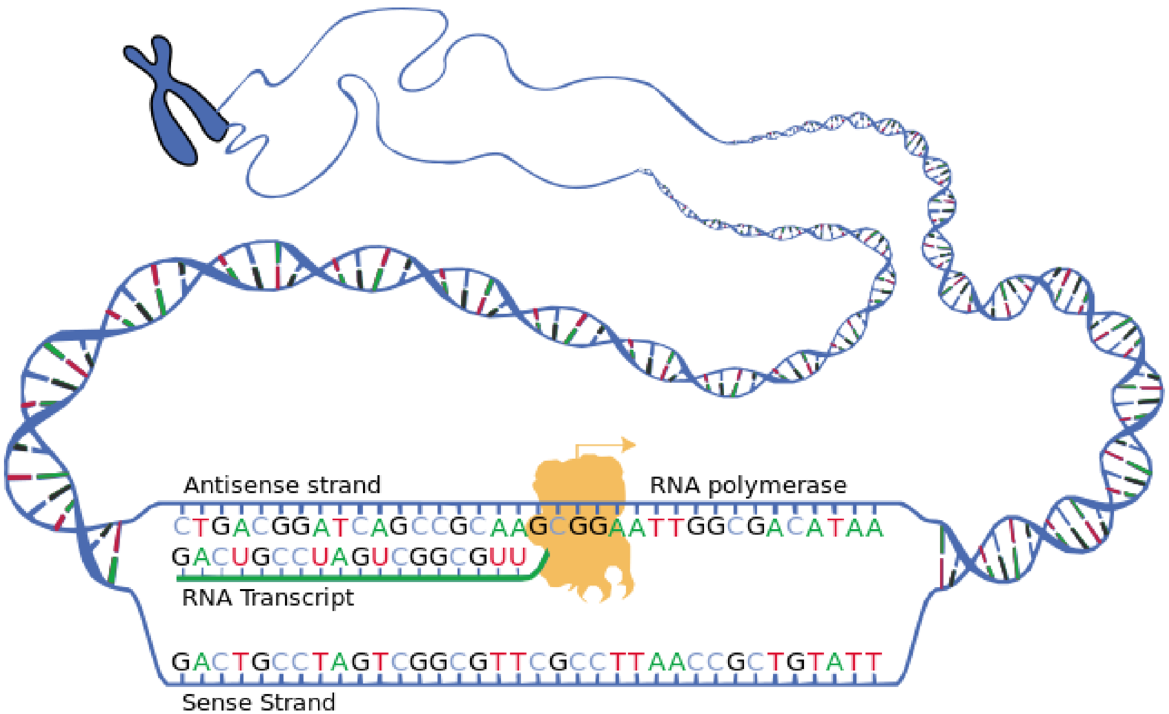
Gene Anatomy



This **control region** is where transcription factors bind to the gene. When all the necessary factors are combined **RNA polymerase** can bind to the gene and initiate **transcription**.

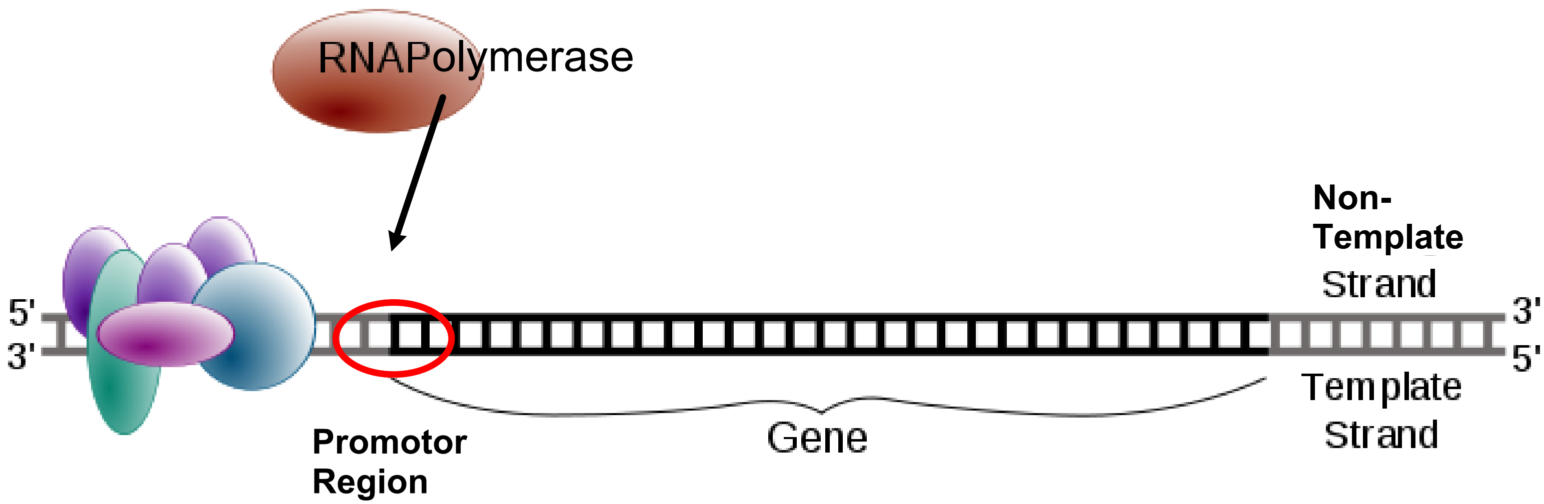
The First Step - Transcription

Transcription is the first step of gene expression, in which a particular segment of DNA is copied into RNA by the enzyme **RNA polymerase.**



Transcription - Pre-Initiation

The first step of transcription is called pre-initiation. RNA polymerase and cofactors bind to DNA and unwind it, creating an **initiation bubble**. This is a space that grants RNA polymerase access to a single strand of the DNA molecule.



Template vs. Non-Template Strands

The RNA polymerase never attaches to the strand that actually contains the gene.

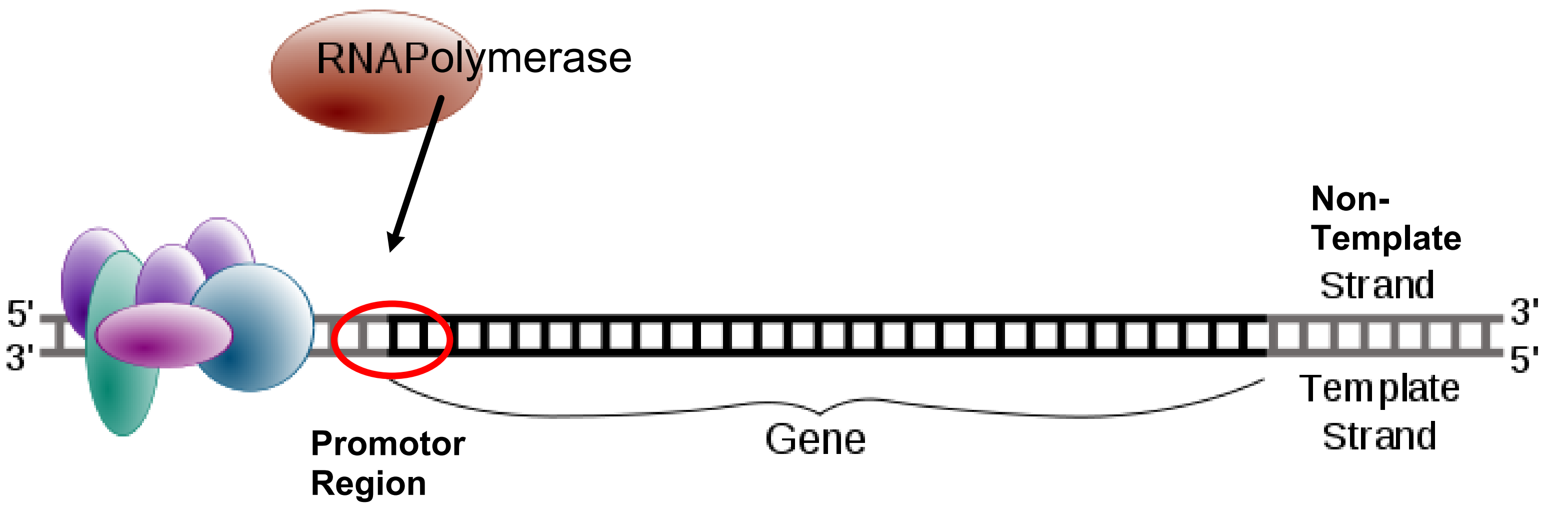
The strand with the genes is called the "non-template strand."
This **IS NOT** the strand that is transcribed.

The other strand is the mirror image of the first, it carries the mirror image of the gene, not the gene itself. It is called the "template strand."
This **IS** the strand where the RNA polymerase attaches.

Transcription - Initiation

To begin, an enzyme called RNA Polymerase attaches to the Promoter region on the DNA.

The Promoter is a specific sequence of bases that the RNA polymerase recognizes.



Transcription: DNA Strands

This makes sense in that the RNA will be the mirror image of the DNA it is transcribed from. And the non-coding strand is the mirror image of the gene.

5' ...A T G G C C T G G A C T T C A... 3' non-template strand of DNA

3' ...T A C C G G A C C T G A A G T... 5' template strand of DNA

transcription of template strand

5' ...A U G G C C U G G A C U U C A... 3' RNA

Note:

the non-template strand of DNA (the gene) matches the new RNA strand

8 The strand that is transcribed into RNA is called the

- A Template Strand
- B Non Template Strand
- C RNA Strand
- D Amino Acid Strand

answer

9The transfer of genetic material from DNA to RNA is called:

- A translation
- B transcription
- C elongation
- D promotion

answer

10 Genes are located on the

- A Template Strand
- B Non Template Strand
- C RNA Strand
- D Amino Acid Strand

answer

11 What is the function of the promoter sequence on the DNA?

- A it is where the RNA polymerase recognizes and binds to initiate transcription
- B it is where the RNA gets copied
- C it where transcription terminates
- D it is where the RNA polymerase binds to on the 3' end of the DNA initiating transcription

answer

12 The strand that is NOT transcribed into RNA is called the

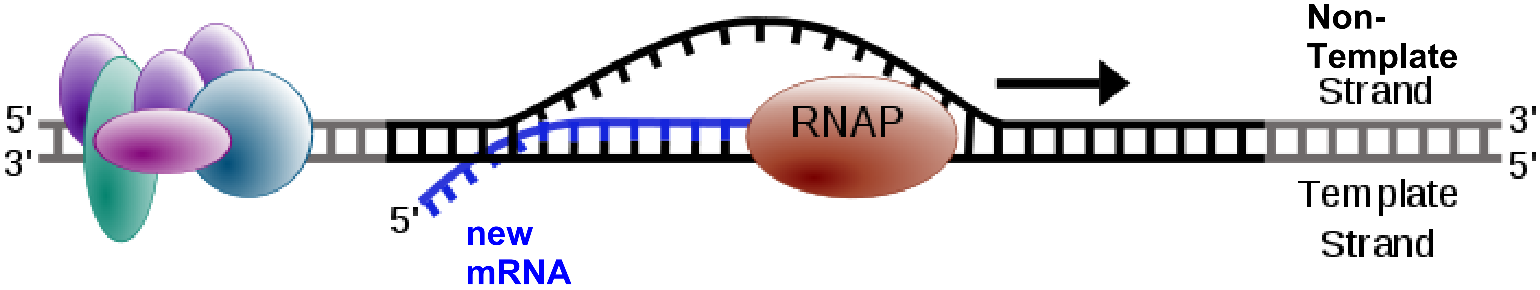
- A Template Strand
- B Non Template Strand
- C RNA Strand
- D Amino Acid Strand

answer

Transcription - Elongation

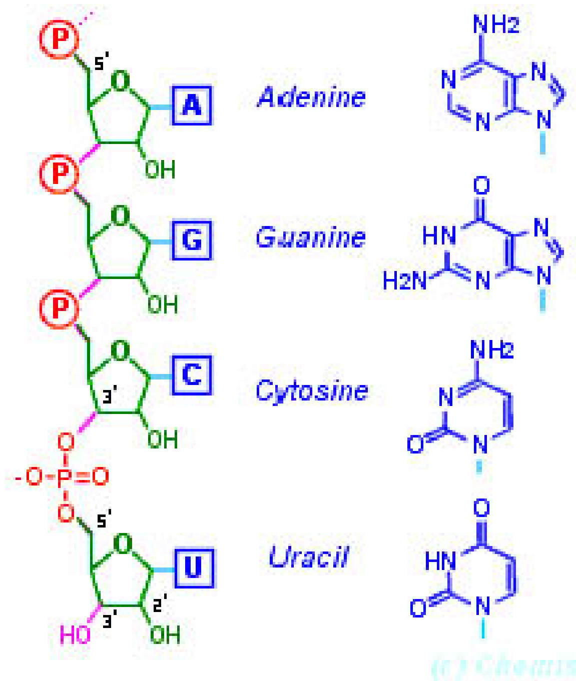
To make the RNA strand, **RNA Polymerase** runs down the DNA template strand reading the bases and bringing in the new RNA nucleotides with the proper complementary bases.

As the RNA Polymerase runs down the DNA, it actually unwinds the DNA!



Base Pairing

Transcription is made possible by the fact that the different bases are attracted to one another in pairs.



RNA DNA

A bonds with T

U bonds with A

G bonds with C

C bonds with G

Note: In DNA replication adenine paired with thymine, in DNA transcription uracil is now paired with adenine. Remember that RNA does not contain thymine as a nucleotide base.

Transcription

Just like in DNA replication,

RNA is made from the 5' end to the 3' end.

DNA ("template strand") 3' TACGGCATTA 5'

RNA 5' AUGCCGUAAU 3'

being made in 5'--------->3' direction.

13 If the template strand of DNA is 5' ATAGATACCATG 3', which is the RNA strand produced from transcription

- A 5' UAUCUAUGGUAC 3'
- B 5' TATCTATGGTAC 3'
- C 3' UAUCUAUGGUAC 5'
- D 3' TATCTATGGTAC 5'

answer

14 If the template strand of DNA is 5' AAAGACACTATT 3', which is the RNA strand produced from transcription

- A 5' UUUCUGUGAUAA 3'
- B 5' TTTCTGTGATAA 3'
- C 3' UUUCUGUGAUAA 5'
- D 3' TTTCTGTGATAA 5'

answer

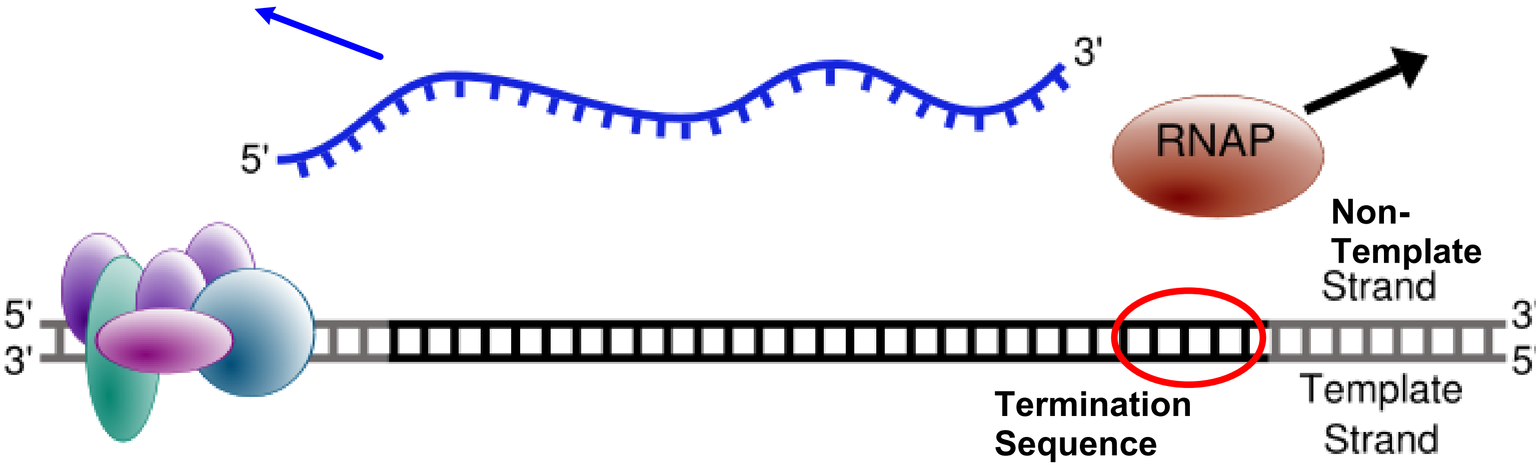
15 If the non-template strand of DNA is 3' ACGATTACT 5', which is the RNA strand produced through transcription

- A 3' TGCTAATGA 5'
- B 3' UGCUAAUGA 5'
- C 5' UGCUAAUGA 3'
- D 5' ACGAUUAGU 3'

answer

Transcription - Termination

RNA Polymerase gets to a sequence on the DNA called a Termination Sequence. This sequence signals the RNA Polymerase to STOP transcription.



The RNA Polymerase falls off the DNA.
The new RNA strand separates from the DNA.
The DNA recoils into a helix.

Click here to see an animation of transcription

DNA Replication vs. Transcription

DNA Replication	Transcription
Two new double-stranded DNA are produced	One new single-stranded RNA is produced
Adenine from the parent strand bonds with thymine on the new daughter strand of DNA	Adenine on the DNA strand bonds with uracil on the new RNA strand.
The whole DNA molecule is replicated	Only the strand with the code for the gene is transcribed.

Synthesis of both occur in the 5' to 3' direction

Translation

Translation is the process by which RNA strands are read to build proteins.

Translate means to convert something from one language to another, you can remember that the process of making protein from RNA is called **translation** because the "language" of nucleotides" is being changed to the "language" of amino acids.

Three Types of RNA

Translation requires 3 types of RNA that are created using transcription.

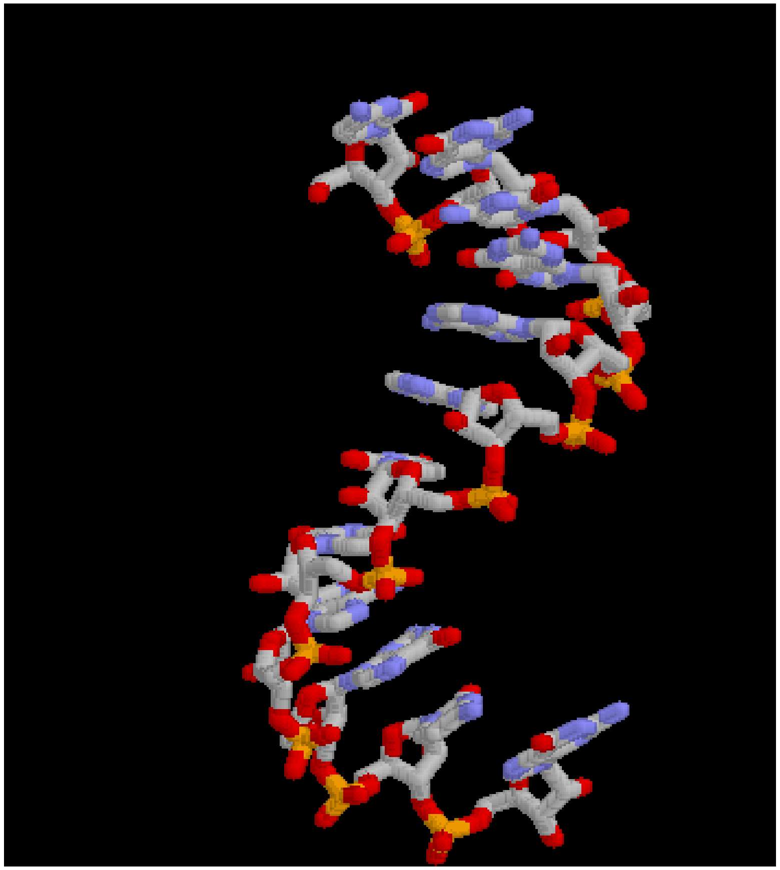
1. mRNA or messenger RNA, carries the information for protein synthesis. This type of RNA is key to The Central Dogma.

2. rRNA or ribosomal RNA, is a catalyst for protein synthesis

3. tRNA or transfer RNA, helps in the assembly of amino acids during protein synthesis

Messenger RNA (mRNA)

The specific RNA that contains the protein's information from DNA is called Messenger RNA (mRNA); it carries the genetic message to ribosomes, where it is translated.

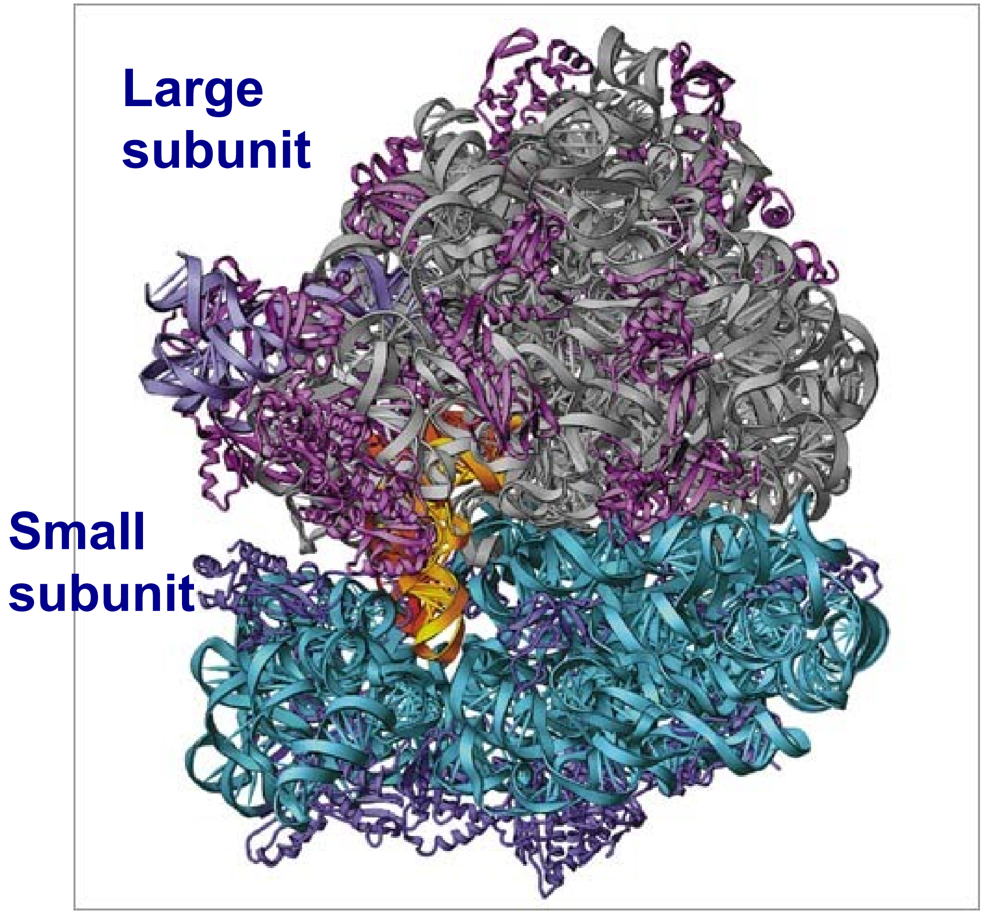


Ribosomal RNA (rRNA)

Ribosomal RNA (rRNA) and some additional proteins make up the ribosome.

The ribosome includes two subunits: one small, and one large.

During translation, the ribosome catalyzes the reaction that makes covalent bonds between amino acids, thus building the protein.



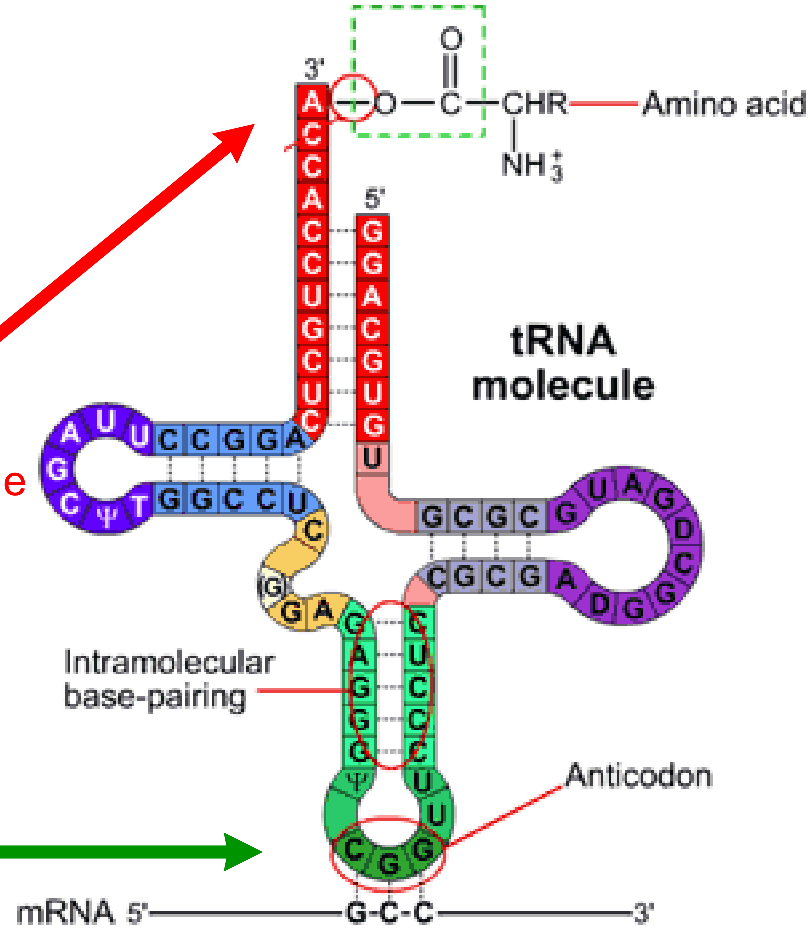
Transfer RNA (tRNA)

Transfer RNA (tRNA) carries amino acids to the ribosome so that the ribosome can covalently bond them together to form the protein.

RNA, being single stranded, can fold in on itself. In tRNA, the RNA folds into a t-shape.

The **Amino Acid Attachment Site** is where the amino acid will attach to the tRNA.

The **Anticodon** Loop is a 3 base sequence on the tip that is complementary to the codon on the mRNA.



16 What 2 components is a ribosome made of?

- A rRNA and DNA
- B rRNA and carbohydrates
- C rRNA and proteins
- D both b and c

answer

17 What is the function of the ribosome?

- A to make an ionic bond between amino acids
- B to make a covalent/peptide bond between amino acids thus building the protein
- C to make hydrogen bonds
- D to make RNA

answer

18 What does the "t" in tRNA stand for?

- A "transfer"- it transfers the amino acid to the ribosome and mRNA codon
- B it refers to the shape
- C "transfer"- it transfers the protein to the DNA
- D Both B and C

answer

19 Why does tRNA fold into its specific shape?

- A The sequence and bonding of its amino acids
- B The sequence of and bonding of nucleotides
- C Its protein structure
- D A and B
- E A and C

answer

Gene Expression: Translation

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Translation - An Overview

All the pieces are ready to begin translation:

a coded strand of mRNA
a set of 20 amino acids
ribosomes
tRNA to match all the amino acids

Translation - An Overview

tRNAs bond to the amino acid specified by their anti-codon.

The opposite side of each tRNA, the anti-codon, bonds to the matching codon on the mRNA, creating a string of amino acids in the proper sequence.

The ribosome makes covalent bonds between the amino acids.

The result is a protein chain with the specified sequence of amino acids.

Proteins: Words
Amino Acids :: Letters

The length and sequence of these amino acids allow all the proteins in the world to be created from only 20 amino acids.

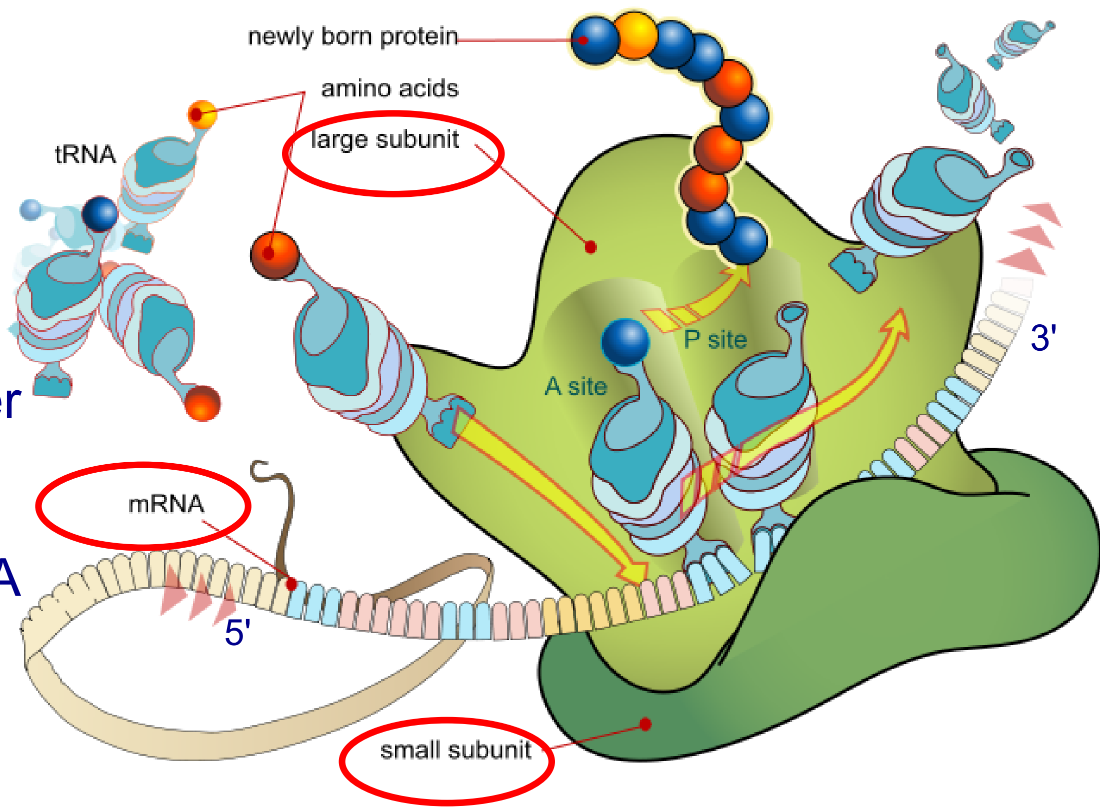
This is very similar to how all the words can be created from only 26 letters in the alphabet.

Translation - Initiation

The small subunit of the ribosome attaches to the mRNA at the bottom of the start codon(at the 5' end).

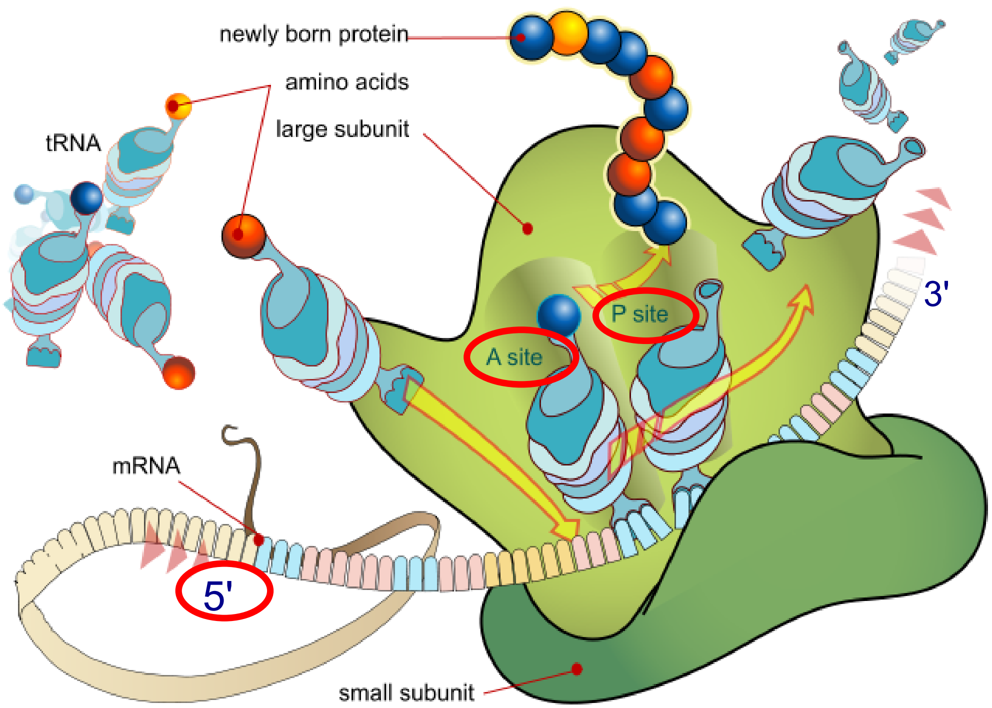
Then the large subunit of the ribosome comes in over the top.

The result is that the mRNA is "sandwiched" between the mRNA at the start codon (and the second codon as well!)



Translation - Initiation

The ribosome goes to the 5' end of the mRNA because the 5' end is the beginning of where the gene on the DNA was transcribed into mRNA.



Also notice that there are 2 sites within the ribosome.

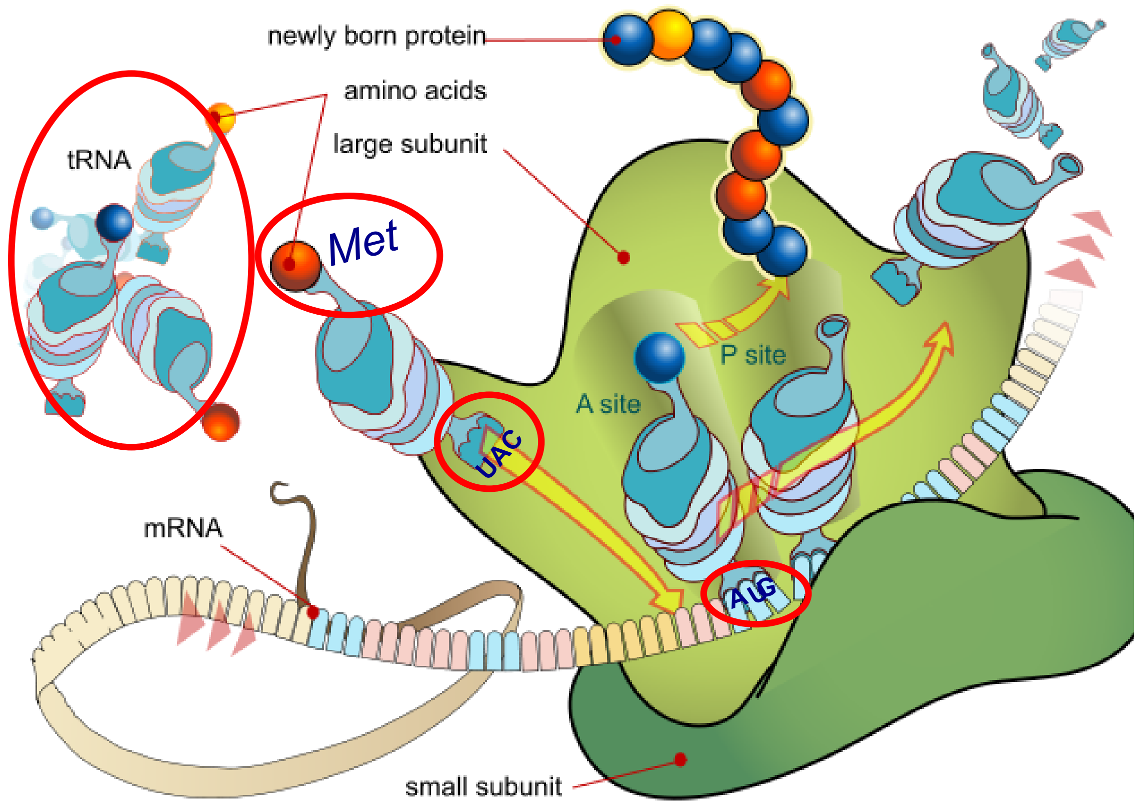
- The P-site - where the new protein will emerge
- The A-site - where the Amino Acids are delivered in

Translation - Initiation

The tRNAs, hydrogen bonded to their specific amino acids, surround the ribosome.

As the leading edge of the mRNA, with the starting code AUG, is exposed in the A site,

the tRNA with the code UAC enters the site and hydrogen bonds with it, carrying methionine into the ribosome.



Translation - Initiation

The methonine is removed from the tRNA and stays in the ribosome to be bonded with the next amino acid. The tRNA leaves the ribosome so another tRNA can enter.

Each tRNA will carry the appropriate amino acid into the ribosome to be bonded in the proper sequence, since each tRNA anticoding site matches the coding site on the mRNA, which is located at the A site of the ribosome.

Because each tRNA has an anticoding sequence it complimentary base pairs with the codon on the mRNA.

20 How does the anticodon on the tRNA and the codon on the mRNA match up?

- A by hydrogen bonding/complimentary base pairing
- B by ionic bonding
- C by peptide bonds
- D none of the above

answer

21 What is the P site of the ribosome?

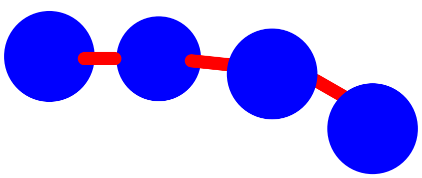
- A it is where the amino acids are delivered in
- B it is where the protein or peptide will emerge
- C it where the tRNA's will deliver in the next amino acid after each translocation
- D it is where the proteins fold into their 3-d shape

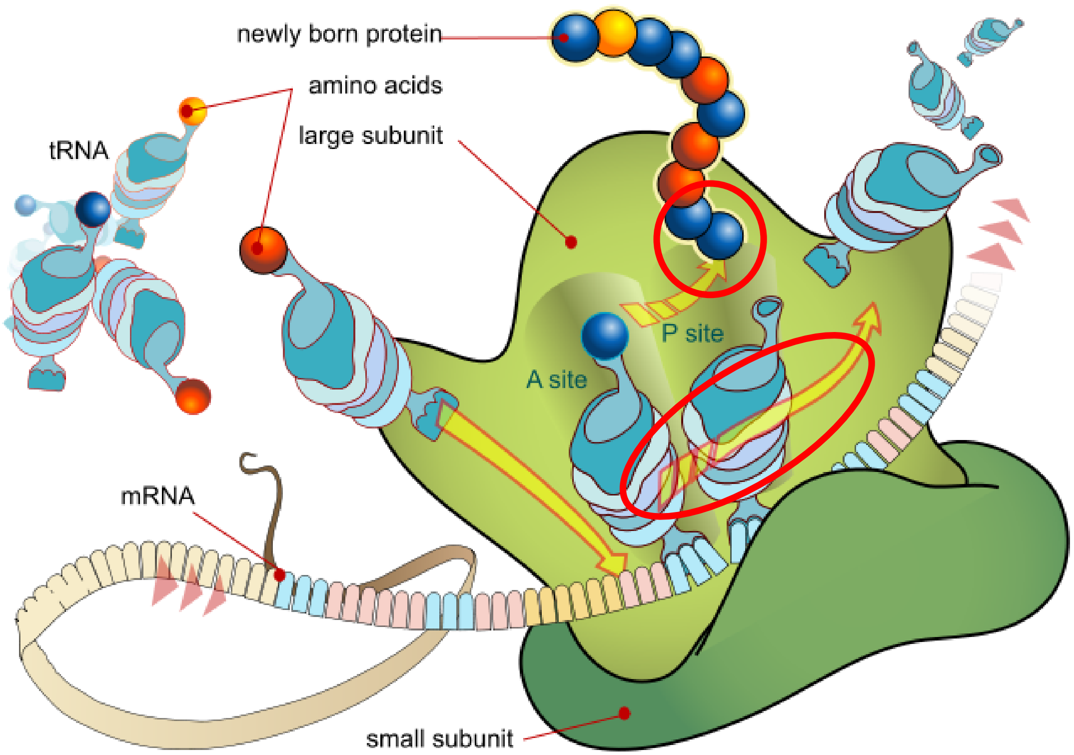
answer

Translation - Elongation

The 2nd tRNA with its amino acid is delivered into the A-site in the ribosome.

The ribosome catalyzes a covalent bond between the amino acids.



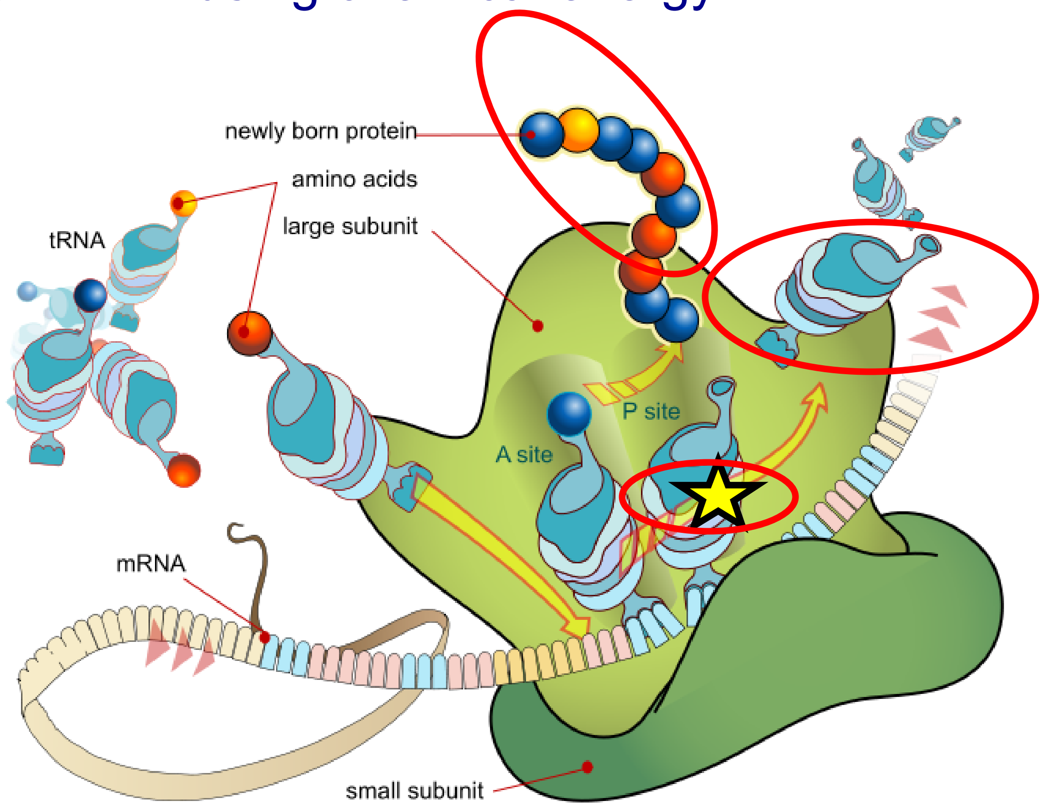


Translation - Elongation

The ribosome moves the mRNA using chemical energy.

The tRNA that was in the A-site moves to the P-site and the tRNA that was in the P-site separates from its amino acid.

Notice the protein emerging from the P-site!



Translation - Elongation

Elongation continues by adding one amino acid after another.

Each amino acid is delivered to the A-site by its matching tRNA.

The ribosome makes a peptide bond between the 2 amino acids in the P and A sites.

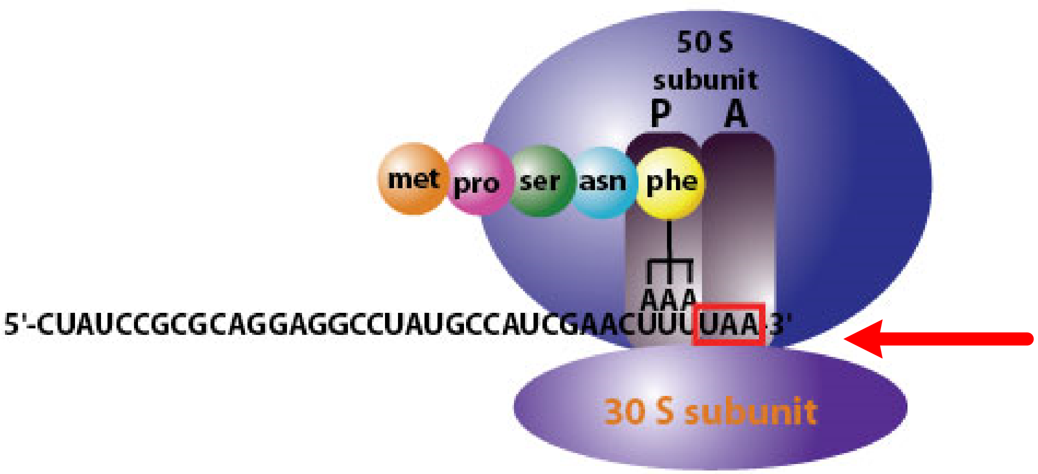
until......

Translation - Termination

The ribosome reaches a STOP codon. Remember that STOP codons do not code for amino acids. This signals the end of translation.

The protein is complete.

The 2 subunits (large and small) separate from each other.

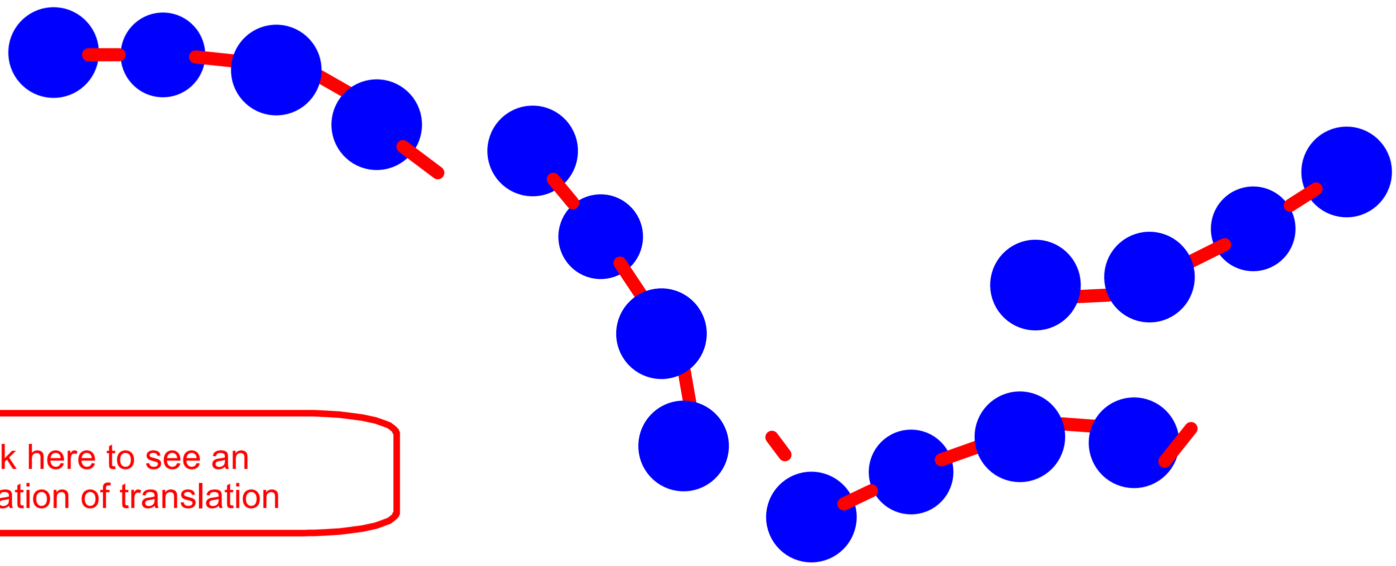


UAA is 1 of the 3 possible STOP codons.

Translation - Termination

The Result- A protein in its "primary sequence".

Remember that Primary level (1^0) of protein structure is the sequence of amino acids.



Click here to see an animation of translation

22 What is the first event of translation?

- A the tRNA comes in
- B the small subunit of the ribosome and the 1st tRNA brings in Methionine to the start codon
- C elongation happens
- D the large subunit of the ribosome comes in

answer

23 What is the first step of translation called?

- A transcription
- B elongation
- C termination
- D initiation

answer

24 What is the function of the ribosome in translation?

- A it makes a peptide/covalent bond using the energy from translocation
- B it makes hydrogen bonds between the codons
- C it makes covalent/peptide bonds between the codons
- D none of the above

answer

25 What does termination in translation involve?

- A translocation of the ribosome
- B the ribosome gets to a stop codon and the small and large subunits of the ribosome separate
- C RNA polymerase falls off the DNA
- D a tRNA brings in an amino acid

answer

26 What is translation?

- A the assembly of the amino acids from the protein code
- B assembly of amino acids coded for by the mRNA codons
- C the making of mRNA
- D assembly of codons from DNA template

answer

Genetics of Development

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Genetics of Development

The most difficult task for the genes of a multi-celled complex organism is to develop the organism from a single cell.

Genetics of Development

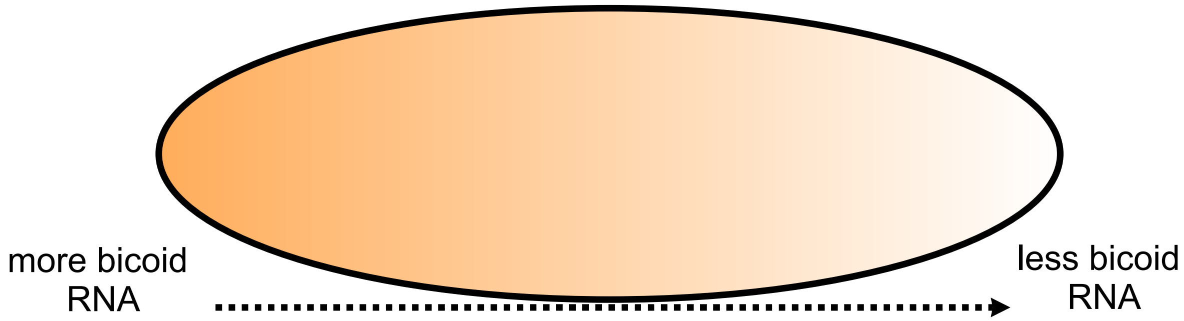
Development is a process where the products of some genes turn other genes on or off. Even before fertilization, development is occurring. The egg supplies information to establish a **molecular coordinate system**. This coordinate system provides a way of determining which future cells will become differentiated into certain parts of the organism.

Genetics of Development

One of the best understood examples of molecular coordination in development is the patterning along the future anterior and posterior of the fruit fly, Drosophila melanogaster. There are fundamental genes that give way to the developmental structure of the fly. The genes that are involved are: **maternal effect genes and homeotic genes.**

Fruit Fly Egg

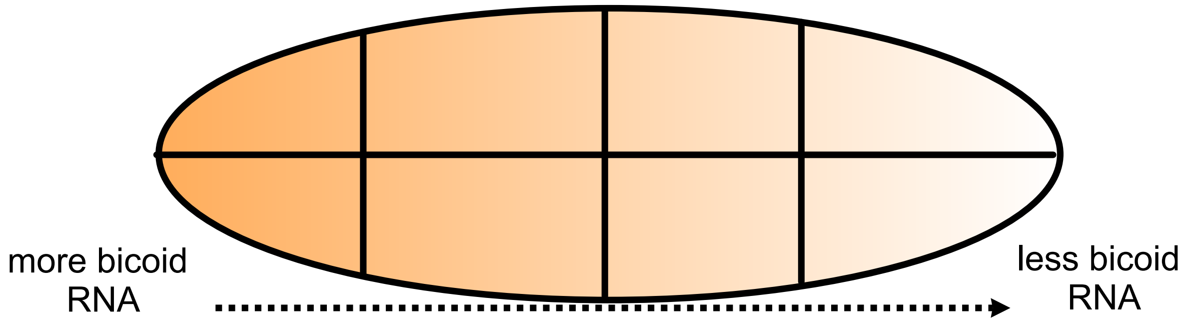
Before a fruit fly egg is fertilized, genes are being expressed. One of those genes is known as the **bicoid** gene. This maternal effect gene produces RNA molecules that float in the cytoplasm. Its uneven distribution (due to the location of the nucleus) creates a gradient.



Fruit Fly Embryo

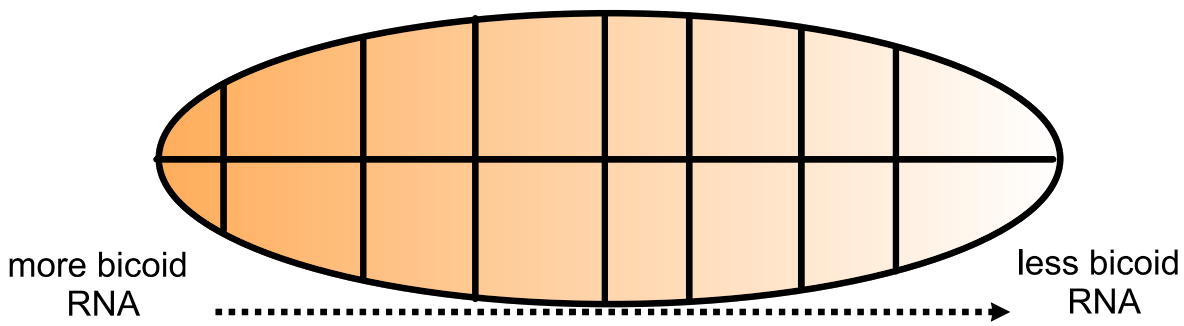
After the egg is fertilized it begins to divide by mitosis but the there is no increase in the amount of cytoplasm as these first quick divisions occur.

Each of these new **stem cells** contains a slightly different concentration of bicoid RNA.



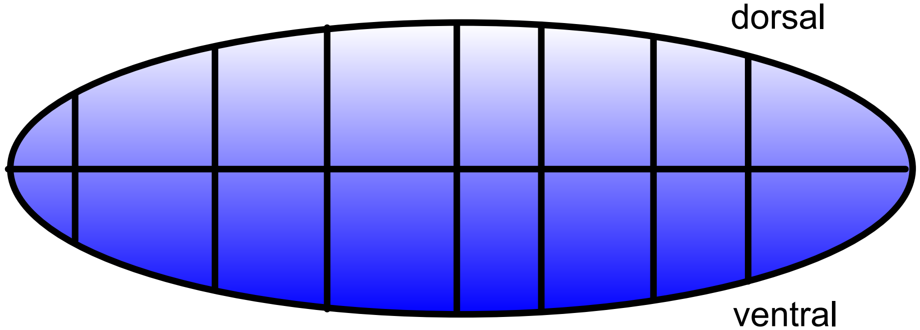
Fruit Fly Embryo

This coordinate system provides a way of determining which end is **posterior** or **anterior**. The cells with high bicoid concentration will activate genes for tissues in the head of the organism. Decreasing amounts lead to the proper order of segments and very low will become the tail.



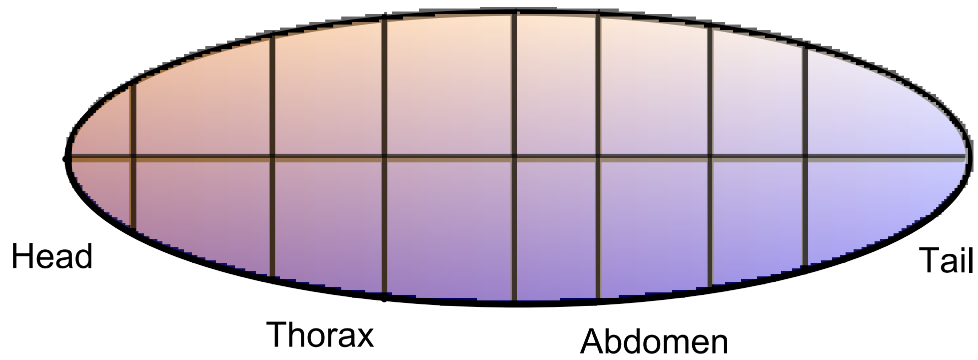
Fruit Fly Embryo

Similar gradients will also determine **dorsal** and **ventral** of the developing fly.



Fruit Fly Embryo

The unique concentration of several maternal effect gene products ensures each cell in the developing embryo has different transcription factors and gene expression.



Stem Cells

Recall that before cells become differentiated into their specific type (bone, neuron, muscle, etc) they are a kind of "blank" cell called a stem cell.

Stem cells contain the entire genome of the organism but have not received the signals needed to become a specific type of cell.

Bicoid concentration is the first step in a cascade of reactions that result in the transformation of stem cells into specialized cells.

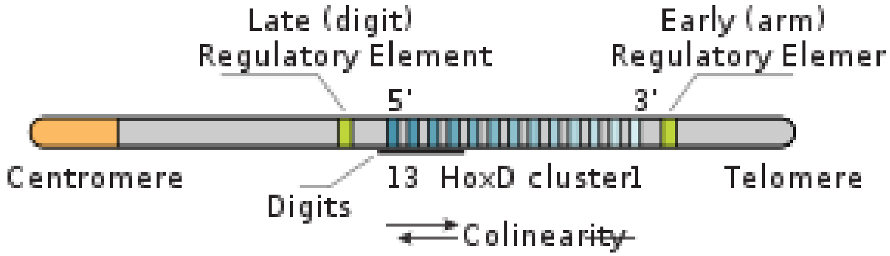
Homeotic Genes

Maternal effect genes act as transcription factors, turning other genes on or off and, as a result, formed the segmented pattern of the Drosophila embryo.

The **homeotic genes**, having been turned on or off by the cascade of gene expression started by the maternal effect genes, also produce transcription factors. They influence the expression of numerous other genes and determine the identity of the segment they are in.

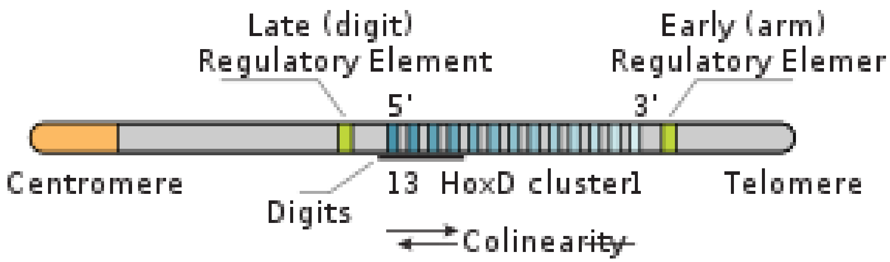
Hox genes

It has been found that the sequence of homeotic genes in fruit flies known as the **Hox genes** are lined up in exactly the same order as the part of the fly they affect. The first gene affects the mouth, the second the face, the third the top of the head and so on up until the eighth and final gene that affects the abdomen.



Hox genes

The proteins that Hox genes produce determine the type of segment structures such as legs, antennae, and wings in fruit flies or the different vertebrate ribs in humans.



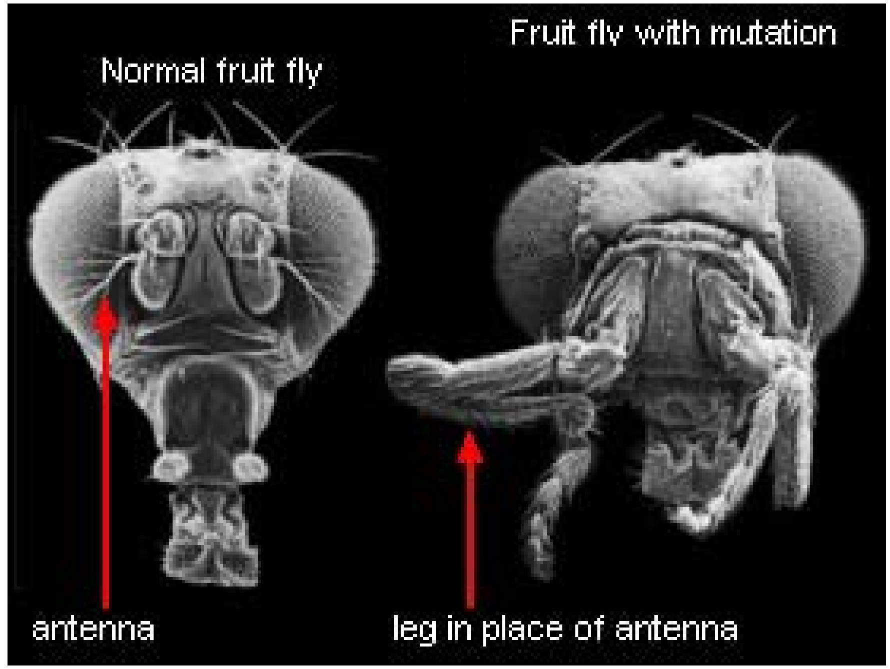
27 A scientist was able to produce a fruit fly with no head, but a tail at each end of its body. He most likely...

- A Disrupted the production of bicoid RNA in the egg before fertilization
- B Saturated the egg with bicoid RNA before fertilization

answer

28 The phenotype shown here is most likely a result of a mutation in

- A the bicoid gene
- B a hox gene
- C a homeotic gene
- D a maternal effect gene



answer

Mutations

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Mutations

A **mutation** is a permanent change in the DNA sequence of a gene. Mutations in a gene's DNA sequence can alter the amino acid sequence of the protein encoded by the gene.

Like words in a sentence, the DNA sequence of each gene determines the amino acid sequence for the protein it encodes. The DNA sequence is interpreted in groups of three nucleotide bases, codons. Each codon specifies a single amino acid in a protein.

Substitution Mutations

When a nucleotide in a gene is copied incorrectly during DNA replication, one nucleotide can be substituted with another. This results in the incorrect amino acid sequence, changing the structure of the protein.

Correct DNASequence: AAA TTT CCC GGG AAA TTT CCC GGG

Correct RNA Transcript: UUU AAA GGG CCC UUU AAA GGG CCC

Correct Polypeptide: Phe - Lys - Gly - Pro - Phe - Lys - Gly - Pro

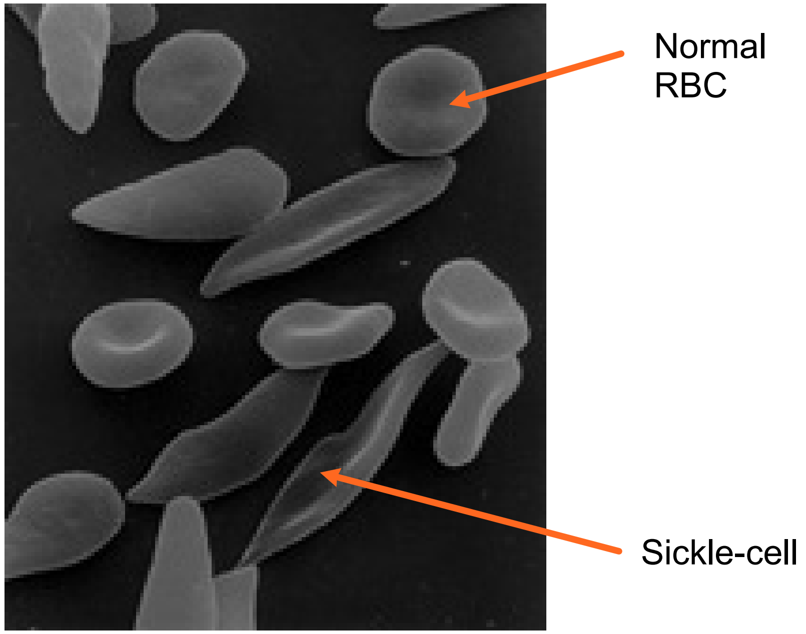
Substitution mutation: AAA TTT CCC GGG ATA TTT CCC GGG

Resulting Transcript: UUU AAA GGG CCC UAU AAA GGG CCC

Resulting Polypeptide: Phe - Lys - Gly - Pro - Tyr - Lys - Gly - Pro

Sickle-Cell Disease

Sickle-cell disease is a blood disorder caused by a substitution mutation. It is characterized by red blood cells that assume an abnormal, rigid, sickle shape.



Sickling decreases the cells' flexibility and ability to carry oxygen. It results in a risk of various complications. The sickling occurs because of a mutation in the hemoglobin gene.

Reading Frame Shifts

We can think about the DNA sequence of a gene as a sentence made up entirely of three-letter words.

Thesunwashot

If you were to split this sentence into individual three-letter words, you would probably read it like this:

The sun was hot

If this sentence represents a gene then each letter corresponds to a nucleotide base, and each word represents a codon. If you shifted the three-letter**reading frame** it would result in a sentence which is not understandable...

_ _T hes unw ash ot_

Or

Th esu nwa sho t _

Insertion and Deletion Mutations

When a nucleotide in a gene is copied incorrectly during DNA replication, nucleotide can be added or deleted. This results in a reading frame shift and the incorrect amino acid sequence, changing the structure of the protein.

Correct DNASequence: AAA TTT CCC GGG
RNA Transcript: UUU AAA GGG CCC
Correct Polypeptide: Phe - Lys - Gly - Pro

Insertion mutation: AAA ATT TCC CGG G_ _
Resulting Transcript: UUU UAA AGG GCC C_ _
Resulting Polypeptide: Phe - STOP

Deletion mutation: AAT TTC CCG GG_
Resulting Transcript: UUA AAG GGC CC_
Resulting Polypeptide: Leu - Lys - Gly ?

A

Tay-Sachs Disease

Tay-Sachs disease is a result of insertion and deletion mutations. It causes a relentless deterioration of mental and physical abilities that commences around six months of age and usually results in death by the age of four.

The disease results from mutations on chromosome 15 in humans. These mutations include base pair insertions and deletions. Each of them alters a protein product, and thus inhibits the function of enzymes in some manner.

Junk DNA and Silent mutations

Junk DNA is the term given to DNA that does not code for proteins. It is the space between genes. A large portion of the DNA in humans is junk DNA, better than 90% in some estimates. That means if a mutation happens in 90% of a humans DNA then the person is unaffected. This is known as a **silent mutation**.

Another way to make a mutation silent is the redundancy in the genetic code. Each amino acid has more than one possible codon. So, if a substitution occurs, the same amino acid may still be coded. This reduces the possibility of a mutation located in a gene causing a change in the protein.

Ala - GCU, GCC, GCA, GCG
Leu - UUA, UUG, CUU, CUC, CUA, CUG
Arg - CGU, CGC, CGA, CGG, AGA, AGG

Are mutations always bad?

Not necessarily...

Very rarely a mutation will cause an individual to become stronger than the rest of its population. Sometimes a mutation can create a polar bear with thicker fur, or a giraffe with a longer neck.

These would be advantages to the individual and they may become fitter to survive in their environment.

Mutagens

A **mutagen** is a physical or chemical agent that can change the DNA of an organism and thus increases the frequency of mutations.

Ionizing radiation - X-rays, gamma rays

Ultraviolet waves - sunlight (mild)

Alkaloid plants - tobacco, coca plant, poppy plant

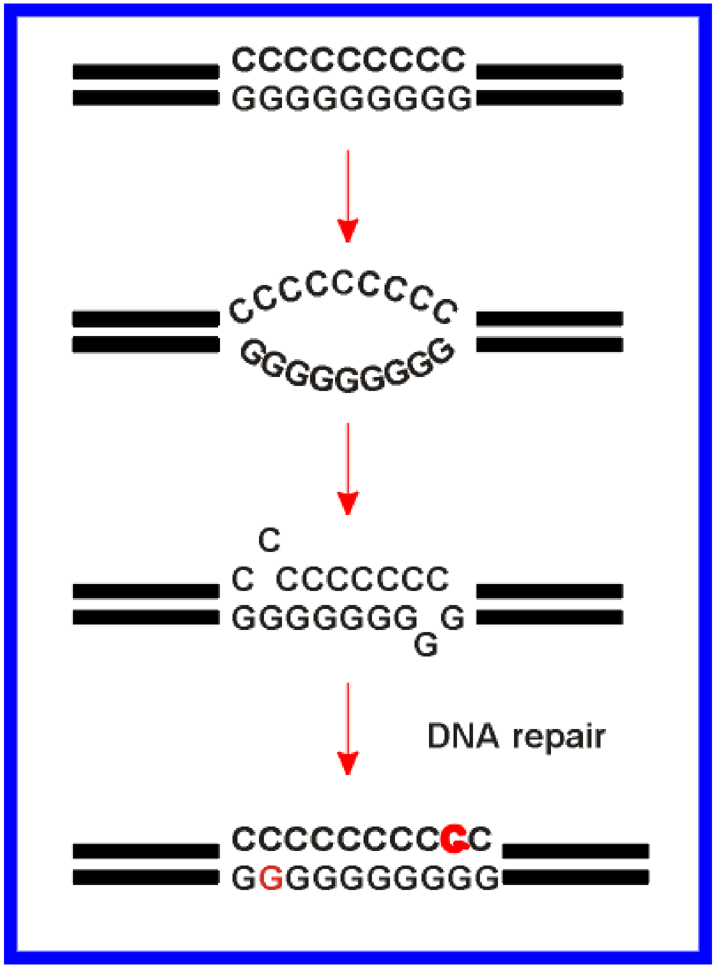
Sodium azide - a component in many car airbags

Benzene - solvent used in plastics, synthetic rubber

Spontaneous Mutation

Not all mutations are caused by mutagens.

Spontaneous mutations occur due to errors involving DNA replication, repair and recombination.



A nucleotide slipping out of place during replication, causing a mutation in the DNA once it has been repaired.

29 The result of a deletion mutation in DNA is that it potentially changes the:

- A structure of the protein
- B chromatin
- C DNA backbone
- D number of chromosomes

answer

30 Changing one nucleotide in a DNA sequence can change __________ in a protein

- A the primary structure
- B the secondary structure
- C the tertiary structure
- D all of the above

answer

31 Using AAA TTT GGG AAA as an example, which of the following would be an example of a frame shift mutation?

- A AAA TTT CCC GGG
- B AAA TTT GGG AAA
- C AA ATT TCC CGG G
- D TTT CCC GGG

answer

32DNA which does not code for proteins is referred to as:

- A Silent DNA
- B Excess DNA
- C Junk DNA
- D Redundant DNA

answer

Genetics of Viruses

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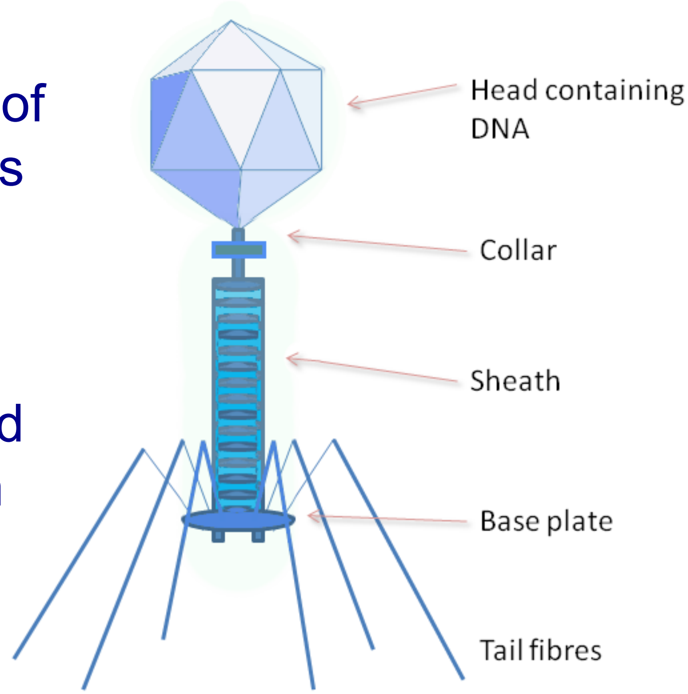
Viruses

A virus is a small infectious agent that can replicate only inside the living cells of an organism. Viruses can infect all types of organisms, though they are not organisms themselves.

Viruses are particles that are considered non-living because they cannot perform all the functions of living things. However, they share the same genetic code and use the mechanism of host cells to reproduce.

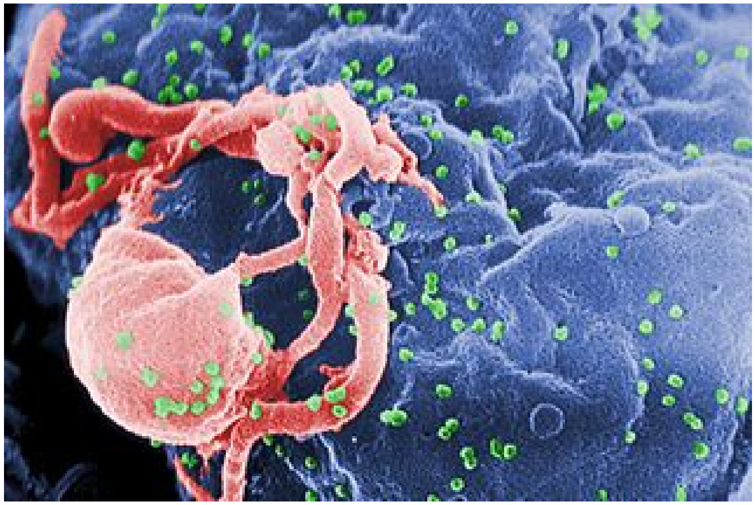


A bacterial phage virus

Viruses

In biology viruses are important because their genetic and reproductive strategies use the same molecular components but their use is unique. Scientists have learned a lot about genetics by understanding their strategies.

In addition, their infectious nature makes them a threat that requires humans to understand how they work to create defensive technology.



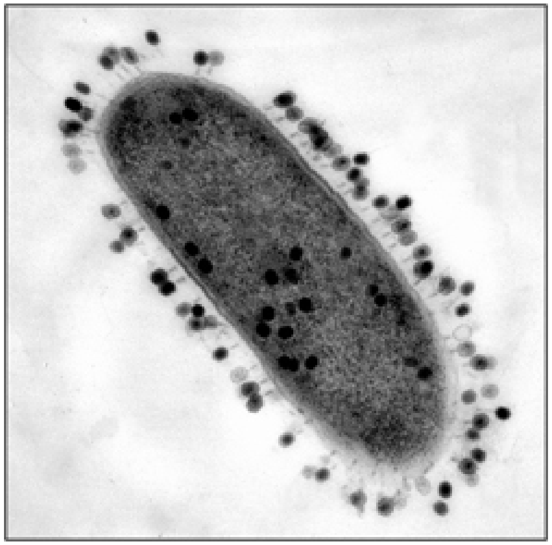
Human immunodeficiency virus (HIV) attacking a human lymphocyte cell

Viruses

The general mode of operation for all viruses is to

- Infect a host cell with its genetic information
- Hijack the molecular machinery of the host cell to manufacture the parts needed to build more viruses
- Package the parts together to form new viruses for release from the host cell

This *E. coli* cell has been attacked by bacteriophage viruses and they have injected their genetic material into the bacterial cell



Viral
Reproductive Cycles

Viruses are **obligate intracellular parasites,** which means they can reproduce only within a host cell.

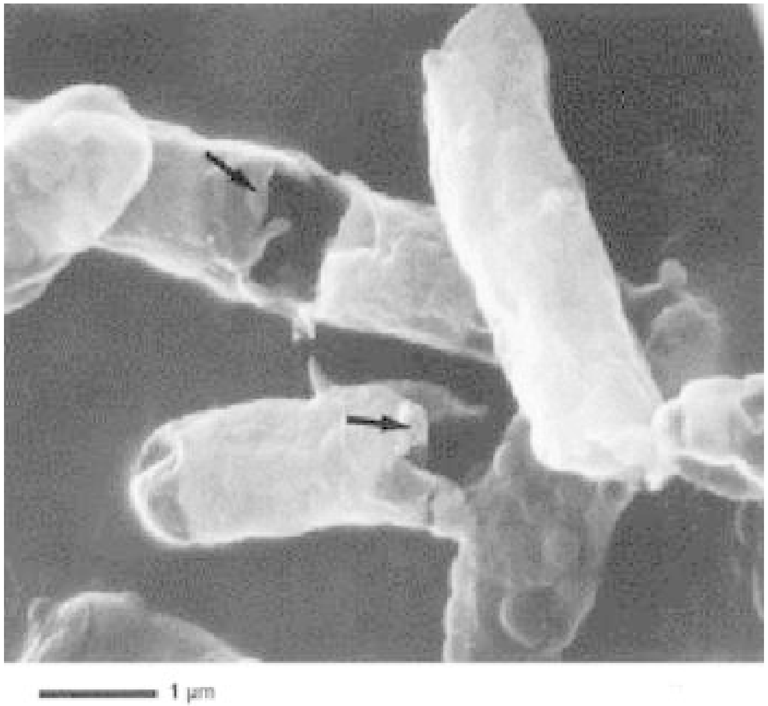
Each virus has a **host range;** it is limited by type of host cells that it can infect.

Viruses use enzymes, ribosomes, and other parts of the host cell to synthesize new viruses.

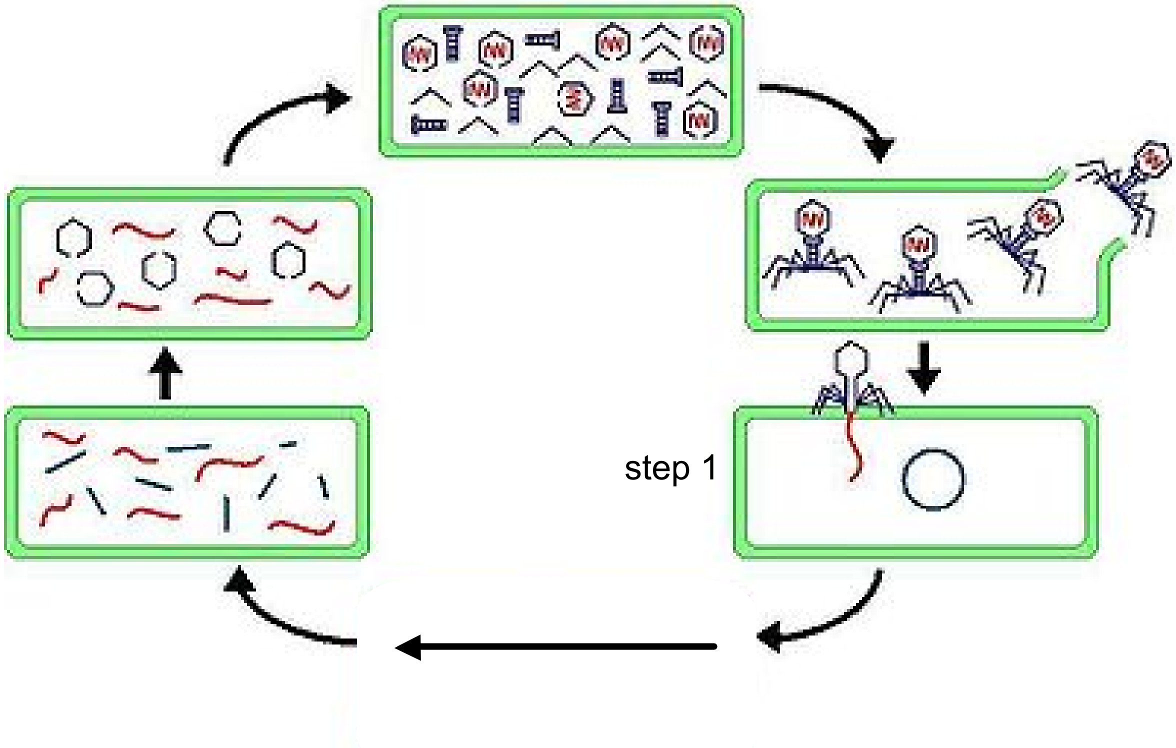
Lytic Cycle

The **lytic cycle** is a viral reproduction that causes the death of the host cell. The lytic cycle produces new phages and digests the host's cell wall, thereby releasing the new viruses.

These cells have been broken open by the lytic cycle of a virus



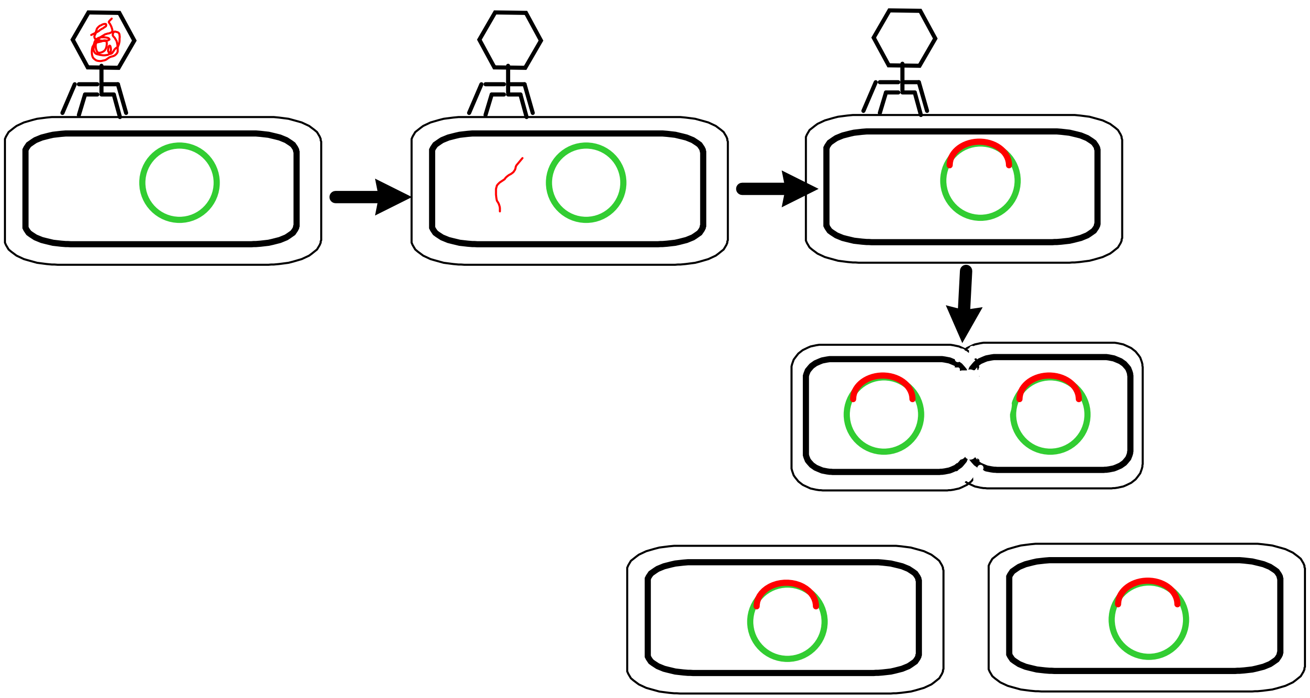
The Lytic Cycle of a Phage



Lysogenic Cycle

Unlike the lytic cycle which is detrimental to the host cell, the **lysogenic cycle** does not cause the cell to die.

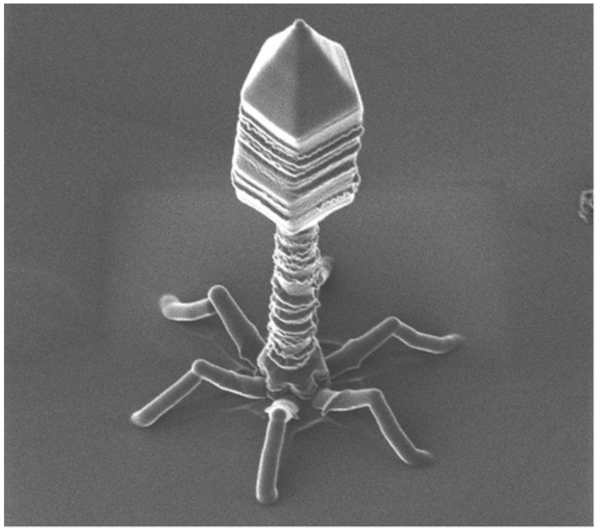
In the lysogenic cycle, the viruses DNA is incorporated into the host's DNA. Then the bacteria cell continues to replicate through binary fission, copying the viruses DNA and its own together.



DNA Viruses

Some viruses use DNA as their genetic material. It does not contain DNA polymerase, so in order for it to reproduce it must inject its DNA into a cell so that it can be copied by the host cell's polymerase.

The bacteriophage example we have seen is a DNA virus.

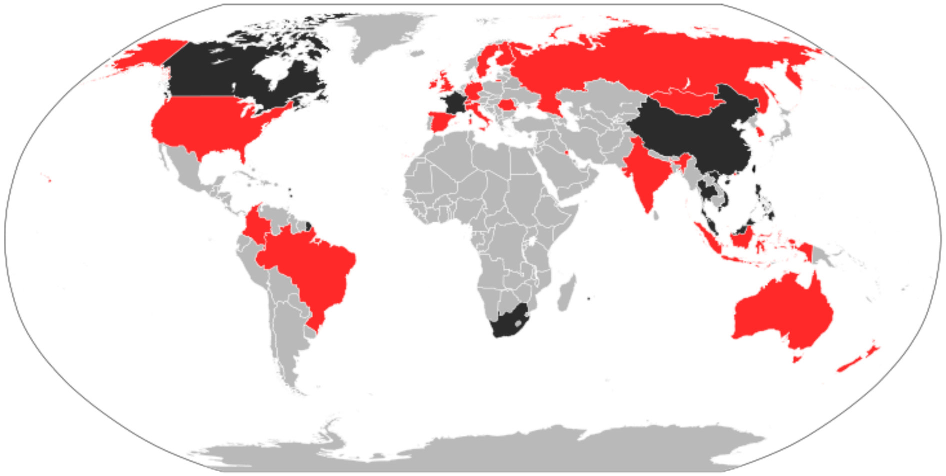


RNA Viruses

An RNA virus is a virus that has RNA as its genetic material. This nucleic acid is usually single-stranded RNA (ssRNA), but may be double-stranded RNA (dsRNA). Some human diseases caused by RNA viruses include SARS, influenza, hepatitis C, West Nile fever, polio and measles.

RNA Viruses

Severe acute respiratory syndrome (SARS) is a viral respiratory disease in humans. An outbreak of SARS in Hong Kong nearly became a pandemic, with 8,273 cases and 775 deaths worldwide. Within weeks, SARS spread from Hong Kong to infect individuals in 37 countries in early 2003.



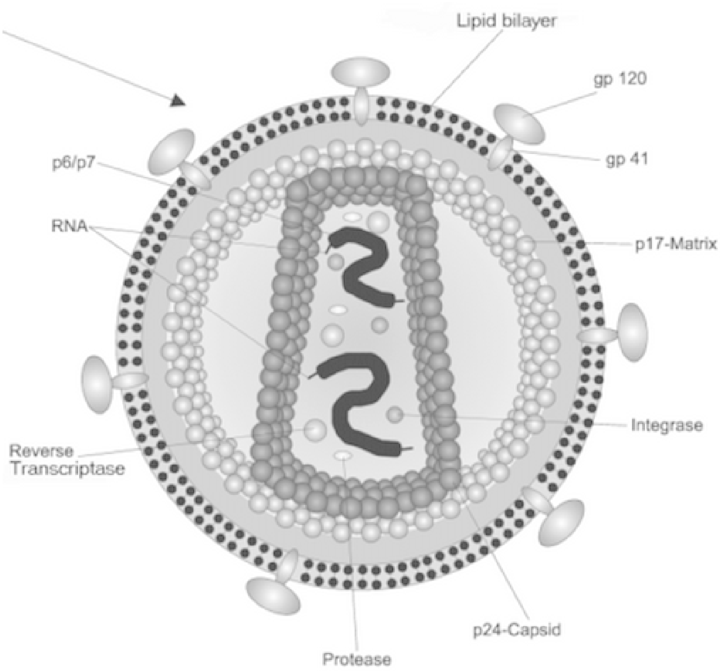
They are enveloped RNA viruses that are pathogens of mammals and birds. This group of viruses cause respiratory tract infections in a variety of animals, including humans

Retro Viruses

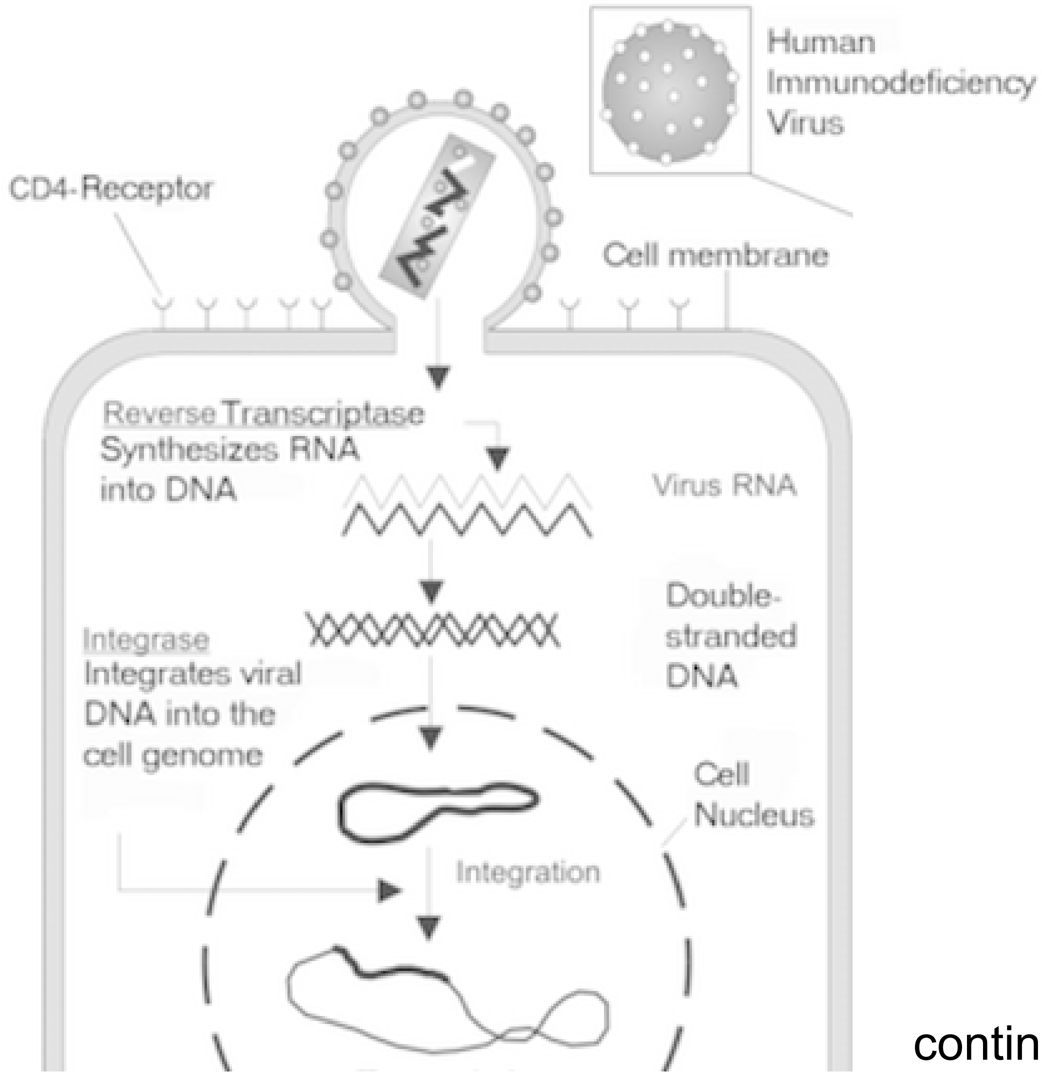
A retrovirus is an RNA virus that replicates in a host cell. First it uses its own **reverse transcriptase** enzyme to produce DNA from its RNA genome, reverse of the usual pattern, thus retro. This new DNA is then incorporated into the host's genome by an**integrase** enzyme. The cell then treats the viral DNA as part of its own instructions, which it follows blindly, making the proteins required to assemble new copies of the virus.

Retro Viruses

These viruses are some of the most complex and believed to be the most advanced from an evolution perspective. For a virus, their entry system into cells is highly complex. They have systems to bypass the usual defenses of their host cell. HIV is a retro virus.



Retro Viruses



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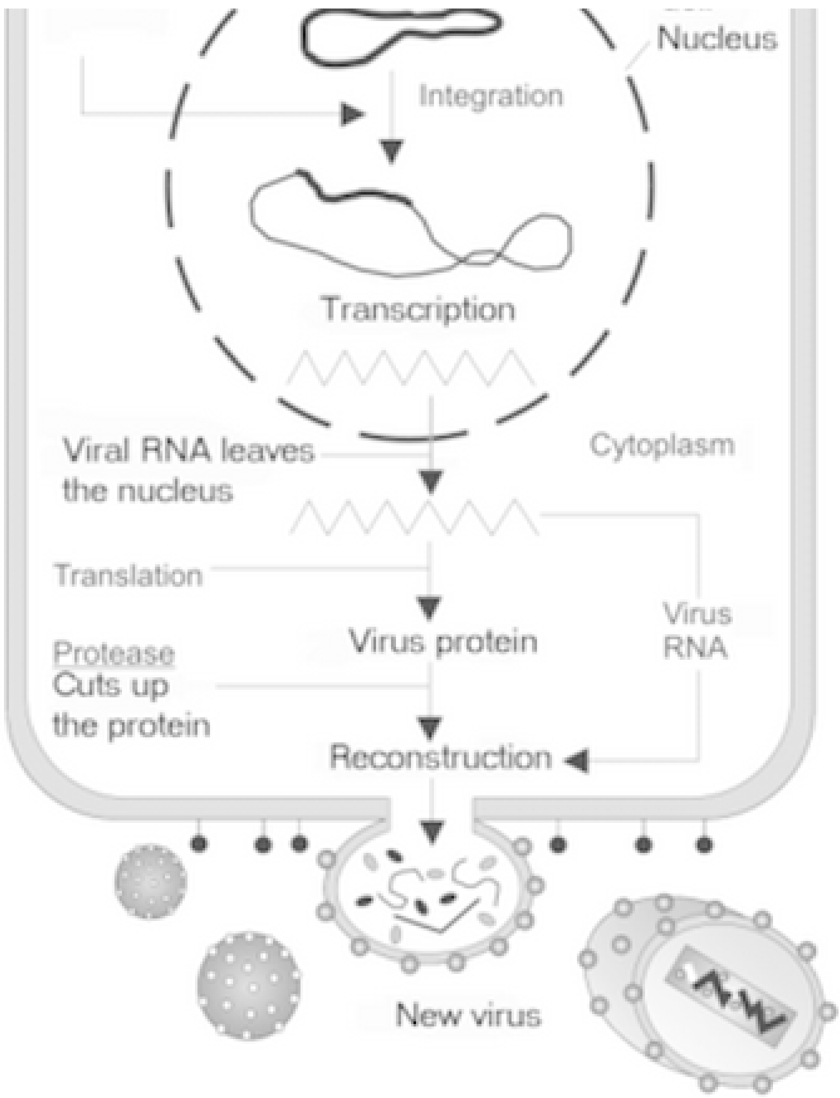
Retro Viruses



HIV

What makes HIV particularly dangerous is that it attacks the human immune system and fools it into treating it is part of the system. No immune attack is offered by the infected cells.

The complex replication system is flawed and many errors are made as the virus replicates its genome. Why would this make treating with medicine difficult?

33Viruses that infect bacteria are called _______.

- A bacterioviruses
- B bacteriophages
- C capsomeres
- D proviruses

answer

34 When a virus infects an E. coli cell, what part of the virus enters the bacterial cytoplasm?

- A the entire virus
- B the nucleic acid
- C the protein capsid and enclosed nucleic acid
- D the tail fibers

answer