

Dangerously Thin: A Case Study on the Genetic Code

Henry Blake (65 y/o male) noticed that his left leg was swollen just inferior to the knee. His doctor diagnosed him with deep vein thrombosis (DVT, a disorder that gets its name from the blood clots that form in a vein deep within the leg) and placed him on a blood thinner called warfarin. Henry returned to the doctor's office after suffering from frequent nose bleeds. A laboratory test revealed that Henry's blood was not clotting normally – even for someone on warfarin. Dr. Strickland immediately took Henry off of his warfarin treatments but his abnormal results continued.

Through genetic testing, Henry was found to carry a mutation in a gene for an enzyme called *CYP2C9*. This enzyme can break down many drugs currently in use, including warfarin. Henry was considered a poor metabolizer (PM) because he was not making any fully functional enzyme. Tests showed that Henry carries mutations in both chromosomes with the *CYP2C9* gene. In both copies of the gene, the 1075th nucleotide has been changed from an adenine (A) to a cytosine (C). This mutation converts an ATT triplet code in the coding strand of the DNA molecule to CTT.

Short Answer Questions

1. Why was Henry's blood dangerously thin even after he stopped taking warfarin?
2. Why did the mutation lead to a malfunction in Henry's *CYP2C9* enzyme?

Questions 3 – 5: Consider the following DNA sequence found on a different region the template strand of Henry's *CYP2C9* gene: **TTACCGAGA**

3. How many codons does this DNA sequence contain?
4. What would be the sequence of the mRNA after this sequence is transcribed?
5. What would be the amino acid sequence for this region? (Use chart on back)

Dangerously Thin Case Study Answer

N Colangelo



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