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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:

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PREDICTED: Gorilla gorilla gorilla BRCA1 DNA repair associated (BRCA1), transcript variant X50, mRNA
Sequence ID: [XM_055388052.1](#) Length: 3774 Number of Matches: 1

Range 1: 29 to 308 GenBank Graphics Next Match Previous Match

Score	Expect	Identities	Gaps	Strand
507 bits(274)	6e-139	278/280(99%)	0/280(0%)	Plus/Plus

Query 1 CTTAGCGGTAGCCCTTGGTTTCCTGGCAACGAAAGCGGGAAATTACAGATAAAAT 60
Sbjct 29 CTTAGCGGTAGCCCTTGGTTTCCTGGCAACGAAAGCGGGAAATTACAGATAAAAT 88

Query 61 AAAACTGCGACTGCCCGCGCTGAGCTCGCTGAGACTTCTGGACGGGGACAGGCTGTGG 120
Sbjct 89 AAAACTGCGACTGCCCGCGCTGAGCTCGCTGAGACTTCTGGACGGGGACAGGCTGTGG 148

Query 121 GGTTCCTCAGATAACTGGGCCCTCGCTCAGGAGGCTTCACCTCTGCTCTGGGTAAT 180
Sbjct 149 GGTTCCTCAGATAACTGGGCCCTCGCTCAGGAGGCTTCACCTCTGCTCTGGGTAAT 208

Query 181 GTTCATTGGAACAGAAAGAAATGGAATTTATCTGCTCTTCGCGTTGAAGAAGTACAAAAG 240
Sbjct 209 GTTCATTGGAACAGAAAGAAATGGAATTTATCTGCTCTTCGCGTTGAAGAAGTACAAAAG 268

Query 241 TCATTAATGCTATGCAGAAAATCTTAGAGTGTCCTCATCTG 280
Sbjct 269 TCATTAATGCTATGCAGAAAATCTTAGAGTGTCCTCATCTG 308

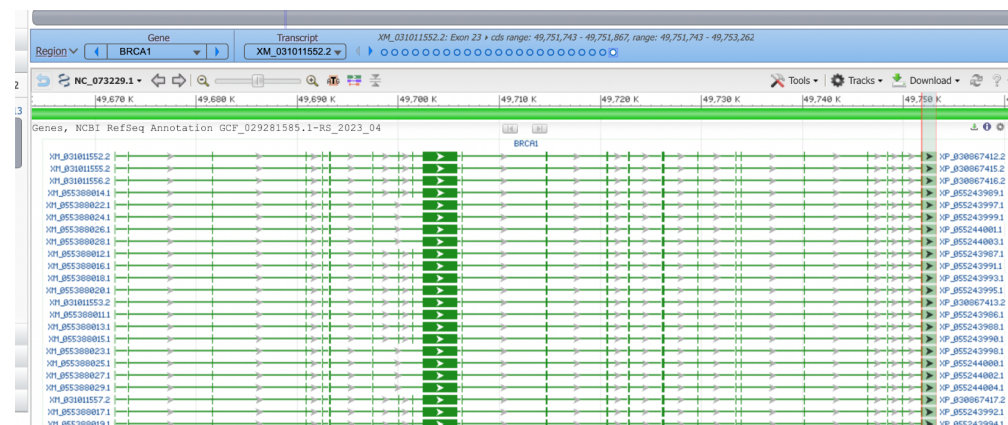
From the screenshot above we can see that there are 2 **substitutions**, one at position 175 (C > A) and another at position 239 (G > T)

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.



Gene: BRCA1
 Name: BRCA1 DNA repair associated
 Location: 49,671,840..49,753,262
 Length: 81,423 nt
 [Positional Info]
 NC_073229.1 position: 49,671,969
 Gene position: 130
 mRNA: XM_031011552.2
 Name: mRNA-BRCA1 DNA repair associated, transcript variant X1
 Location: 49,671,840..49,753,262
 [Length]
 Span on NC_073229.1: 81,423 nt
 Aligned length: 7,253 nt
 Sequence length: 7,253 nt
 [Positional Info]
 NC_073229.1 position: 49,671,969
 Exon: 1 of 23
 mRNA position: 130
 mRNA sequence: TACAGATAAATTA[A]CTGCGACTGCGCGG
 [Qualifiers]
 experiment: COORDINATES: polyA evidence [ECO:0006239]
 Download FASTA: XM_031011552.2
 XM_031011552.2 exons
 Links & Tools
 GeneID: 101138567 (BRCA1)
 BLAST mRNA: XM_031011552.2
 BLAST nr: NC_073229.1 (49,671,840..49,753,262)
 FASTA record: XM_031011552.2
 GenBank record: XM_031011552.2
 Graphical View: XM_031011552.2

3. What is the length of the mRNA transcript?

The length is 3774

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PREDICTED: Gorilla gorilla gorilla BRCA1 DNA repair associated (BRCA1), transcript variant X50, mRNA

Sequence ID: [XM_055388052.1](#) Length: 3774 Number of Matches: 1

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

716 amino acids

CDS: XP_055244027.1
 Name: breast cancer type 1 susceptibility protein homolog isoform X27
 Location: 229..2,379
 [Length]
 Span on XM_055388052.1: 2,151 nt
 Protein length: 716 aa
 [Positional Info]
 XM_055388052.1 position: 2,041
 CDS position: 1,813
 Protein position: 605
 Protein sequence: RGDVVNQRNHQGPKR[A]RESQDRKIFRGLDI
 Download FASTA: [XP_055244027.1](#)
 Links & Tools
 GeneID: 101138567 (BRCA1)
 BLAST Protein: XP_055244027.1
 BLAST nr: XM_055388052.1 (229..2,379)
 FASTA record: XM_055388052.1 (229..2,379)
 GenBank record: XM_055388052.1 (229..2,379)
 Graphical View: XP_055244027.1

5. What is the molecular weight of the protein?

Protein weight mol weight is 80068

Features Annotated : Gene; mRNA; CDS; ncRNA
 Annotation Date : 04/14/2023
 ##Genome-Annotation-Data-END##
 COMPLETENESS: full length.

FEATURES

source
 Location/Qualifiers
 1..716
 /organism="Gorilla gorilla gorilla"
 /isolate="KB3781"
 /sub_species="gorilla"
 /db_xref="taxon:3595"
 /chromosome="5"
 /sex="male"
 /tissue_type="Fibroblast"
 /dev_stage="Mature"
 1..716
 /product="breast cancer type 1 susceptibility protein
 homolog isoform X2"
 /calculated_mol_wt=80068
 1..716
 /gene="BRCA1"
 /coded_by="XM_055388052.1:229..2379"
 /db_xref="GeneID:101138562"

ORIGIN

BRCA1 [Gorilla gorilla gorilla]
 Gene
 PREDICTED: Gorilla gorilla gorilla BRCA1
 DNA repair associated (BRCA1), tra Nucleotide
 See more...

NCBI Orthologs

General protein information

Preferred Names
 breast cancer type 1 susceptibility protein homolog

NCBI Reference Sequences (RefSeq)

[NEW](#) Try the new [Transcript table](#)

RefSeqs of Annotated Genomes: GCF_029281585.1-RS_2023_04

The following sections contain reference sequences that belong to a specific genome build. [Explain](#)

Reference NHGRI_mGorGor1-v1.1-0.2.freeze_pri

Genomic

1. [NC_073229.1](#) Reference NHGRI_mGorGor1-v1.1-0.2.freeze_pri

Range 49671840..49753262

Download [GenBank](#), [FASTA](#), [Sequence Viewer \(Graphics\)](#)

mRNA and Protein(s)

1. [XM_031011552.2](#) → [XP_030867412.2](#) breast cancer type 1 susceptibility protein homolog isoform X1
 UniProtKB/Swiss-Prot [Q46485](#)
2. [XM_031011555.2](#) → [XP_030867415.2](#) breast cancer type 1 susceptibility protein homolog isoform X2
3. [XM_031011556.2](#) → [XP_030867416.2](#) breast cancer type 1 susceptibility protein homolog isoform X3
4. [XM_055388014.1](#) → [XP_055243989.1](#) breast cancer type 1 susceptibility protein homolog isoform X5
5. [XM_055388022.1](#) → [XP_055243997.1](#) breast cancer type 1 susceptibility protein homolog isoform X9
6. [XM_055388024.1](#) → [XP_055243999.1](#) breast cancer type 1 susceptibility protein homolog isoform X10
7. [XM_055388026.1](#) → [XP_055244001.1](#) breast cancer type 1 susceptibility protein homolog isoform X11

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.