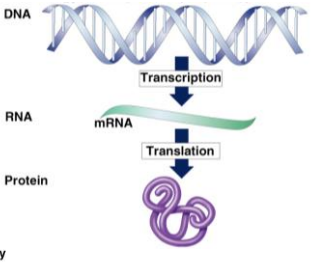
Mutations



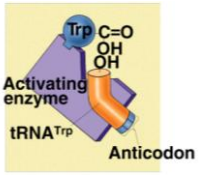
10 μm

Val His Leu Thr Pro Val Glu
1 2 3 4 5 6 7

(b) Sickled red blood cells and the primary structure of sickle-cell hemoglobin

Universal Code

- Code is redundant
  - several codons for each amino acid
  - "wobble" in the tRNA
  - "wobble" in the aminoacyl-tRNA synthetase enzyme that loads the tRNA

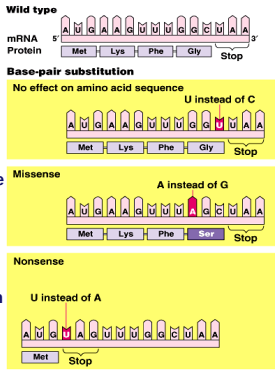


First base (5′ end)	Second base: U	Second base: C	Second base: A	Second base: G	Third base (3′ end)
U	UUU Phe	UCU Ser	UAU Tyr	UGU Cys	U
	UUC Phe	UCC Ser	UAC Tyr	UGC Cys	C
	UUA Leu	UCA Ser	UAA Stop	UGA Stop	A
	UUG Leu	UCG Ser	UAG Stop	UGG Trp	G
C	CUU Leu	CCU Pro	CAU His	CGU Arg	U
	CUC Leu	CCC Pro	CAC His	CGC Arg	C
	CUA Leu	CCA Pro	CAA Gln	CGA Arg	A
	CUG Leu	CCG Pro	CAG Gln	CGG Arg	G
A	AUU Ile	ACU Thr	AAU Asn	AGU Ser	U
	AUC Ile	ACC Thr	AAC Asn	AGC Ser	C
	AUA Ile	ACA Thr	AAA Lys	AGA Arg	A
	AUG Met or start	ACG Thr	AAG Lys	AGG Arg	G
G	GUU Val	GCU Ala	GAU Asp	GGU Gly	U
	GUC Val	GCC Ala	GAC Asp	GGC Gly	C
	GUA Val	GCA Ala	GAA Glu	GGA Gly	A
	GUG Val	GCG Ala	GAG Glu	GGG Gly	G

Mutations

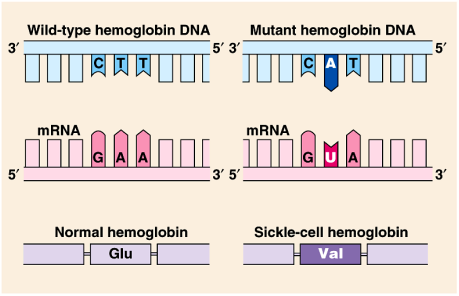
- Point mutations
  - single base change
  - base-pair substitution
    - silent mutation
      - no amino acid change
      - redundancy in code
    - missense
      - change amino acid
    - nonsense
      - change to stop codon

When do mutations affect the next generation?

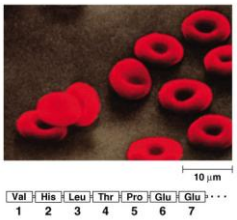


A Mutation Leads to Sickle Cell Anemia

What kind of mutation?



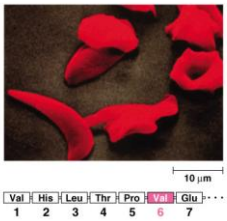
Sickle Cell Anemia



10 μm

Val His Leu Thr Pro Glu Glu
1 2 3 4 5 6 7

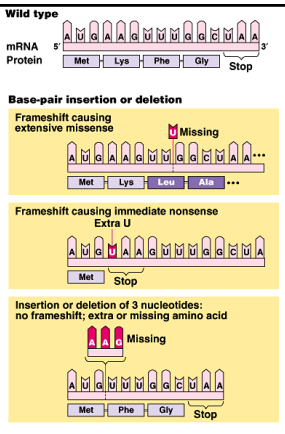
(a) Normal red blood cells and the primary structure of normal hemoglobin



10 μm

Val His Leu Thr Pro Val Glu
1 2 3 4 5 6 7

(b) Sickled red blood cells and the primary structure of sickle-cell hemoglobin

Mutations

- Frameshift
  - shift in the reading frame
    - changes everything "downstream"
  - insertions
    - adding base(s)
  - deletions
    - losing base(s)