

**“I forgot to make a back-up copy of my brain,
so everything I learned last semester was lost.”**

Don't let this happen to you!!

BELLRINGER

What happens, if in recording your grade, a teacher transposes the numbers for your 91 test grade?

Sometimes a small mistake can have a big effect!



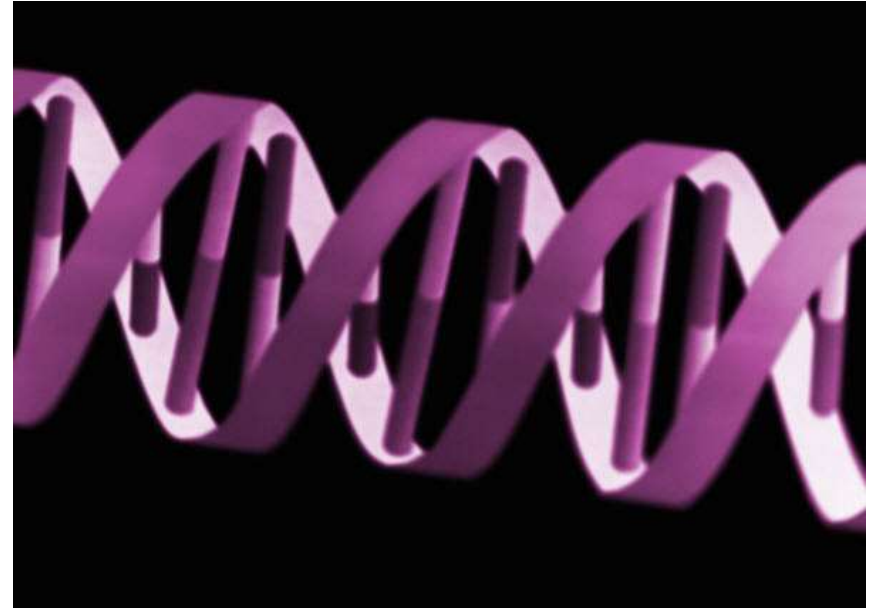
KEY CONCEPT - Mutations are changes in DNA that may or may not affect phenotype (physical characteristics).

EQ – How can mutations affect an organism's traits?

8.7 Mutations

► Significance of Mutations

- Most are **neutral**
 - Eye color
 - Birth marks
- Some are **harmful**
 - Sickle Cell Anemia
 - Down Syndrome
- Some are **beneficial**
 - Sickle Cell Anemia to Malaria
 - Immunity to HIV

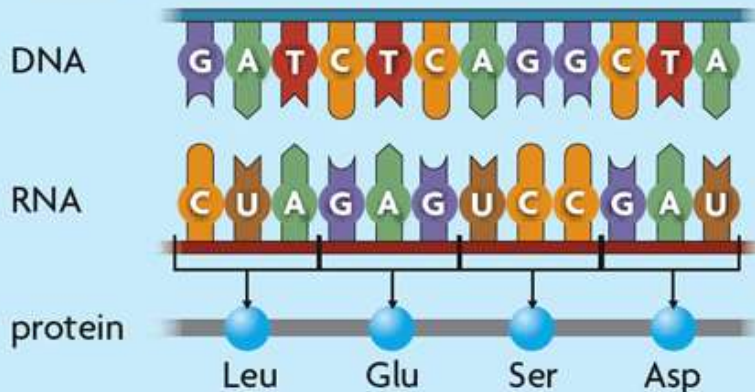


▶ Some mutations affect a single gene, while others affect an entire chromosome.

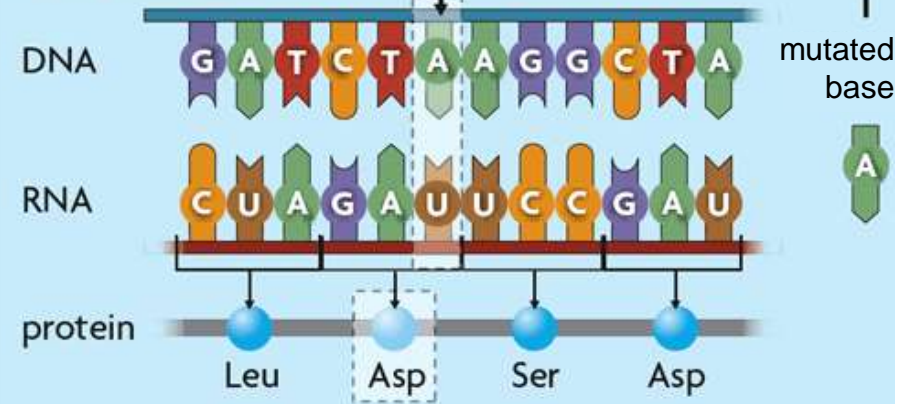
- A mutation is a change in an organism's DNA.
- Many kinds of mutations can occur, especially during **replication**.
- A type of point mutation substitutes one nucleotide for another (called a *substitution* – does not affect the reading frame.).

A **mutation** is a change in an organism's DNA.

Normal



Point mutation



8.7 Mutations

▶ POINT MUTATIONS:

- **Point Mutations** – changes in one or a few nucleotides that do not change the reading frame

- **Substitution**

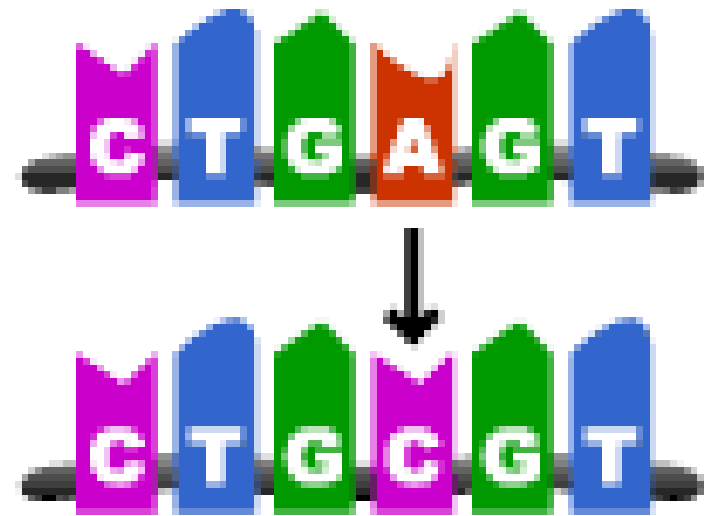
THE FAT CAT ATE THE RAT
THE FAT HAT ATE THE RAT

- **Insertion of 3 bases**

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- **Deletion of 3 bases**

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Point mutation

8.7 Mutations

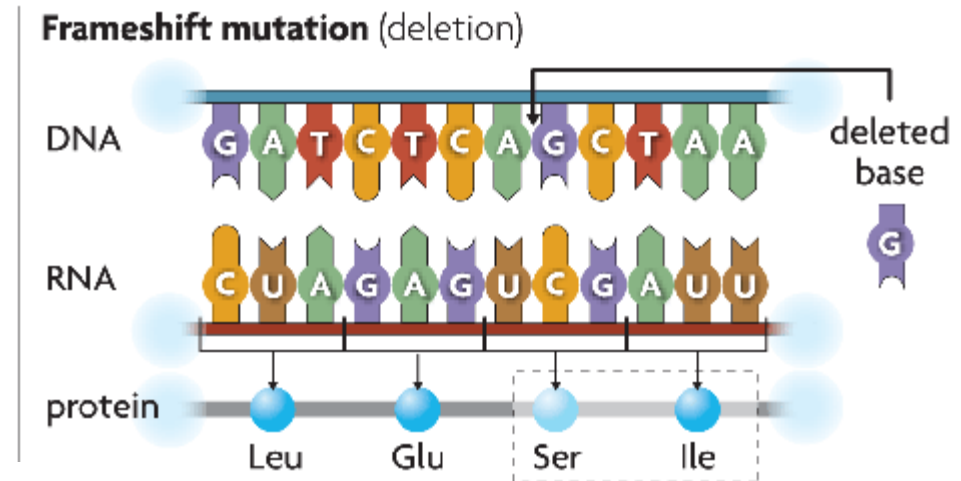
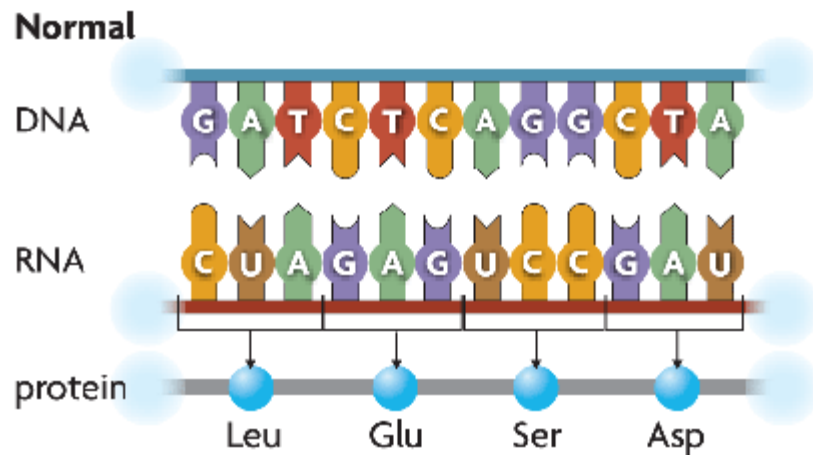
▶ Point Mutation

- **Sickle Cell Anemia** is the result of one nucleotide substitution
- Occurs in the **hemoglobin gene**



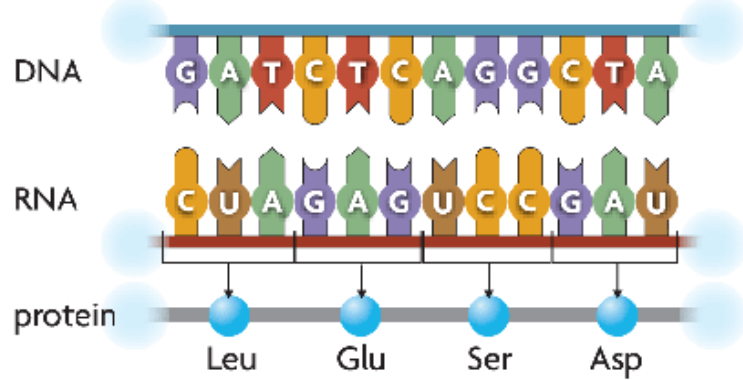
8.7 Mutations

- A **frameshift mutation** inserts or deletes a nucleotide(s) in the DNA sequence, ***changing the reading frame***.

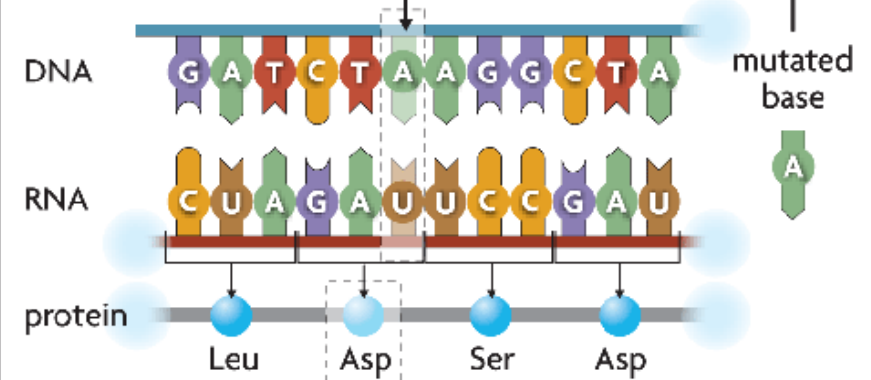


8.7 Mutations

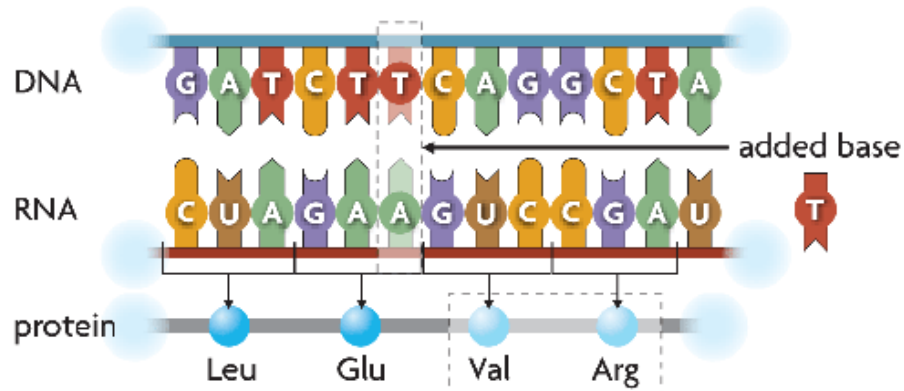
Normal



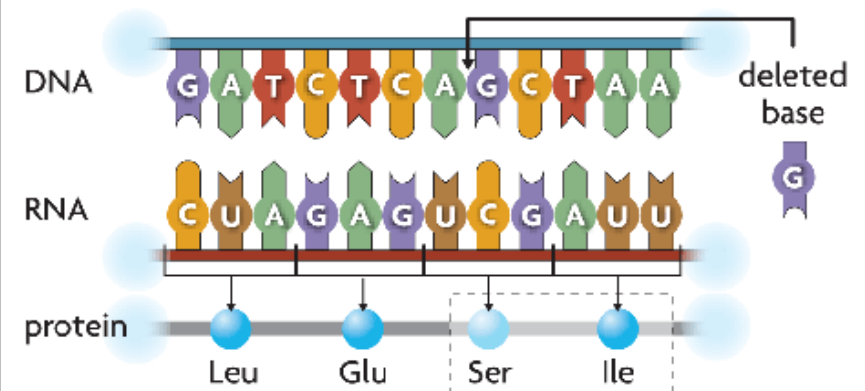
Point mutation



Frameshift mutation (insertion)



Frameshift mutation (deletion)



8.7 Mutations

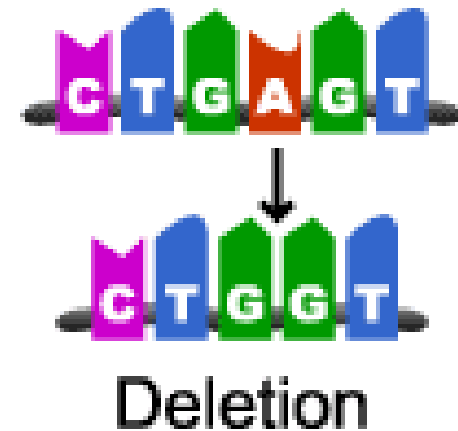
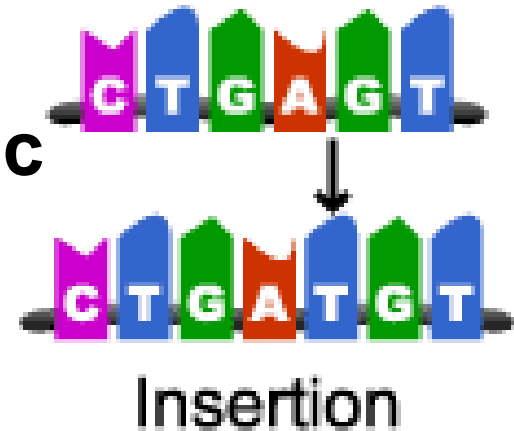
- **Frameshift Mutations** – shifts the reading frame of the genetic message so that the protein may not be able to perform its function.

- Insertion

THE FAT CAT ATE THE RAT
THE FAT HCA TAT ETH ERA T

- Deletion

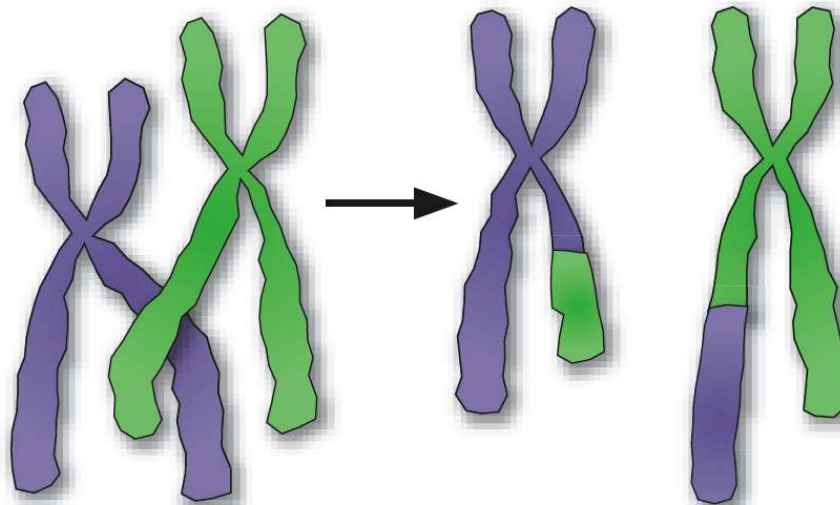
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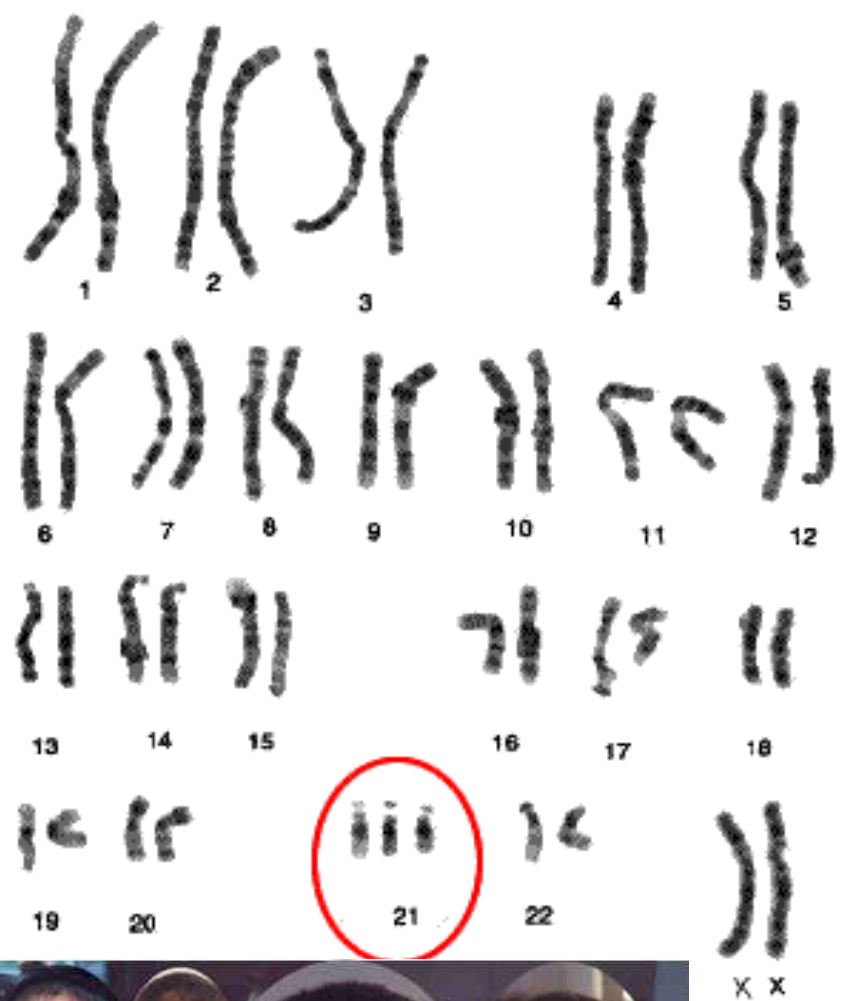
8.7 Mutations

- **Chromosomal mutations** affect many genes (we will look at these more in depth when we cover genetic disorders).
 - Chromosomal mutations may occur during **crossing over (prophase I of meiosis I)**
 - Chromosomal mutations affect many genes.
 - Gene duplication results from unequal crossing over.

Gene duplication

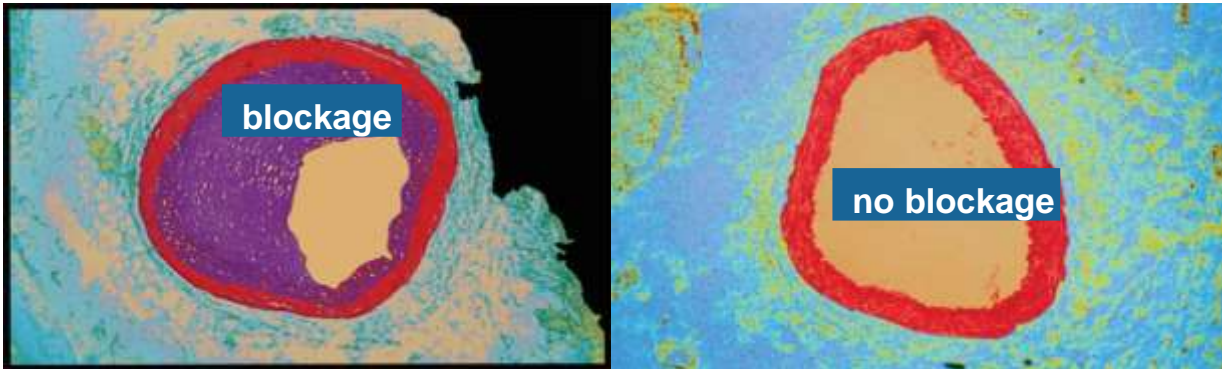


- **Nondisjunction** is another form of chromosomal mutation.
 - Occurs during **meiosis** when sister chromatids do not separate properly in Meiosis **II**.
 - One gamete has **2** copies of the chromosomes and the other gamete has **none**.
 - **Down's Syndrome** is caused by the gamete (typically the egg) that has two copies of chromosome **#21**. The resultant embryo would have **3** copies of #21 instead of two (Called **Trisomy 21**).



▶ Mutations may or may not affect phenotype (physical characteristics).

- Chromosomal mutations tend to have a **big effect**.
- Some gene mutations change phenotype.
 - A mutation may cause a premature stop codon.
 - A mutation may change protein shape or the active site.
 - A mutation may change gene regulation.



CYSTIC FIBROSIS - a genetic disease most commonly caused by a specific deletion – the most common one is a DNA deletion of a TTT sequence. It causes an overproduction of thick, sticky mucus.

8.7 Mutations

- **There are two ways in which DNA can become mutated:**
 - **Mutations can be inherited.**
Parent to child
 - **Mutations can be acquired.**
Environmental damage
Mistakes when DNA is copied
- ▶ **Mutations can be caused by several factors.**
 1. Replication errors can cause mutations.
 2. Mutagens, such as UV light rays and chemicals, can cause mutations.
 3. Some cancer drugs use mutagenic properties to kill cancer cells.



Rachel Carson was one of the first ecologists to warn against the widespread use of pesticides and other potential mutagens & toxins.

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"YOUR CASE WOULD MAKE A GREAT MEDICAL JOURNAL ARTICLE,
BUT FRANKLY, THE NATIONAL ENQUIRER PAYS MORE."

8.7 Mutations

The genetic code matches each RNA **codon** with its amino acid or function.

		Second base										
		U		C		A		G				
First base	U	UUU	phenylalanine (Phe)	UCU	serine (Ser)	UAU	tyrosine (Tyr)	UGU	cysteine (Cys)	U		
		UUC		UCC			UAC		UGC		C	
		UUA	leucine (Leu)	UCA			UAA	STOP	UGA	STOP	A	
		UUG		UCG			UAG	STOP	UGG	tryptophan (Trp)	G	
	C	CUU	leucine (Leu)	CCU	proline (Pro)	CAU	histidine (His)	CGU	arginine (Arg)	U		
		CUC				CAC		CGC			C	
		CUA				CCA		CAA		glutamine (Gln)	CGA	A
		CUG				CCG		CAG			CGG	G
	A	AUU	isoleucine (Ile)	ACU	threonine (Thr)	AAU	asparagine (Asn)	AGU	serine (Ser)	U		
		AUC				AAC		AGC		C		
		AUA				AAA	lysine (Lys)	AGA	arginine (Arg)	A		
		AUG	methionine (Met)	ACG			AAG		AGG	G		
	G	GUU	valine (Val)	GCU	alanine (Ala)	GAU	aspartic acid (Asp)	GGU	glycine (Gly)	U		
		GUC				GCC		GGC			C	
		GUA				GCA		GAA		glutamic acid (Glu)	GGA	A
		GUG				GCG		GAG			GGG	G
		Third base										

1 Find the first base, C, in the left column.

2 Find the second base, A, in the top row. Find the box where these two intersect.

3 Find the third base, U, in the right column. CAU codes for histidine, abbreviated as His.