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**Bioinformatics**

**Assignment 6**

1. **Using Ensembl Ensembl Release 111 answer the following questions:**

1.) How many splice variants does the human BRCA2 gene have? **15**

2.) How many exons are in the longest splice variant(s) of BRCA2? **27**

3.) How many introns are in the longest splice variant(s) of BRCA2? **26**

4.) What chromosome is this gene on? **Chromosome 13**

5.) What are the gene synonyms of BRCA2? **BRCC2, FACD, FAD, FAD1, FANCD, FANCD1, XRCC11**

6.) How many orthologs of human BRCA2 are found in the African ostrich? **172 orthologues**

7.) In the human hemoglobin transcript ENST00000330597.5 HBG1-201, what is the sequence of the second exon?

**G**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1429943325;vf=735355253)**T**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847923528;vf=741376696)**CTG**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1343156232;vf=731218509)[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1589906467;vf=739070804)**TGTCT**[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1335958398;vf=730873977)**CC**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs41404150;vf=706976586)**A**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs35700518;vf=706959283)**GGACCC**[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs35977759;vf=706969491)**GA**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs33974602;vf=706896498)**G**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1589906450;vf=739070801)**T**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847923183;vf=741376580)**T**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1394056016;vf=733644608)**T**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs41475844;vf=706976788)**A**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1395960485;vf=733732899)**AGCTT**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1589906444;vf=739070800)**G**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847923033;vf=741376523)**CAACCTGT**[**CCTC**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1409662507;vf=734391537)

[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1409662507;v=rs1589906439;vf=734391537;vf=739070798)[**GCCTCTG**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1409662507;vf=734391537)[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1409662507;v=rs35746147;vf=734391537;vf=706960945)[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1409662507;vf=734391537)[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1173059624;vf=723098646)[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs778172072;vf=713544984)**CA**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1434319312;vf=735562609)**G**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1422261857;vf=734988779)**G**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1187559666;vf=723792915)**AACCC**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1485842420;vf=738016136)[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs539011424;vf=709947693)**AAGT**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847922732;vf=741376424)[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs34747494;vf=706924389)**A**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847922666;vf=741376399)[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847922641;vf=741376390)**CAC**[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1213522116;vf=725034610)**T**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs747508521;v=rs756489020;vf=712059015;vf=712495096)[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs747508521;vf=712059015)**CAAG**[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847922517;vf=741376345)[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847922517;v=rs1589906410;vf=741376345;vf=739070788)[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1208095608;vf=724775097)**GT**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs752995575;vf=712325850)**CTGACTTC**

[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1589906400;vf=739070785)**T**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1225872760;vf=725625116)**G**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs34049890;vf=706898654)**GA**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs33965337;vf=706896416)**A**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847922133;vf=741376219)**G**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1256614625;vf=727088779)**CA**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1061234;vf=706477848)**AAAG**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs755174349;vf=712431430)[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1589906380;vf=739070780)**CCTG**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs34435255;vf=706912864)**A**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847921855;vf=741376110)[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs33947112;vf=706896259)**ATCTCAAGGG**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1359816903;vf=732014326)**AC**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs751636715;vf=712259925)**TTTGCC**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847921684;vf=741376065)[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1415219099;vf=734657005)**GCTG**[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1376275271;vf=732796568)[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs766448778;vf=712977746)**TGA**

**AC**[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1464732955;vf=737010311)**GCACT**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847921541;vf=741376013)**T**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs527266118;vf=709349841)**ACA**[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1847921469;vf=741375992)**GCT**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs750370911;vf=712198199)**C**[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs34127117;vf=706901542)[**T**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1418054559;vf=734788224)**GTGGAT**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1190498683;vf=723934545)**CT**[**G**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1267554412;vf=727611202)**AGAACTT**[**C**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs765208645;vf=712917744)**A**[**A**](https://useast.ensembl.org/Homo_sapiens/ZMenu/TextSequence?db=core;factorytype=Location;g=ENSG00000213934;r=11:5248269-5249857;t=ENST00000330597;v=rs1257266010;vf=727120011)**G**

8.) What is the length of the first intron of this hemoglobin transcript? **122**

9.) How many pseudogenes are in the narwal genome annotation (NGI\_Narwhal\_1, INSDC Assembly GCA\_005190385.2)? **723**

1. **Using Genomicus web-code version 2021-08-15 and database version 109.01, answer the following questions:**

10.) What two genes flank BRCA2 in the human genome? **N4BP2L1 and ZAR1L**

11.) What genes flank the sonic hedgehog gene(s) in the betta fish? **rnf32 and rbm33a**

12.) How many copies of shh (sonic hedgehog) are in the betta fish? **19,556,708** - **19,563,316**

13.) How many copies of shh are in Atlantic salmon? **9,678,850** - **9,686,853**

1. **Using NCBI answer the following questions:**

14.) How many genome assemblies are available for Bos taurus on NCBI? **17**

15.) How many of the above assemblies are assembled into chromosomes? **11**

16.) How many chromosomes does Bos taurus have excluding sex chromosomes? **29**

17.) Which cow chromosome is the longest? **Seqoccin.Bt.char.v1.0**

18.) How many microRNAs are annotated on chromosome the assembly: Bos taurus ARS-UCD2.0? **1,033**

19.) If I am interested in learning more about the gene associate with the transcript: NM\_001104822, what is a good publication to read on PubMed?

**A good publication to read is Gene evolution and gene expression after whole genome duplication in fish: The PhyloFish database by Pasquier et al.**

20.) What is the translated coding sequence of NM\_001104822?

**1 gtcagaagca tcagcattta aaaaaaaaaa aaaatggacc tgctggccaa agcatctgtg**

**61 ctgctgttgc tgctcctgag cctcagcaac gctcaaactg acaatatgga ggaagcagaa**

**121 aacggttcat caaaggagga aatagacgag tctgaactgg aggacgtgtc ctccatcatc**

**181 ttcagaatga acaacaactc tatggaggaa ctgttggaag gagatcttgt tcttcccaaa**

**241 accaggaatg ccatgaagtg ctttggcgct ccagatagct gccgctggcc aaagtcttcc**

**301 aatggcatcg tgaaggttcc ttatgtggtt agcgacaact atgaaagtga cgagaaggaa**

**361 accattcgga acgccatgaa ggagtttgca gaaaaaacct gcattcactt tgttcctcgc**

**421 aacaatgaga gggcctacct gagccttgaa cccagatttg gctgcaagtc tatgatgggc**

**481 tatgttggtg acaaacaagt ggtggtgctt cagcggtttg gctgcataaa gcacgccgtc**

**541 atccagcatg agctcctgca tgctctgggt ttctaccacg agcacactcg gagcgaccgc**

**601 gaccagcatg taaaaatcaa ctgggaaaac atcattaaag atttcacaca caactttgat**

**661 aagaatgata ccgacaatct gggcaccccg tatgactatg gctccatcat gcactatgga**

**721 agaactgcct ttggaaaaga cagaaaggag accataaccc ccatccctaa ccccaaagct**

**781 gccattggcc aaacagagag gatgtcggac atcgatatac ttcgagtcaa taagctttac**

**841 aaatgttgag atgacatgag ctgaaaatga tatttgatgc ttcatgtggg tgtacgacat**

**901 taaagtgaat cggtaaaggg gagcgtttat ccttgt**