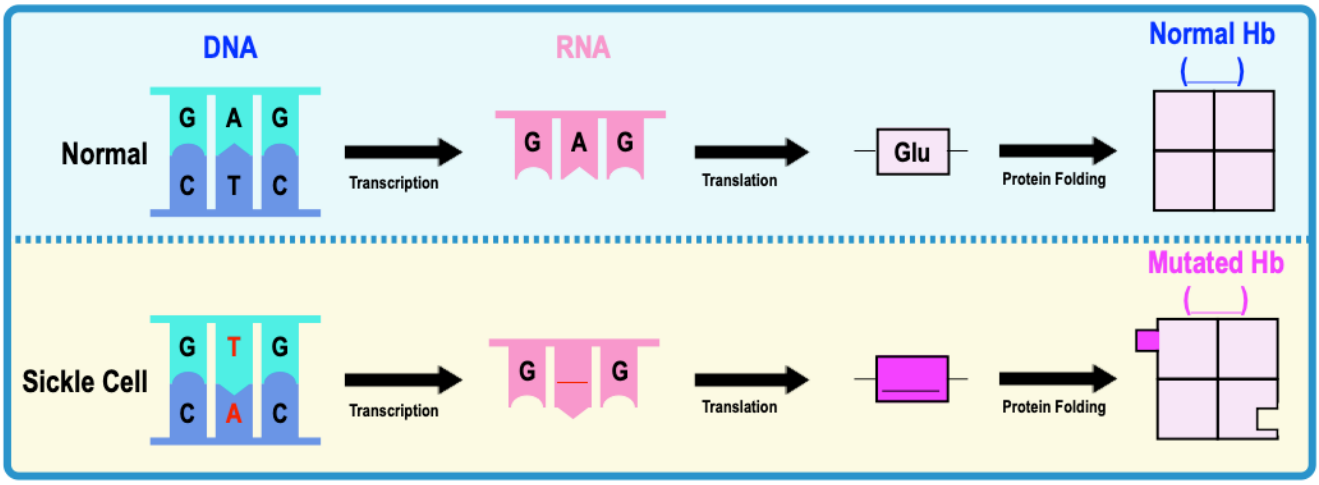
CONCEPT: SICKLE CELL ANEMIA

●*Sickle-Cell Anemia*: a disease due to a Hb _____ where red blood cells take a *sickle*-shape & cause health issues.

□ *Anemia*: _____ number of erythrocytes (red blood cells).

□ *Homozygous point* mutation causes a Glu-6 to change to a _____-6 in the β subunit of hemoglobin.



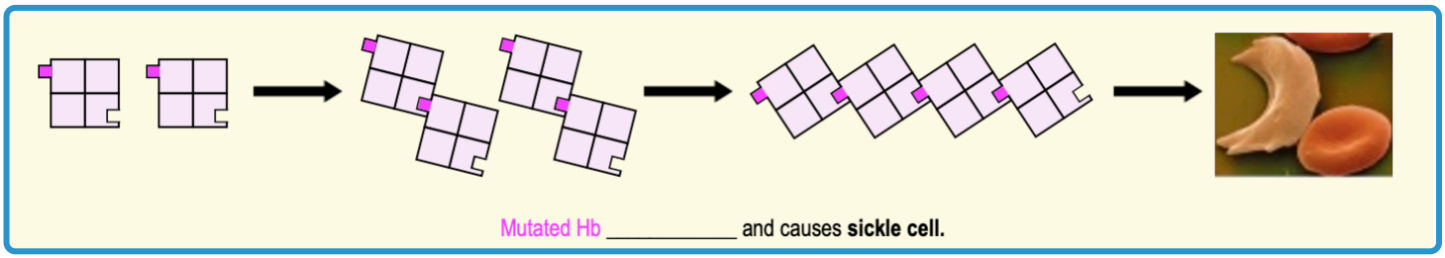
PRACTICE: The amino acid substitution of Val for Glu in Hemoglobin S (sickle-cell hemoglobin) is problematic because it substitutes a _____ amino acid residue with a _____ amino acid residue.

a) Polar ; nonpolar. b) hydrophobic ; charged. c) Aromatic ; Polar. d) Nonpolar ; aromatic.

Agglutination of Mutated Hb Causes Sickle-Cell

●The mutation above leads to _____ of mutated hemoglobin chains due to the *hydrophobic effect*.

□ Hemoglobin's O_2 affinity & allosteric properties are unaffected, but the Hb aggregates *deform* the red blood cell.



PRACTICE: In sickle cell anemia, the molecular basis of the malfunction of the hemoglobin molecule is:

a) Faulty binding of iron by the heme groups.

b) Insufficient iron in the diet.

c) Substitution of a single amino acid in the DNA sequence.

d) Reduced affinity for oxygen.

e) Silent mutation in the DNA sequence causing a single amino acid substitution.

f) Point mutation in the DNA sequence causing a single amino acid substitution.

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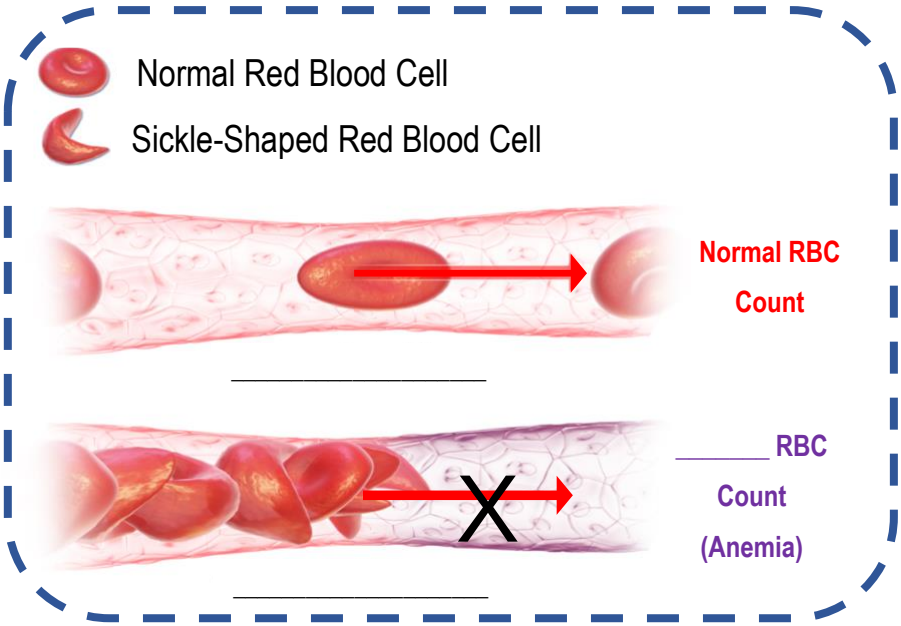
Physiological Effects of Sickle Cell Anemia

●Sickled red blood cells can become trapped in small blood vessels, which *impairs* circulation & can cause organ *damage*.

□ Sickled red blood cells also *rupture* more easily, leading to _____ (*low* red blood cell count).

EXAMPLE: Sickle Cell Anemia Physiological Effects.

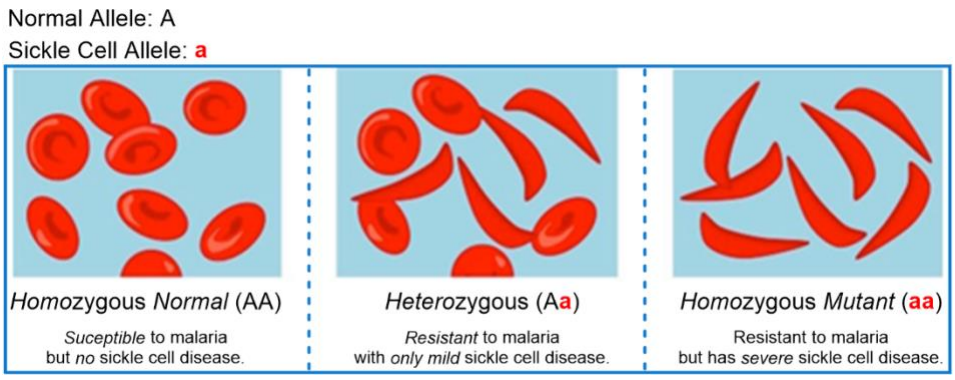


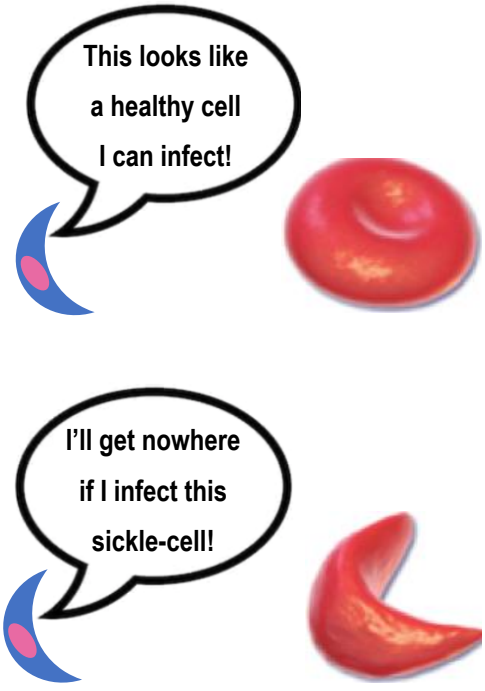
Malaria Resistance & Sickle Cell Anemia

●*Heterozygous* individuals for sickle-cell trait have _____ to malaria but do NOT have sickle-cell health issues.

□ *Heterozygous* sickle-cell trait is selected for by *natural selection* in areas of the world with malaria.

□ Exact mechanism of malaria resistance is not yet fully understood.





CONCEPT: SICKLE CELL ANEMIA

PRACTICE: Match the following key terms with their appropriate image (A or B):

a) HbS: Image _____
b) HbA: Image _____
c) Malaria resistance: Image _____
d) Heterozygous (Aa): Image _____
e) Homozygous (AA): Image _____
f) Low RBC count: Image _____
g) RNA sequence **GAG**: Image _____
h) RNA sequence **GUG**: Image _____

Image A

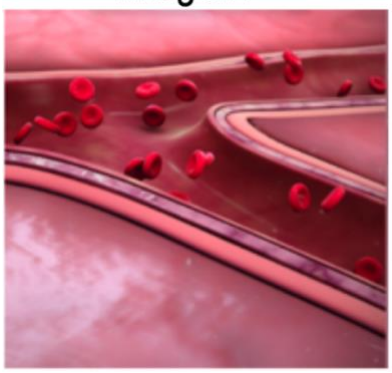
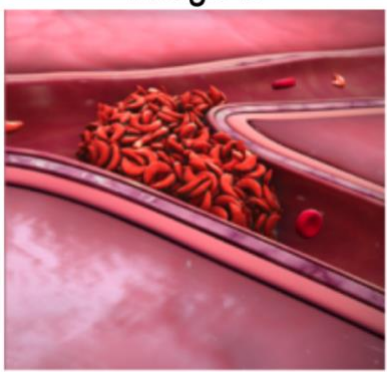


Image B



PRACTICE: Researchers investigating the sickle-cell mutation want to separate the mutant hemoglobin protein (HbS) from the normal adult hemoglobin (HbA) that was extracted from a heterozygous patient's blood serum sample. In order to do this, they perform gel electrophoresis at a pH of 8.5, where most normal hemoglobin proteins are negatively charged. The protein sample is added to the negative pole and migrates towards the positive pole when current is applied. The result of the gel is shown below. Label which band represent HbS and which band represents HbA.