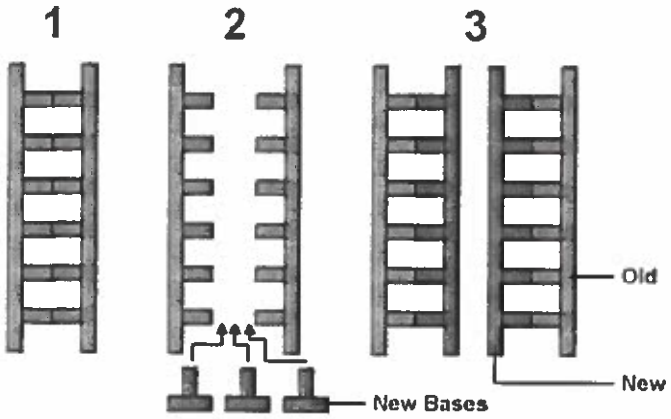
Genetics

Key

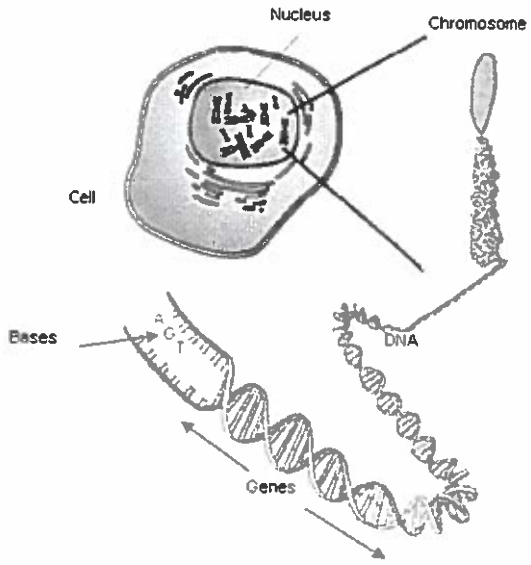
Main Concept #1: Describe how the process of DNA replication results in the transmission and/or conservation of genetic information.

- DNA Replication – copying of DNA
  - Ensures that each resulting cell will have a complete set of DNA molecule
  - During DNA replication, the DNA molecule separates into two strands, then produces two new complementary strands following the rules of base pairing. Each strand of the double helix of DNA serves as a template against which the new strand is made → called semiconservative replication



Main Concept #2: Explain the functional relationships between DNA, genes, alleles, and chromosomes and their roles in inheritance.

- Two conclusions from Mendel's experiments with the pea plant
  1. biological inheritance is determined by factors that are passed from one generation to the next = **genes**
     - each gene controlled one trait with two contrasting characters
     - different forms of a gene = **alleles**
  2. principle of dominance – states that some alleles are dominant and others are recessive
     - organism with dominant allele for a particular form of a trait will always have that form
     - organism with recessive allele for a particular form of a trait will have that form only
- Segregation – separation of alleles
  - Done during formation of gametes (reproductive cells)



DNA is a molecule that carries genetic information which determines which proteins to make when

A **gene** is a specific segment of the DNA molecule that codes for a specific protein – EX: eye color

A **chromosome** is a whole bunch of genes linked together into organized structure (eukaryotes it is linear / prokaryotes it is circular)

An **allele** is a form of a gene – meaning it still is going to do what is asked (make eye color) but it has different information – blue eyes, brown eyes, hazel eyes...)

Main Concept #3: Describe and/or predict observed patterns of inheritance (ie. dominant, recessive, co-dominance, incomplete dominance, sex-linked, polygenic, and multiple alleles).

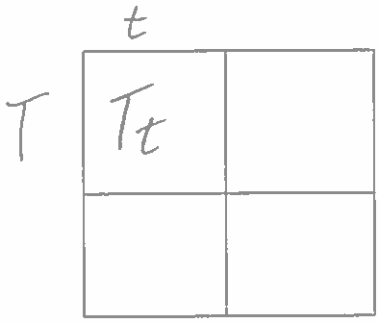
- Probability – likelihood that a particular event will occur
  - Probability of two events happening, you multiply the individual probabilities
    - Past outcomes do not affect future ones
  - The principles of probability can be used to predict the outcomes of genetic crosses
- Punnett square – diagram that helps determine gene combinations that might result from a genetic cross
- Capital letters represent dominant alleles; lower case letters represent recessive alleles
- Homozygous – have two identical alleles – true-breeding
- Heterozygous – have two different alleles – hybrid – carrier
- Phenotype – physical feature
- Gentoype – genetic make-up

- for two genes, alleles segregate independently
  - independent assortment – genes segregate independently and do not influence each other's inheritance
    - the principle of independent assortment states that genes for different traits can segregate independently during the formation of gametes
- some alleles are neither dominant nor recessive, and many traits are controlled by multiple alleles or multiple genes
  - incomplete dominance – one allele is not completely dominant over another
    - heterozygous phenotype is somewhere between two homozygous phenotypes
  - codominance – both alleles contribute to the phenotype of the organism
    - heterozygous phenotypes have some of both homozygous phenotypes
  - multiple alleles – genes that have more than 2 possible alleles
  - polygenic traits – traits that result from the interaction of many genes
    - these traits are also greatly influenced by the environment

Monohybrid Cross

1. Two fish meet at the coral reef, fall in love, and get married that same night. They decide to make babies right away. The mom fish has a big fluffy tail (TT) while the dad has a very boring flat tail (tt). The dad is worried that he will pass his ugly tail down to his kids. What is the chance that the first child will have a flat tail?

T = fluffy tail t = flat tail



Genotypic Ratio: All Tt Phenotypic Ratio: All Fluffy

Incomplete Dominance:

1. In Japanese four-o'clocks, the gene for red flower color (R) is incompletely dominant over the white flower color(r). Predict the genotypic and phenotypic ratios of a red plant crossed with a white plant.

All RW All Pink

Codominance

1. The palomino horse is a hybrid (mix) showing a golden coat with a lighter mane and tail. A pair of codominant alleles, D1 and D2 is known to be involved in this trait. Horses with the D1D1 genotype are chestnut colored, horses with the D1D2 genotype are palomino, and horses with the D2D2 genotype are white in color.

   A. Two palomino horses are mated by artificial insemination. What types of offspring could be produced?

1 Chestnut
2 Palomino
1 White

Sex-Linked Traits

1. White eyed fruit flies are the result of a sex-linked recessive gene. Show the results from a cross between a red-eyed (R) male and white-eyed (r) female fruit fly.

$X^rX^r \cdot X^RY$

♀ All X^RX^r red
♂ All X^rY White

Main Concept #4: Explain how genetic engineering has impacted the fields of medicine, forensics, and agriculture (e.g., selective breeding, gene splicing, cloning, genetically modified organisms, gene therapy).

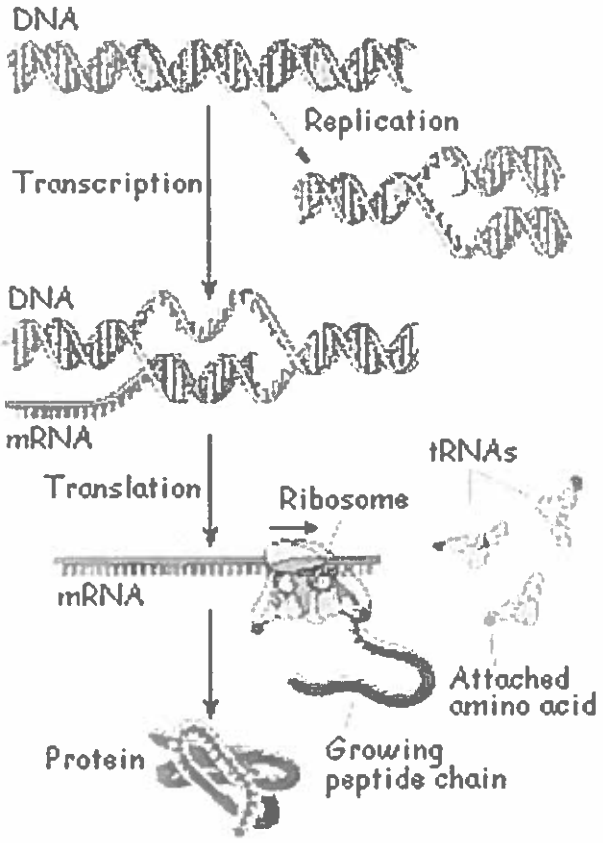
- selective breeding – allowing only those animals with desired characteristics to produce the next generation
  - humans use selective breeding to pass desired traits on to the next generation of organisms
- genetic engineering – making changes in the DNA code of living organisms
- Cutting / Splicing DNA
  - Restriction enzymes – cuts DNA at a specific sequence of nucleotides
  - cutting and pasting
    - recombinant DNA – taking DNA and "pasting" it to another organism's DNA
- transgenic organisms /genetically modified organisms– organisms that contain genes from other organisms
  - using the basic techniques of genetic engineering, a gene from one organism can be inserted into cells from another organism. These transformed cells can then be used to grow new organisms
- clone – member of a population or genetically identical cells produced from a single cell
- gene therapy is the process of attempting to cure genetic disorders by placing copies of healthy genes into cells that lack them

Protein Synthesis

Vocabulary

Transcription
Translation
Translocation
Chromosomal mutation
Deoxyribonucleic acid
Frame-shift mutation
Gene expression
Mutation
Point mutation

Concepts to Know



The Central Dogma: How our DNA code makes Phenotype
DNA → RNA → Protein

How are we so different? Why are we not identical to a plant? Or a bacterium? Or each other?

The DNA code is the same in all organisms but the sequence of the letters is different. All life uses A,C,T,G in double-stranded base pairs. This is the same concept that War and Peace is not identical to your IPod Warranty, but they're still written in English. DNA is just a language. A very, very, very important language.

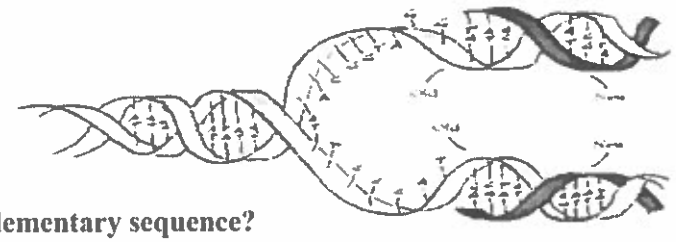
This is why scientists can manipulate life in the laboratory so easily. This is also the key to understanding a single common ancestor.

DNA is the code of life – ACTGs are the alphabet of life, just like 0s and 1s are the binary code of computers. The ACTGs of DNA can be read by enzymes to create a triplet codon that is interpreted into 20 amino acids to make very diverse proteins. **DNA is letters; codons are words; proteins are messages that make sense.**

1st Idea: Life Contains DNA: it is a SELF-REPLICATING molecule.

DNA replicates itself (via DNA polymerase and other enzymes) in ***a semi-conservative*** manner. This means that at the end of replication, each of the daughter molecules has one old strand, from the parent strand of DNA, and one strand that is newly synthesized. (see pic).

Adenine pairs with Thymine (A = T)
Guanine pairs with Cytosine (G ≡ C)
The bonds between the base pairs are **hydrogen bonds**



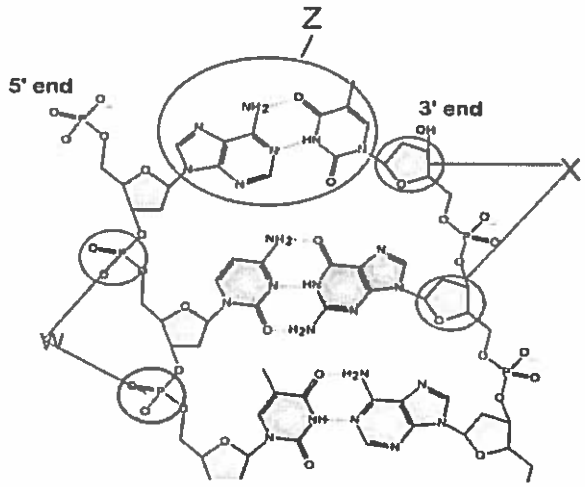
If given the template strand of DNA below, what is the complementary sequence?

5' A T G T A T G C C A A T G C A 3'

3' T A C A T A C G G T T A C G T 5'

FYI: ***How'd they figure this stuff out?*** *With radioactive isotopes of nitrogen, sulfur and phosphorus.*

DNA proofreading: the enzymes involved make sure this process makes an exact message (only 1 in 10 billion base pairs would be incorrect; better than computer coding mechanisms)



DNA STRUCTURE

Nucleic Acid (polymer) is made of nucleotides (monomer)

A nucleotide is made of: a sugar X, deoxy

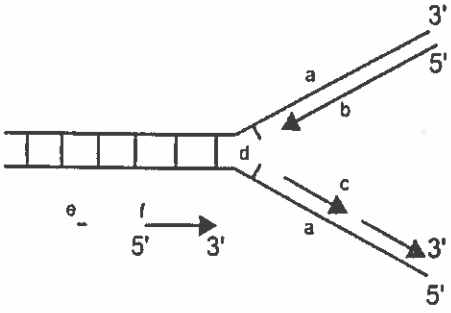
a phosphate group W,

and a nitrogenous base Z. A, T, C, G

IDENTIFY EACH AS W, X, OR Z IN THE DIAGRAM

I. DNA REPLICATION

DNA polymerase is an enzyme *(ends in –ase)*. **All enzymes have a specific active site.** The DNA in this example is the substrate and only can fit into the enzyme (DNA polymerase) a certain way. This is why DNA replication has a **leading** and a **lagging** strand when made. The enzyme can only fit onto DNA via the 3' hydroxyl side, not the 5' phosphate side.

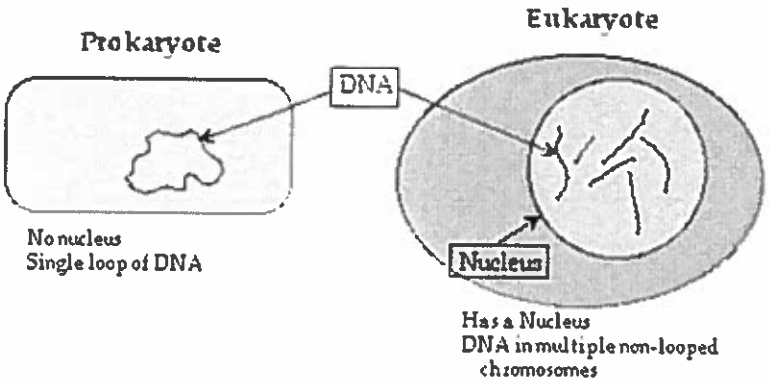


What letter in this diagram represents the *continuous* leading strand? b

What letter represents the Okazaki *fragments* of the lagging strand? c

II. P ROKARYOTES VS EUKARYOTES

Prokaryotes ("before nucleus") evolved before eukaryotes ("true nucleus") and have slight differences in their DNA structure.



➔ **What can you deduce from the picture?**

No Lagging Strand in Prok.

2nd Idea: DNA is the source message but RNA is the working copy

MAJOR DIFFERENCES BETWEEN DNA AND RNA

DNA	RNA
deoxiribose sugar	ribose sugar
thymine	uracil
double helix	single strand (mRNA) or unit (tRNA)
permanent	temporary
in nucleus (some in mitochondria)	leaves nucleus, works in cytoplasm
one kind	many kinds (at least 3)

The DNA is like the encyclopedia you can never check out of the library. However, you are allowed to make copies of the information. That's what RNA is - a copied message of the important pages. Making copies ensures that you don't 'ruin' the original by taking it out of the nucleus (*this only applies to eukaryotes*), you can make copies in bulk, AND you only have to make copies of what you need. You wouldn't copy all 6000 pages of an encyclopedia would you? No! Only the 4-5 pages you might need for a report. In eukaryotes, we only code for ~ 2% of our DNA!

RNA (ribonucleic acid) is the intermediate between DNA and protein. It has slight differences to DNA. See the chart.

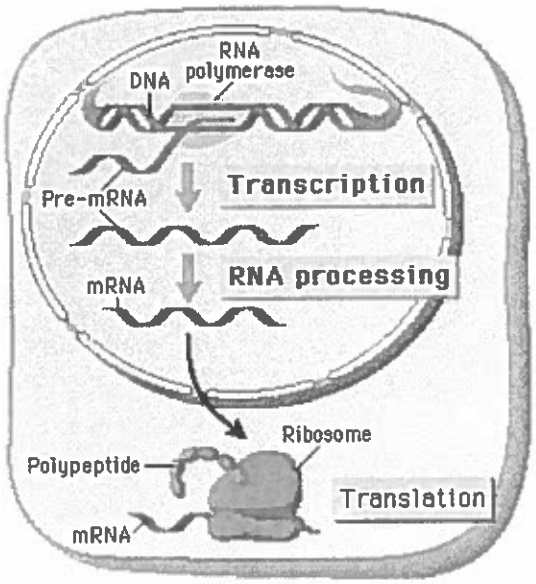
TRANSCRIPTION is the process of making RNA from DNA (via the enzyme RNA polymerase). This happens in the nucleus for eukaryotes, but would happen in cytoplasm for prokaryotes.

Watch a refresher video of the process on the protein synthesis page for www.udkeystone.wikispaces.com

Can you complete this message?

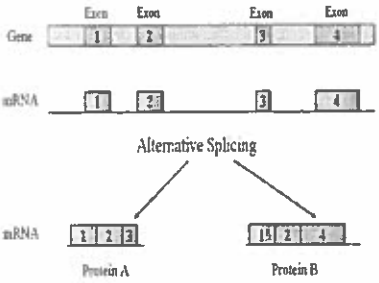
T A C C C C T T T G G C A T A G A

A U G G G G A A A C C G U A U C U



Important Points about TRANSCRIPTION: DNA → RNA → Protein

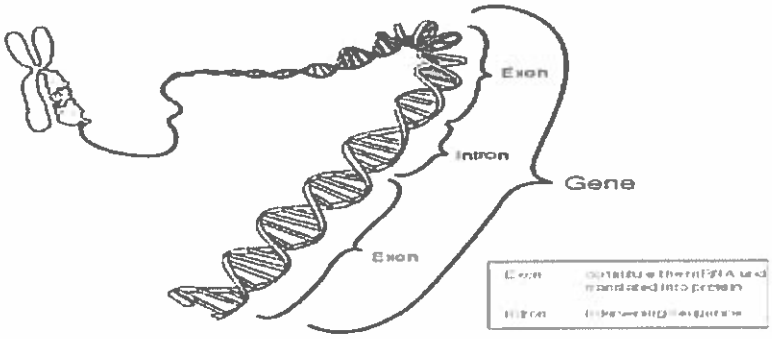
- **RNA Polymerase** scans the genome for the promoter region of DNA (the start signal)
- A single-stranded copy of RNA is made of the DNA gene, where **U is complementary to A instead of T.**
- Transcription and Translation occur simultaneously in the cytoplasm for prokaryotes, with no editing needed.
- **Transcription occurs in nucleus for eukaryotes.**
- Eukaryotic messageRNA has **EXONS** (expressed message) and **INTRONS** (in-between message)
- Introns get spliced (cut out) of the mRNA to make the mature transcript.



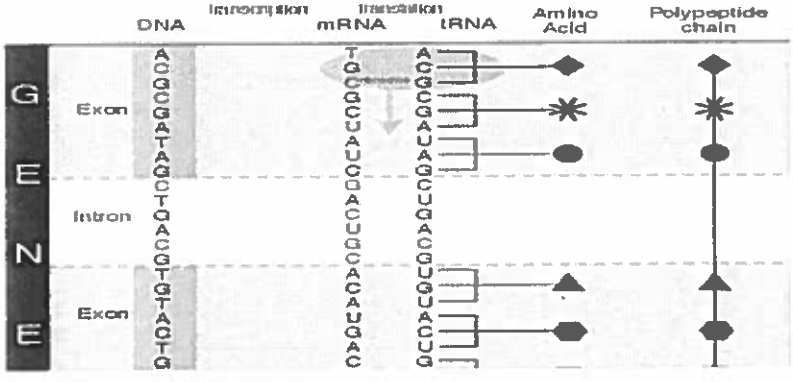
WHY INTRONS?

Alternative splicing allows to mix-n-match exons to make different proteins from the same sequence. This is a major source of eukaryotic evolution!

This is like you being able to make 20 different outfits in your closet from 4 pairs of pants and 5 shirts.



3rd Idea: Translation is matching an amino acid to the messageRNA in order to make the protein code



Important points about TRANSLATION

- The mRNA leaves the nucleus → cytoplasm *(in eukaryotes)*
- Message is read at the **ribosome**
- mRNA is read 3 letters at a time
- AUG is the start signal
- 1 Codon (3 letter message) is translated into 1 amino acid
- transferRNA molecule has one end (anticodon) that matches the mRNA . Each anticodon specifies an amino acid.
- There are 20 amino acids
- The amino acids are bonded together as peptide chains…which fold into proteins

If a mature mRNA transcript has 300 nucleotides, how many amino acids would that code for?

100aa

TRANSLATION :

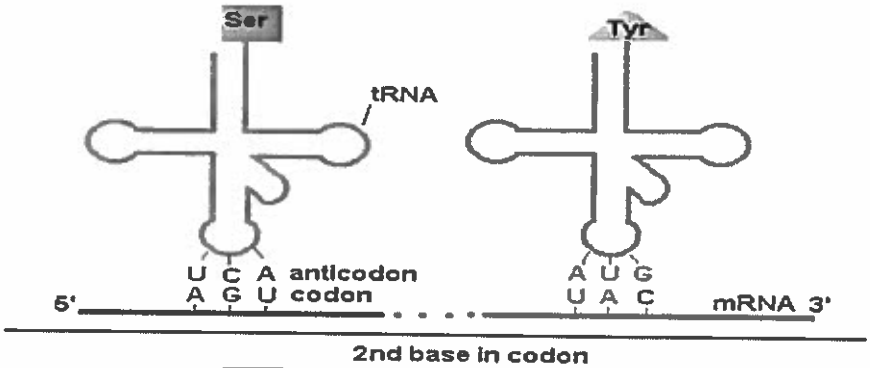
DNA → **RNA → Protein**

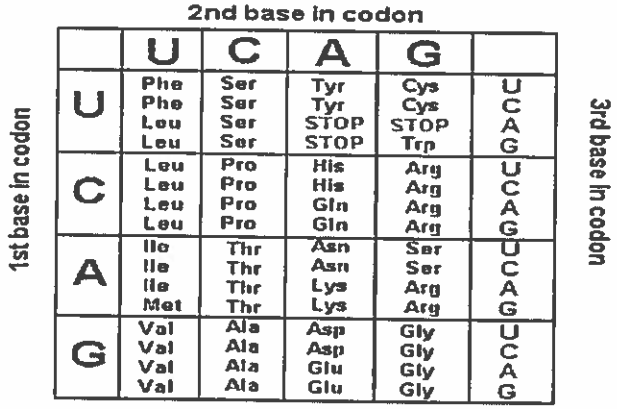
Ex: the message AUGGGGCAAUAA codes for Met-Gly-Gln-*
(the * tells the ribosome to stop)

What does this message code for?

AUG CUU CCA GAG UGA

Meth Leu Pro Glu ____





The Genetic Code

- After a polypeptide chain is made from amino acids (at a ribosome), it might be used right away in the cytoplasm, or it might be sent to the Golgi apparatus to have more folding or carbohydrates added.
- Proteins made on free ribosomes will work in the cytoplasm
- Proteins made on the rough ER will go the cell membrane or be excreted

4TH Idea: Mutations in the DNA or RNA sequence produce the wrong amino acid sequence.

*****THE ULTIMATE SOURCE OF EVOLUTION IS MUTATION*****

MUTATION : A change in DNA sequence

- **Point Mutations**: Change one or two base pairs
  - → **Insertion, Deletion, Substitution**

Only 2 of these are "**frameshift mutations**" - that is, they change the codon reading frame.

Other mutation vocabulary

- ➔ **Silent Mutation** = the mutation goes unnoticed – it does not change the amino acid sequence or is not in a coding region (the mutation is in an intron, or the 98% of the genome that doesn't code for protein, or in the 3rd base of a codon)
- ➔ **Missense** – an insertion, deletion, or substitution that would make the message different
- ➔ **Nonsense** – really bad; a stop codon is created and the message stops prematurely

Example: Remember that DNA and RNA are just a language. To emphasize the point of mutation, I am using English (an alphabet with 26 letters, not 4!) Imagine you have the following message:

THE CAT ATE THE RAT

Using the above bolded mutations, label the type of mutation each must be:

Sub THE HAT ATE THE RAT

Ins TTH EHA TAT ETH ERA T

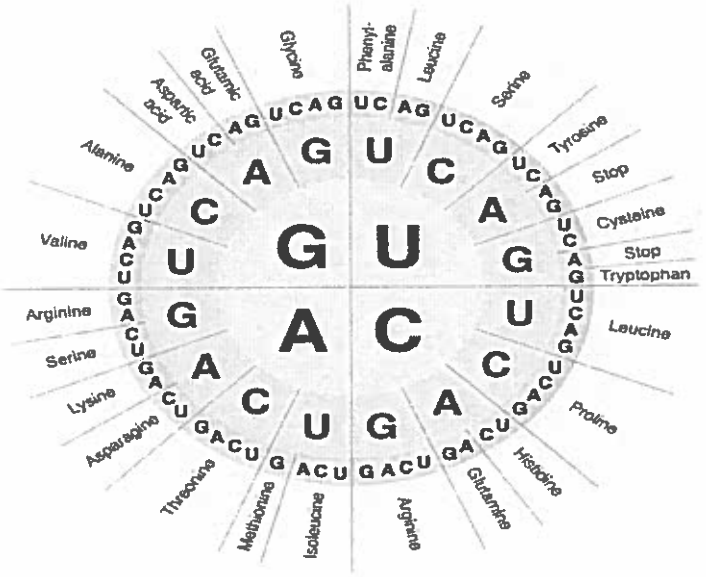
Del THE ATA TET HER AT

The "Central Dogma of Biology" is summarized as:

DNA → RNA → Protein

Fill in the chart:

DNA Triplet	mRNA Codon	tRNA Anticodon	Amino Acid
TAC	AUG	UAC	met
GGA	CCU	GGA	Pro
TTC	AAG	UUC	Lys
ATC	UAG	AUC	STOP



➔ ORDER THE FOLLOWING

3 Intron sequences are spliced out and exons are joined together

7 amino acids form peptide bonds as tRNA molecules match the mRNA

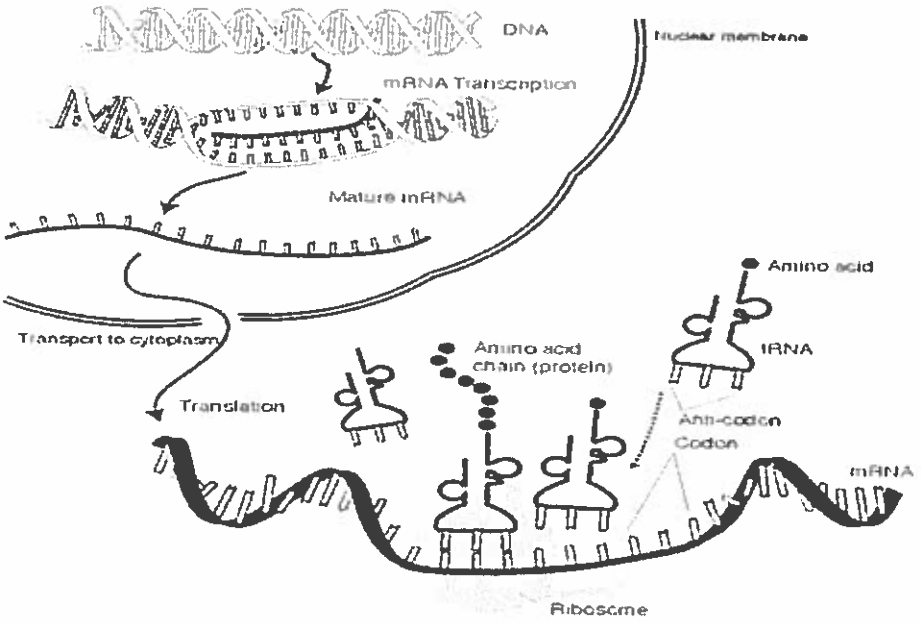
2 RNA polymerase reads the DNA and builds complimentary sequence

5 The mRNA attaches to the ribosome

4 The ends of the mature transcript are protected before it leaves the nucleus

1 RNA polymerase finds the promoter sequence on DNA

6 transfer RNA arrives at the ribosome and the anticodon complements to the mRNA codon



Practice Questions:

1. Which process helps to preserve the genetic information stored in DNA during DNA replication?
   A. the replacement of nitrogen base thymine with uracil
   B. enzymes quickly linking nitrogen bases with hydrogen bonds
   C. the synthesis of unique sugar and phosphate molecules for each nucleotide
   (D) nucleotides lining up along the template strand according to base pairing rule

2. In a flowering plant species, red flower color is dominant over white flower color. What is the genotype of any red-flowering plant resulting from this species?
   A. red and white alleles present on one chromosome
   B. red and white alleles present on two chromosomes
   C. a red allele present on both homologous chromosomes
   (D) a red allele present on at least one of two homologous chromosomes

3. The endoplasmic reticulum is a network of membranes within the cell, and it is often classified as rough or smooth, depending on whether there are ribosomes on its surface. Which statement **best** describes the role of rough endoplasmic reticulum in the cell?
   A. It stores all proteins for later use.
   B. It provides an attachment site for larger organelles.
   (C) It aids in the production of membrane and secretory proteins.
   D. It stores amino acids required for the production of all proteins.

Use the table below to answer the question.

Blood Types

Genotypes	Phenotypes
ii	O
I^AI^A, I^Ai	A
I^BI^B, I^Bi	B
I^AI^B	AB

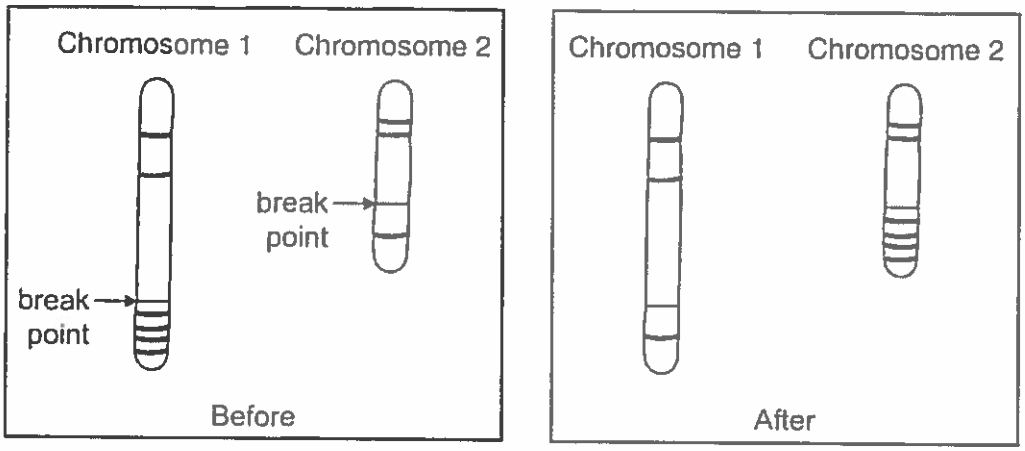
4. Blood type is inherited through multiple alleles, including I^A, I^B, and i. A child has type A blood. If the father has type AB blood, what are all the possible phenotypes of the mother?
   A. phenotypes O or A
   B. phenotypes A or AB
   C. phenotypes A, B, AB
   (D.) phenotypes O, A, B, AB

$I^AI^B \times ii$, I^A-, I^AI^B, I^Bi

I^A i or I^A

Use the diagram below to answer the question.

Chromosome Change



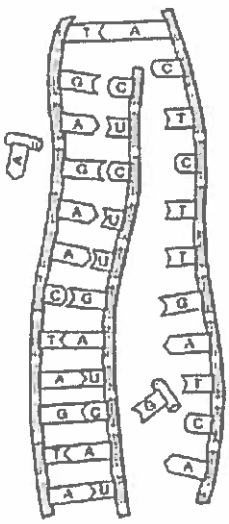
5. Which type of change in chromosome composition is illustrated in the diagram?
   A. deletion
   B. insertion
   C. inversion
   D. translocation

6. Which statement describes a cell process that is common to both eukaryotic and prokaryotic cells?
   A. Both cell types carry out transcription in the nucleus.
   B. Both cell types use ribosomes to carry out translation.
   C. Both cell types assemble amino acids to carry out transcription.
   D. Both cell types carry out translation in the endoplasmic reticulum.

7. A genetic mutation resulted in a change in the sequence of amino acids of a protein, but the function of the protein was not changed. Which statement **best** describes the genetic mutation?
   A. It was a silent mutation that caused a change in the DNA of the organism.
   B. It was a silent mutation that caused a change in the phenotype of the organism.
   C. It was a nonsense mutation that caused a change in the DNA of the organism.
   D. It was a nonsense mutation that caused a change in the phenotype of the organism.

8. Genetic engineering has led to genetically modified plants that resist insect pests and bacterial and fungal infections. Which outcome would **most likely** be a reason why some scientists recommend caution in planting genetically modified plants?
   A. unplanned ecosystem interactions
   B. reduced pesticide and herbicide use
   C. improved agricultural yield and profit
   D. increased genetic variation and diversity

9. Which of the following is primarily responsible for the coding of the amino acids used in the synthesis of cellular proteins?
   A. DNA
   B. transfer RNA
   C. ribosomes
   D. Golgi apparatus



Key
T = thymine
G = guanine
C = cytosine
A = adenine
U = uracil

10. Which statement describes the diagram above?
    a. DNA transcription is producing ribosomal RNA.
    b. DNA translation is producing messenger RNA.
    (c.) DNA transcription is producing messenger RNA.
    d. DNA translation is producing ribosomal RNA.

Open-ended Question:

11. A cattle farmer genetically crosses a cow (female) with a white coat with a bull (male) with a red coat. The resulting calf (offspring) is roan, which means there are red and white hairs intermixed in the coat of the calf. The genes for coat color in cattle are co-dominant.

Part A: Although a farm has cattle in all three colors, the farmer prefers roan cattle over white or red cattle. Use the Punnett square to show a cross that would produce only roan offspring.

	W	W
R		
R		

Part B: Explain how a roan calf results from one white- and one red-coated parent. In your explanation, use letters to represent genes. Be sure to indicate what colors the letters represent.

The R Allele Produces Red Hair } Both of These Show UP
The W " " White "

Part C: Predict the possible genotypes and phenotypes of the offspring produced from two roan cattle.

1 Red
2 Roan
1 White