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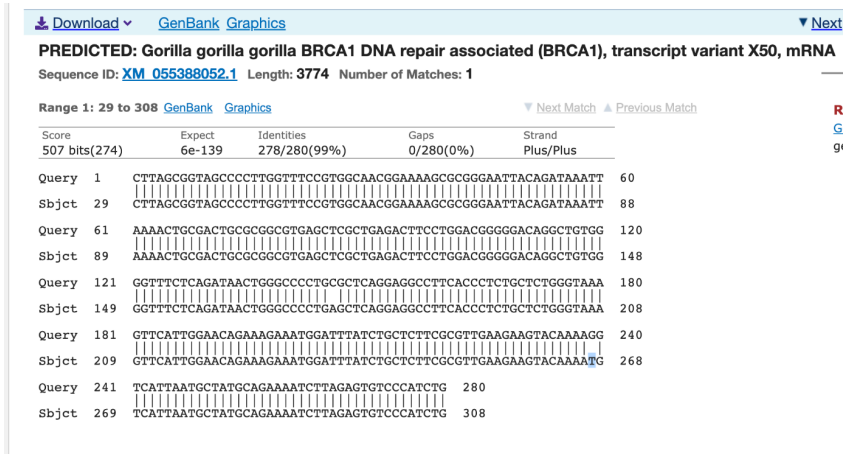
Part 1 Question:

1. Where is the mutation located and what is the nature of the mutation? (example substitution, nonsense mutation, deletion, insertion).

Substitution mutation:



From the screenshot above we can see that there are 2 **substitutions**, one at position 175 (C > A) and another at position 239 (G > T)

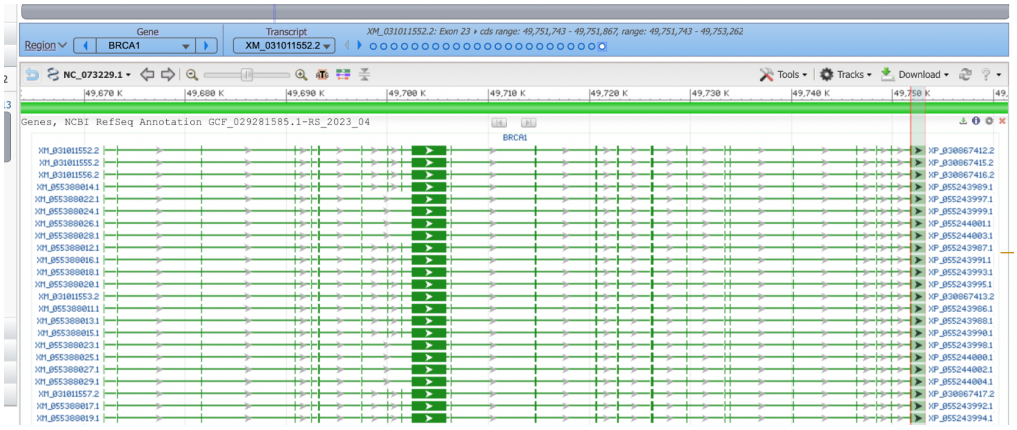
Part 2.

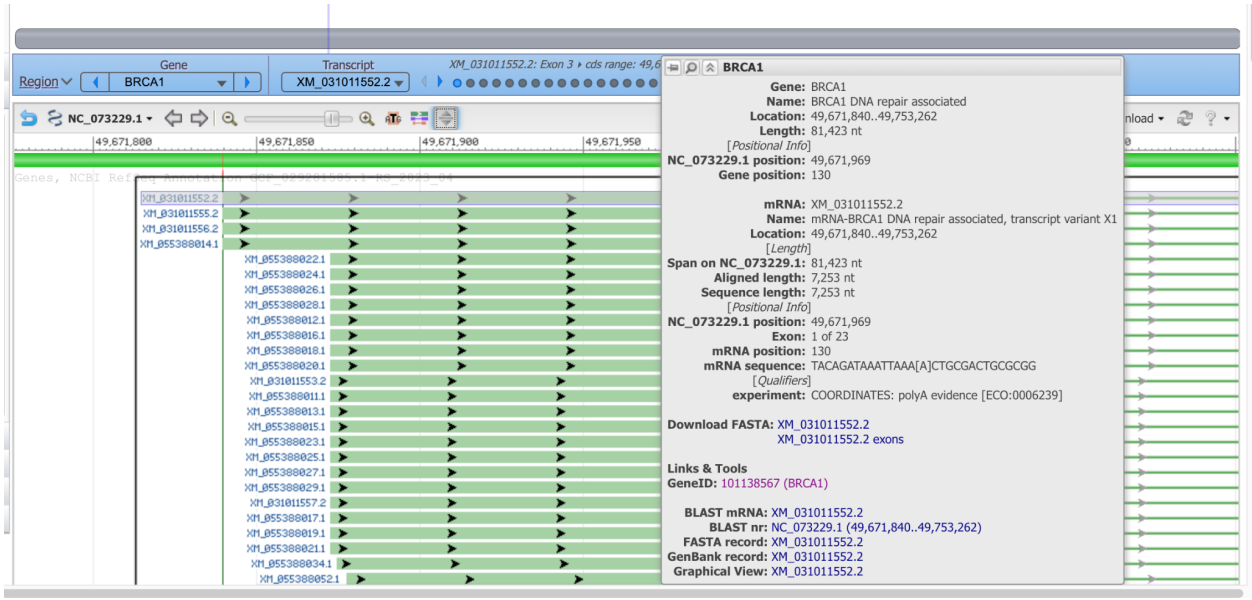
1. In regards to genomic DNA, would you expect to see introns or exons or both? Explain.

Both sequences with mutations are the same length, so both introns and exons can be expected. If there was only one or the other, the lengths would differ.

2. How many introns and exons are in the first transcript listed?

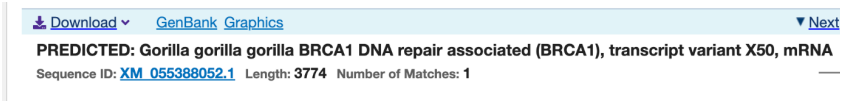
23 exons and 87 introns.





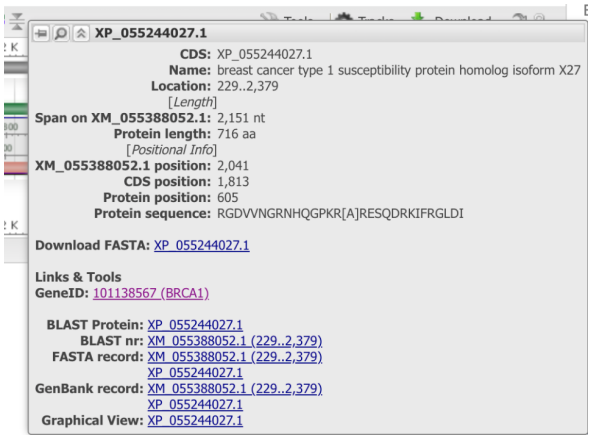
3. What is the length of the mRNA transcript?

The length is 3774



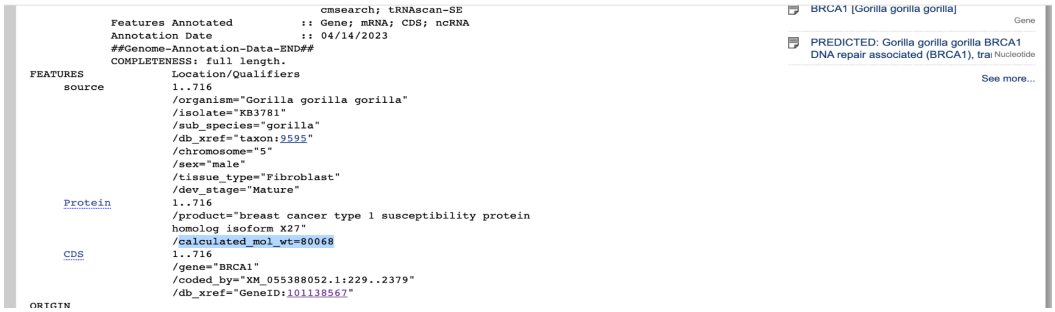
4. What is the number of amino acids of the protein?

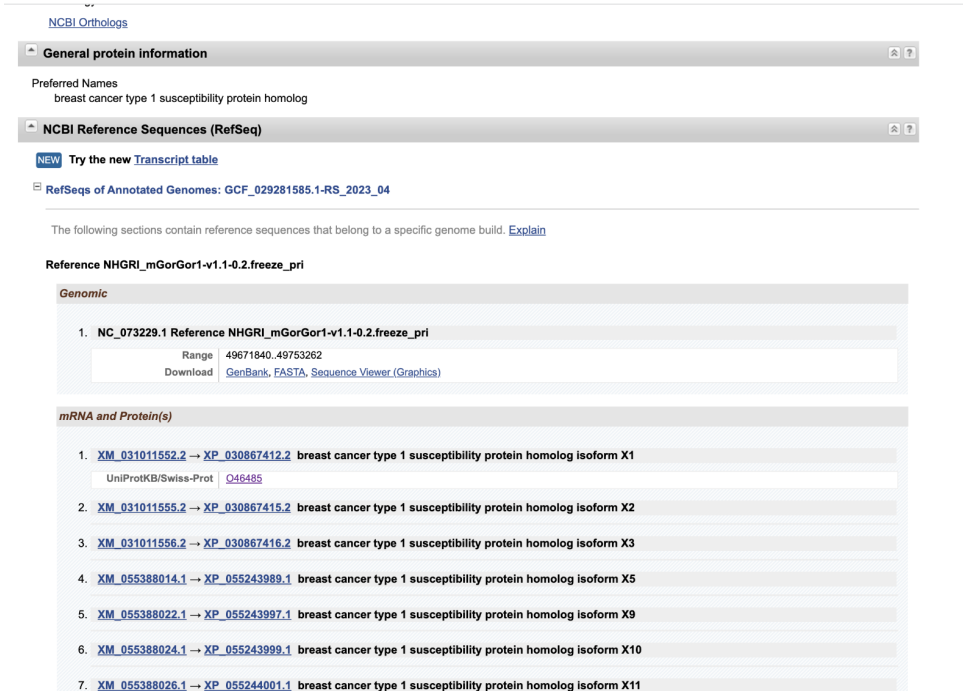
716 amino acids



5. What is the molecular weight of the protein?

Protein weight mol weight is 80068





Part 3.

1. How was the gene mutation initially identified?

The gene mutation was initially identified by noting the difference between letter on the coding and transcribed strands of DNA. The mutation was noted to be a substitution mutation because the letters were different rather than missing on the transcribed strand.

2. State which diseases you can find this mutated gene.

If the tumor suppressant is suppressed due to the overlap of introns and excess of substitution mutations, tumors will begin to appear. There can be an under expression of this gene or and over expression, of course because the body loves homeostasis this would affect the body. An under expression can cause health issues, additionally an overexpression can cause issues like tumor growth and cancer.

3. What chromosome is this gene located on?

The gene is located on chromosome 17q21. Now, people can get tested for the gene to know if they have an over-expression of this gene (at risk for tumors\ cancer).

4. What is the function of the normal gene?
The function of the normal gene BRCA 1, would be to provide instructions for making proteins, to prevent or repair damaged DNA.