

Transformation Project

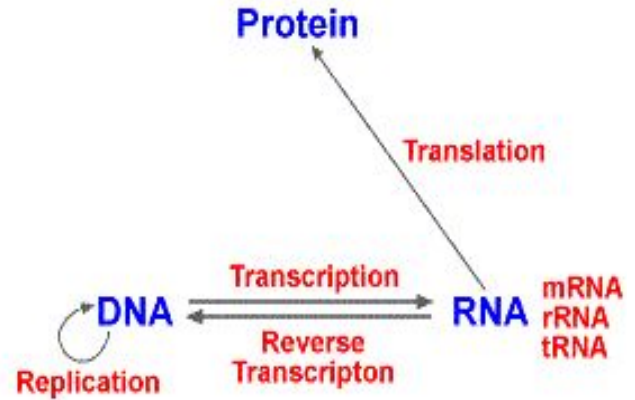
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Outline

- Central Dogma
- Mutations
- Mutations in Genes
- vWF- von Willebrand Factor
- von Willebrand Disease
- vFW Diagram
- Treatment
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Central Dogma

- Central Dogma is the transfer of genetic information within an organism.
- Often used when referring to the the process of DNA “making” proteins.
- The DNA makes a complementary mRNA to send to the ribosomes.
- The mRNA then uses codons (3 Nitrogen Bases) to send for a specific amino acid
- These amino acids make up the protein.



-An image of the process that sends the DNA signals to the Ribosomes

Mutations



-An image of DNA, and the red marking an area of mutation.

- Mutations are changes in DNA, more specifically their Nitrogen bases.
- These Mutations can only happen in DNA
- Caused by damage to DNA.

Mutations in Genes

- When there is a mutation there can be an effect or no effect.
- This is due to the variability of the codons that can code for an amino acid.
- For instance, Leucine has 3 different codons that call for it, 4 of which share the first 2 bases, meaning as long as the first 2 bases are CU, the 3rd doesn't matter. It will always code for Leucine.

Point Mutations

- **Silent mutation** = no change to protein

AUGCGUGUAUACGCAUGCGAGUGA



MetArgValTyrAlaCysGluStop

AUGCGUGUAUAGCUUGCGAGUGA



MetArgValTyrAlaCysGluStop

Regents Biology

The code has repeats in it!



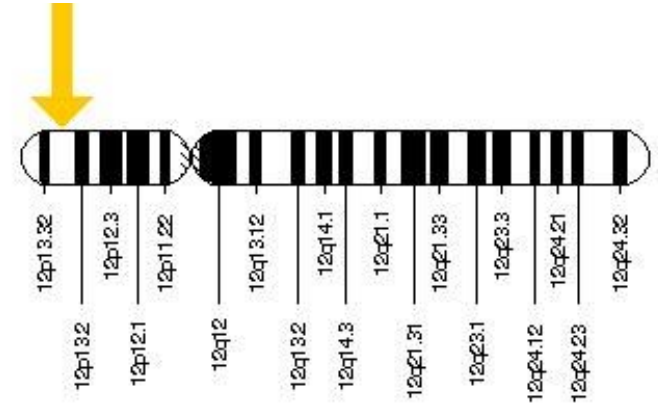
-Another example of mutation that does not affect the protein.

Mutations in Genes -Continued

- When a mutation occurs then the gene will produce a different amino acid and in turn a protein that is either completely different or not functioning.
- This is due to that proteins are 3-Dimensional and that the sequence of amino acids changes the shape, thus the effectiveness of the protein
- There is a rare chance that this mutation will not affect the proteins shape, and that it will work.

vWF- von Willebrand Factor

- Located on chromosome 12
 - Base pairs 5,948,873 to 6,124,669 (contains 175,796 base pairs)
- Protein “von Willebrand Factor”
 - Binds to other proteins to contribute to blood clotting.



Model of VWF's location on chromosome 12

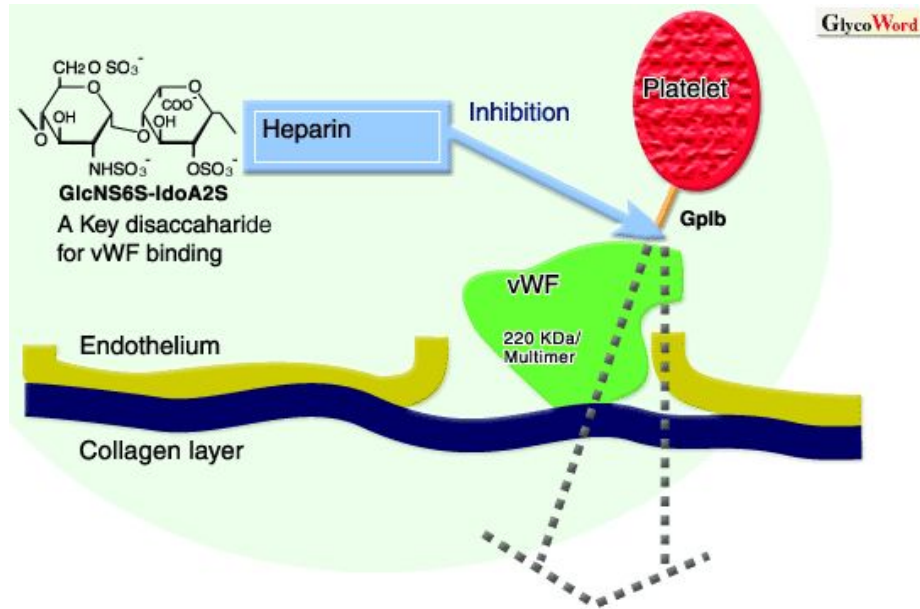
von Willebrand Disease

- Lack of von Willebrand Disease leads to reduced blood clotting
- Causes lack of clotting or slower clotting.
- Symptoms include massive nose bleeds, heavy bruising, prolonged bleeding from cuts, surgery, menstruation etc.

von Willebrand Disease -Continued

- Usually inherited from only one parent though it is possible in rare cases for it to be inherited from both.
- Affects 1 in 100 to 10,000 people (most cases are too minor to take notice to)

vWF Diagram



-The diagram shows the vWF attaching to the blood vessel wall, allowing other cells to bind to it to allow clotting.

Treatments

- Most cases are mild and do not require treatment
 - Only necessary if an accident or surgery occurs
- Hormone desmopressin is taken by nasal spray or injection.
 - Stimulates release of VWF cells
- If unable to take desmopressin, or VWF is nonresponsive, an infusion of von Willebrand Factor can also be another treatment

Questions

Any Questions?



Citation

- http://www.ndsu.edu/pubweb/~mcclean/plsc731/dna/central_dogma.gif
- <http://www.nhlbi.nih.gov/health/health-topics/topics/vwd/treatment>
- <http://www.doctortipster.com/wp-content/uploads/2012/02/Mutation.jpg>
- <http://upload.wikimedia.org/wikipedia/commons/thumb/3/3e/Desmopressin.svg/220px-Desmopressin.svg.png>
- <http://image.slidesharecdn.com/12-150201134109-conversion-gate02/95/124-mutations-7-638.jpg?cb=1422819712>
- <http://ghr.nlm.nih.gov/gene/VWF>
- <http://www.beyondpositive.org/wp-content/uploads/2014/01/question.png>