

## NONSENSE MUTATION

Wife: He treats our marriage like it's a Biochemistry lesson, he's always talking nonsense.

Therapist: Is that true?

Husband: UAG, UAA, UGA.



*NONSENSE MUTATIONS* OCCUR WHEN THE DNA CHANGE CREATES A PREMATURE STOP CODON WHICH TERMINATES THE POLYPEPTIDE

- CYSTIC FIBROSIS IS AN EXAMPLE OF A DISEASE WHICH CAN RESULT FROM A NONSENSE MUTATION (THIS MAY NOT BE THE ONLY CAUSE THOUGH)

## SILENT MUTATION

First position (5' end)	Second position				Third position (3' end)
	U	C	A	G	
U	Phe	Ser	Tyr	Cys	U
	Phe	Ser	Tyr	Cys	C
	Leu	Ser	Stop	Stop	A
	Leu	Ser	Stop	Trp	G
C	Leu	Pro	His	Arg	U
	Leu	Pro	His	Arg	C
	Leu	Pro	Gln	Arg	A
	Leu	Pro	Gln	Arg	G
A	Ile	Thr	Asn	Ser	U
	Ile	Thr	Asn	Ser	C
	Ile	Thr	Lys	Arg	A
	Met	Thr	Lys	Arg	G
G	Val	Ala	Asp	Gly	U
	Val	Ala	Asp	Gly	C
	Val	Ala	Glu	Gly	A
	Val	Ala	Glu	Gly	G

▲ Table 1

*SILENT MUTATIONS* OCCUR WHEN THE DNA CHANGE DOES NOT ALTER THE AMINO ACID SEQUENCE OF THE POLYPEPTIDE

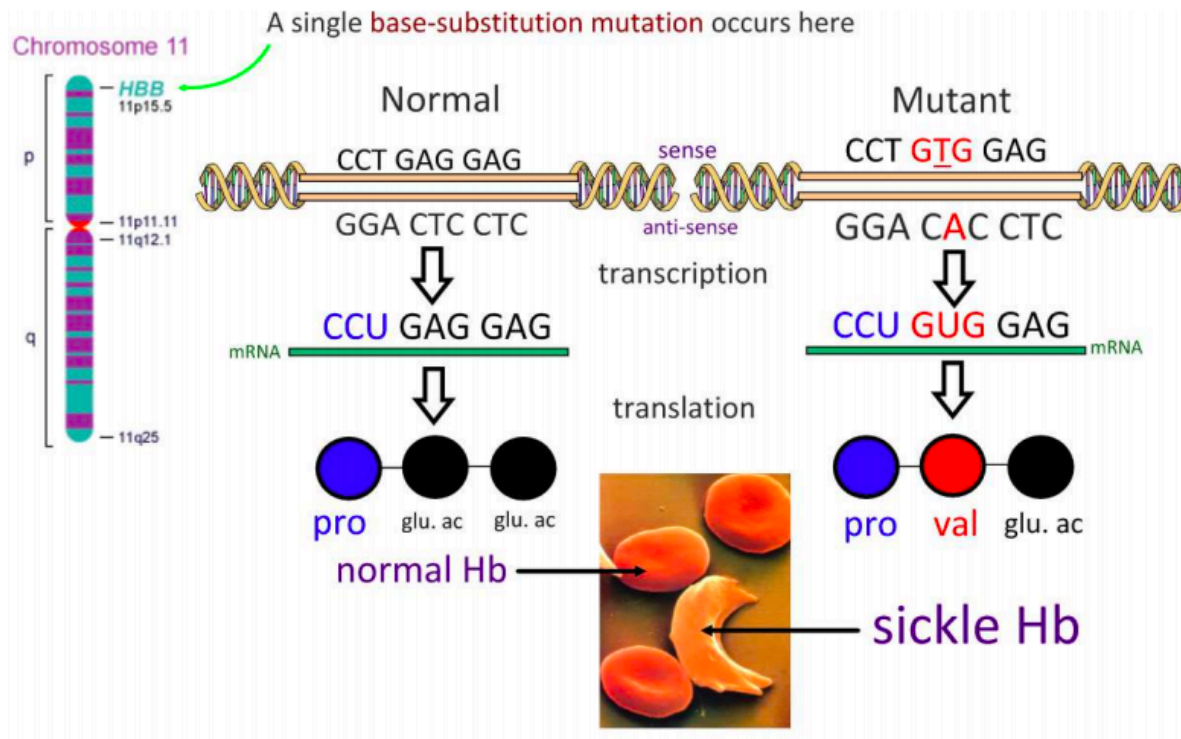
■ THIS IS POSSIBLE BECAUSE THE GENETIC CODE IS **DEGENERATE** AND CERTAIN CODONS MAY CODE FOR THE SAME AMINO ACID.

LOOK AT THE CODON WHEEL, WHICH CODONS WOULD YOU DESCRIBE AS DEGENERATE?

## MIS-SENSE MUTATION

*MISSENSE MUTATIONS* OCCUR WHEN THE DNA CHANGE ALTERS A SINGLE AMINO ACID IN THE POLYPEPTIDE CHAIN

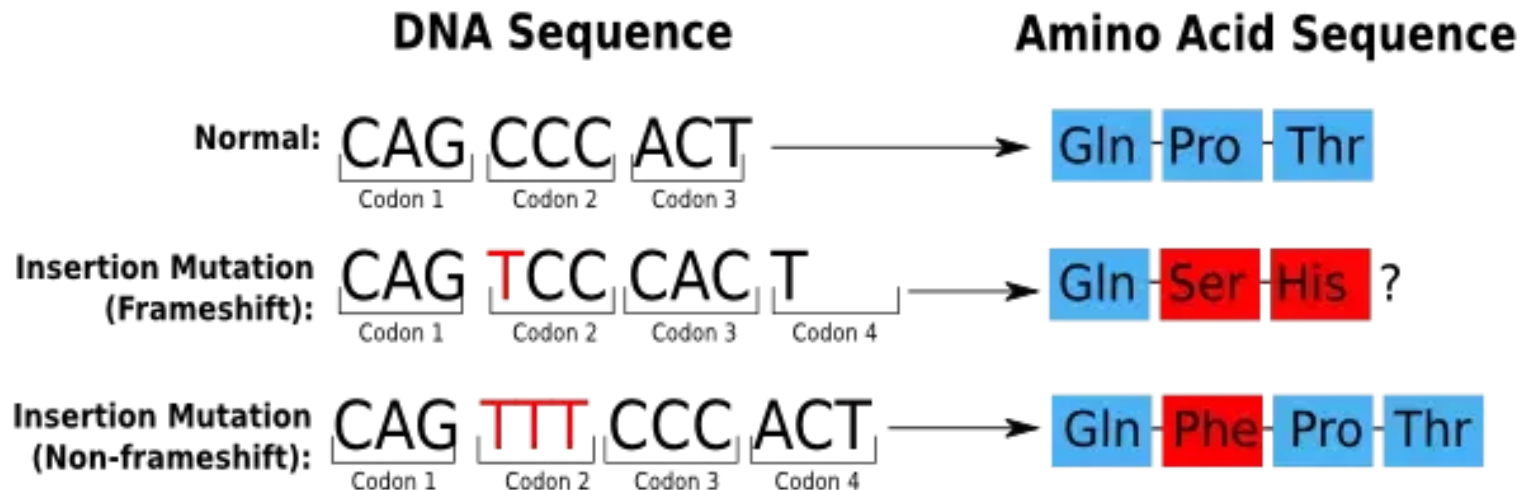
- SICKLE CELL ANEMIA IS AN EXAMPLE OF A DISEASE CAUSED BY A **SINGLE BASE SUBSTITUTION MUTATION** ( WHICH AMINO ACID CHANGES AND WHAT DOES IT CHANGE TO ?)



## ADDITION (INSERTION)

*THIS IS AN EXAMPLE OF A 'FRAMESHIFT MUTATION' MEANING THAT THE ADDITION OF A BASE ALTERS THE READING FRAME OF THE GENE*

- THIS CHANGE WILL AFFECT EVERY CODON BEYOND THE POINT OF MUTATION AND THUS MAY DRAMATICALLY CHANGE THE AMINO ACID SEQUENCE



# DELETION

*THIS IS AN EXAMPLE OF A 'FRAMESHIFT MUTATION' MEANING THAT THE DELETION OF A BASE ALTERS THE READING FRAME OF THE GENE*

- THIS CHANGE WILL AFFECT EVERY CODON BEYOND THE POINT OF MUTATION AND THUS MAY DRAMATICALLY CHANGE THE AMINO ACID SEQUENCE

AROUND 70% OF CASES OF CYSTIC FIBROSIS DUE TO THE DELETION OF THREE NUCLEOTIDES THAT RESULTS IN A LOSS OF THE AMINO ACID PHENYLALANINE. THIS RESULTS IN A FAULTY MEMBRANE PROTEIN AND SUBSEQUENT RESPIRATORY, DIGESTIVE ISSUES DUE TO THE BUILD-UP OF MUCUS.

