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Biology 294

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Genome Assignment

Chromosome Maps:

1. **What chromosome did you choose?**
   Chromosome 6

2/3. State the number of genes and base pairs on the chromosome you chose.

1900 genes

170 million base pairs (95% have been determined)

4. List one disease (due to a mutated gene) associated with this chromosome.

Spinocerebellar ataxia due to mutations on gene SCA1

5. State the normal function of the gene (not the disease) you listed in #4.

Gene SCA1 function is protein production of atazin-1 which will vary in size due to the mutation.

Introduction to BLAST:

6. What is the top sequence description match for your query sequence?

Homo sapiens CFTR promoter region (LOC111674463) on chromosome 7

7. What is the function of this sequence?

Long range interactions with enhancers which suggest complex regulation.

8. What gene (Protein) is the sequence associated with?

Cystic fibrosis transmembrane conductance regulator gene, and protein kinase A.

9. What does the encoded protein do in the body?

Encodes ATP-binding cassette transporters. Protein kinase A induces phosphorylation of the R domain that alters its position and allows NBD's dimerization to happen.

Citation:

Sala, A. et al. Role of Protein Kinase A-Mediated Phosphorylation in CFTR Channel Activity Regulation. *Front Physiol* **12**; https://doi.org/10.3389%2Ffphys.2021.690247 (2021).

10. For what disease is a mutated form of this gene responsible?

A mutation form of this gene could cause Cystic fibrosis.

11. On what chromosome is the gene located?

It is located on chromosome 7.

12. What species (stated the scientific name) other than Homo sapiens also has a 100% similarity for this sequence?

Another species with 100% similarity is the *Pongo abelii.*

13. Does it surprise you that this species also has 100% similarity in identity? Why or why not?

Yes, I was surprised to find out that the *Pongo abelii* has 100% similarity in this gene. I do know that we share a lot of similarities, but I did not expect it to be as far as to how we regulate our bodies by using proteins.

For each, state what the gene is.

14. NM_145556

Mus musculus TAR DNA binding protein (Tardbp), transcript variant 1, mRNA

15. NM_013444

Homo sapiens ubiquilin 2 (UBQLN2), mRNA

16. NM_001010850

Homo sapiens fusion (involved in t(12;16) in malignant liposarcoma) (FUS), transcript variant 2, mRNA

17. KJ174530

Homo sapiens superoxide dismutase-1 (SOD-1) gene, exon 1 and partial cds

18. What disease is associated with mutations of the genes referenced in 14-17? What is a common name of the disease?

ALS or Lou Gehrig's disease.

19. What is GenBank?

GenBank is a database available to the public that shares nucleotide sequences and protein translations.

20. What is the hierarchy among the National institutes of health, the national library of medicine, and the national center for biotechnology information? Where do GenBank and Blast belong?

The NIH would take primary hierarchy since it is the main association responsible for research in general. The next would come the NLM which is one of the many institutions under the NIH. After this comes the NCBI which is a division within the NLM. Following this would be GenBank as it is a publicly shared tool for the NCBI and lastly would come blast as it used GenBank's data for its search.

21. Reflect on one thing that you learned from doing this assignment.

That there are many public resources at our disposal which utilizes years of research and constant reviews. I also learned that it is a lot easier to research and learn about chromosomes and genes.