

CHAPTER 18

OVERHEAD

Mutations

BLM 18.3.1

GUU-CAU-UUG-ACU-CCC-GAA-GAA
val - his - leu - thr - pro - glu - glu

- A** The normal coding sequence, with the codons in the top row and the resulting amino acids below them.

GUU-CAU-UUG-ACC-CCC-GAA-GAA
val - his - leu - thr - pro - glu - glu

- B** This mutation is silent, since the change in nucleotide sequence has no effect on the polypeptide product.

GUU-CAU-UUG-ACU-CCC-GUA-GAA
val - his - leu - thr - pro - val - glu

- C** This is a mis-sense mutation, since it causes the amino acid valine to be inserted in the place of glutamate within the polypeptide chain. The resulting protein is unable to transport oxygen effectively and produces a disorder known as sickle cell disease.

GUU-CAU-UAG
val - his - stop

- D** This substitution causes a nonsense mutation by changing the codon for the amino acid leucine (UUG) into a premature stop codon. No functional polypeptide will be produced from this gene.

GUU-CAU-UUG-ACU-CCC-GAA-GAA
val - his - leu - thr - pro - glu - glu

- A** The normal coding sequence, with the codons in the top row and the resulting amino acids below them.

↓
GUU-CAU-GUU-GAC-UCC-CGA-AGA A
val - his - val - asp - ser - arg - arg

- B** The insertion of a single nucleotide, in this case guanine, results in a frameshift mutation.

↑
GUU-CAU-UUG-CUC-CCG-AAG-AA
val - his - leu - leu - pro - lys

- C** Similarly, a deletion of even a single nucleotide, in this case adenine, also results in a frameshift mutation.

(Left) A nucleotide substitution can have varied effects, as shown on this portion of the gene that codes for human betaglobulin, one of the two polypeptides in the blood protein hemoglobin. (Right) Frameshift mutations are usually nonsense mutations.