

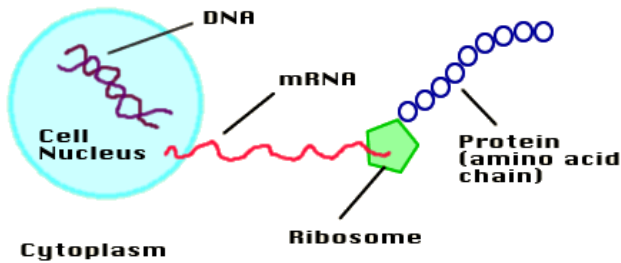
## 15 - Genes and Proteins [Google Slides](#)

### Gene Theory → Genes specify a proteins

⚓ How can a change in one gene lead to disease?  
→ Cystic Fibrosis

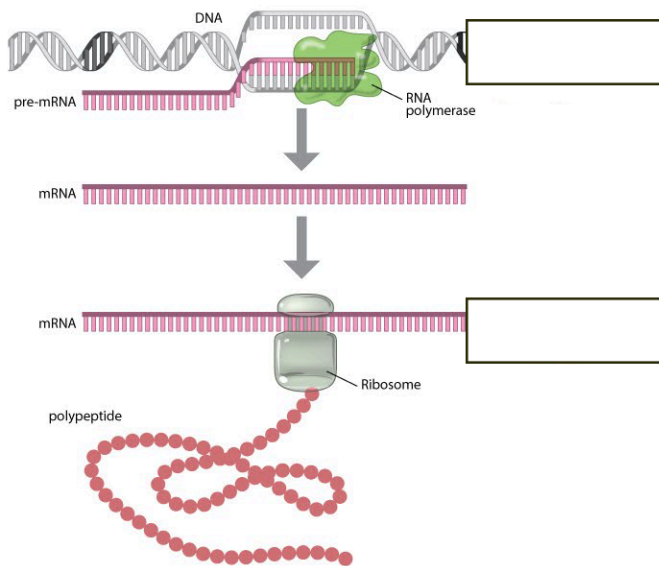
What do you remember about cystic fibrosis?

Three Important Points to Remember



- 1) Chromosomes are made of \_\_\_\_\_
- 2) Segments of DNA code for a \_\_\_\_\_
- 3) Protein codes for a \_\_\_\_\_

### The Central Dogma DNA → mRNA → Protein



### RNA = ribonucleic acid

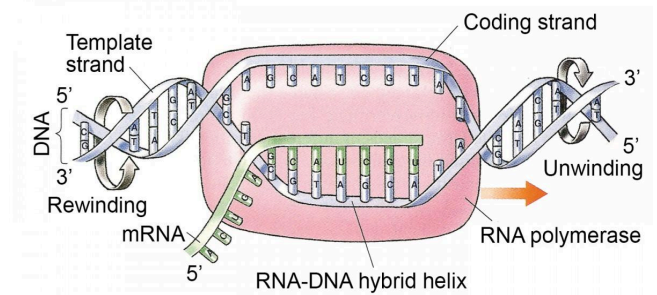
- ribose, uracil, \_\_\_\_\_

Three forms

\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

## Transcription - mRNA is made from DNA

- Follows base-pair rule but has uracil instead of \_\_\_\_\_
- RNA polymerase is used to build the strand of \_\_\_\_\_

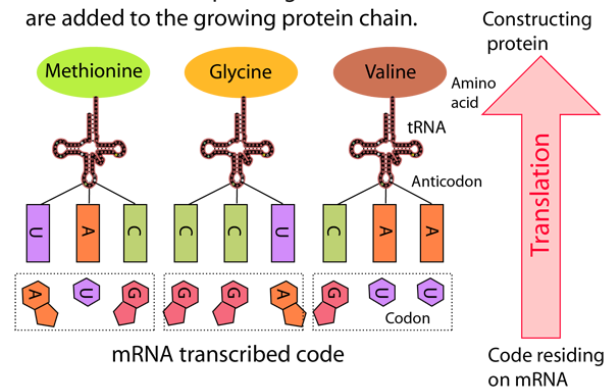


DNA : **A A T C C A T T G G A G**

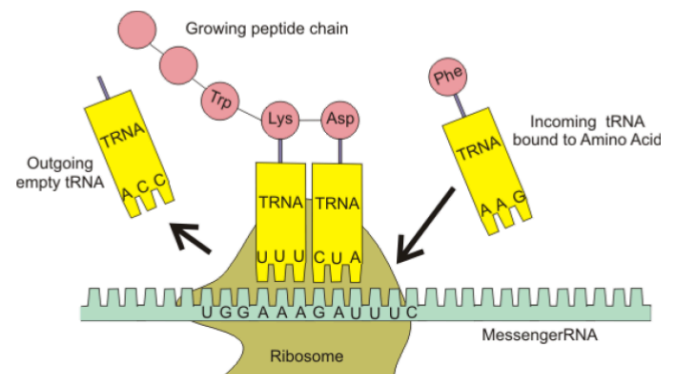
RNA:

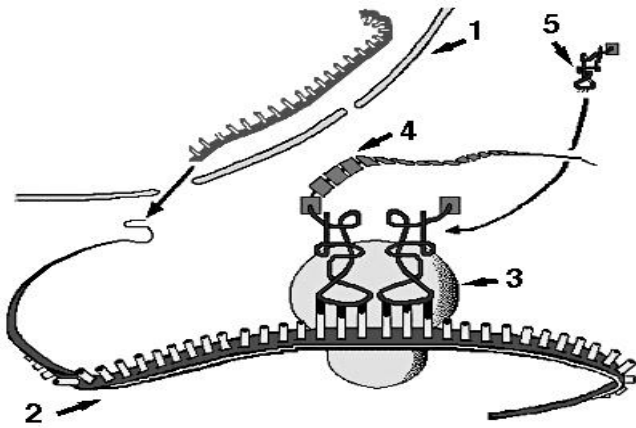
## Translation - protein is built from MRNA

Amino acids corresponding to the codons are added to the growing protein chain.



- occurs on the \_\_\_\_\_
- mRNA threaded through large and small subunit
- \_\_\_\_\_ delivers amino acids, forming a chain
- 3 bases on mRNA (codon) specify 1 amino acid
- translation begins with AUG \_\_\_\_\_
- ends at the stop codon
- proteins can contain any number of amino acids





**Codon charts** are used to determine the amino acid sequence

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

DNA: T A C A A T C G G G G G A T T

RNA:

A.A.

### Regions of DNA

\_\_\_\_\_ = coding regions of DNA

\_\_\_\_\_ = non-coding regions

Spliceosomes = coding areas, introns removed

**SNP** = single nucleotide polymorphism  
(curly vs straight coat in dogs)

**Mutations** - changes in DNA

Mutations in LMNA gene cause \_\_\_\_\_

## Gene Mutations

Point Mutations - single \_\_\_\_\_ change

Missense - changes the \_\_\_\_\_

## Hemoglobin - protein found in blood

Fragment of Beta Chain of Wild-type (normal) Adult Hemoglobin (HbA)					
DNA 3'to 5'	G A C	T G A	G G A	C T T	C T C
RNA					
Amino Acid					
Fragment of Beta Chain of Sickle Cell (mutant) Hemoglobin (HbS)					
DNA	G A C	T G A	G G A	C A T	C T C
RNA					
Amino Acid					

Nonsense mutation - results in a \_\_\_\_\_ codon  
- this shortens the resulting protein, Ex. DMD

**Gene Therapy** and Exon Skipping - skips over the stop codon, (repairs gene)

**Frameshift Mutation** - base is added or deleted  
- alters reading frame - changes protein

Diseases and nonfunctional proteins:

- Hemophilia
- Sickle Cell
- Huntington's
- Cystic Fibrosis

## Case Study: Cystic Fibrosis (Part 2)

Normal	Type I	Type II	Type III	Type IV	Type V
No synthesis	Block in processing	Block in channel regulation	Altered conductance	Reduced synthesis	
Q540X 12%	F508del 87%	G551D 5%	R117H 5%	A455E 5%	