1. WHAT CHROMOSOME DID YOU CHOOSE? **5**
2 & 3. STATE THE NUMBER OF GENES AND BASE PAIRS ON THE CHROMOSOME YOU CHOSE. **1700 genes and 180 million base pairs**
4. LIST ONE GENE THAT IS LOCATED ON THIS CHROMOSOME. **SMN1**

5. STATE THE FUNCTION OF THE GENE YOU LISTED IN #4. **The function is currently being investigated by studies in rats and transgenic mice.**

6. WHAT IS THE SECOND SEQUENCE DESCRIPTION MATCH FOR YOUR QUERY SEQUENCE? **Homo sapiens CFTR (CFTR) gene, partial cds**

7. WHAT DOES THE ENCODED PROTEIN DO IN THE BODY? **Functions as a chloride channel**

8. FOR WHAT DISEASE IS A MUTATED FORM OF THIS GENE RESPONSIBLE? **Cystic fibrosis**

9. ON WHAT CHROMOSOME IS THE GENE LOCATED? **7**

10.CHOOSE A SPECIES (STATE THE
SCIENTIFIC NAME) OTHER THAN HOMO SAPIENS THAT ALSO HAS A 100% IDENTITY FOR THIS SEQUENCE? **Pongo abelii**

11. WHAT IS THE COMMON NAME FOR THIS SPECIES? **Sumatran orangutan**
12. DOES IT SURPRISE YOU THAT THIS SPECIES ALSO HAS A 100% SIMILARITY IN IDENTITY? **No**
WHY OR WHY NOT? **Both humans and Sumatran orangutans are primates so it would make sense that there are sequences that have 100% identity for both**
13.
a. WHAT IS THE GENUS AND SPECIES WITH THIS NUCLEOTIDE SEQUENCE? **Genus: sapajus species: apella**
b. WHAT IS THE COMMON NAME? **tufted capuchin**
c. HOW MANY GAPS OCCUR BETWEEN THE TWO SEQUENCES (THE ONE YOU ORIGINALLY
SUBMITTED AND ONE THAT HAS LESS THAN 100% QUERY COVER)? **1/119**

14. WHAT IS A GAP IN SEQUENCE ALIGNMENTS? **The absence of a region which is present in another sequence.**

FOR EACH, STATE WHAT THE GENE IS (#15-18). (Again, give the description of the gene or gene
product, not the nucleotide sequence.)

15. NM_145556 **Enables RNA polymerase II cis-regulatory region sequence-specific DNA binding activity and pre-mRNA intronic binding activity. Involved in positive regulation of protein import into nucleus; regulation of circadian rhythm; and regulation of protein stability. Acts upstream of or within RNA splicing.**

16. NM_013444 **Encodes a ubiquitin like protein**

17. NM_001010850 **Fusion protein produced as a result of chromosomal translocations in human cancers**

18. KJ174530 **The protein encoded by this gene binds copper and zin cions and is responsible for destroying free superoxide radicals in the body**

19. Search Google to answer the following: WHAT DISEASE IS ASSOCIATED WITH MUTATIONS OF
THE GENES REFERENCED IN #15-#18? WHAT IS A "COMMON NAME" OF THE DISEASE? (The
name of a person; Hint, hint...Baseball season just finished...)

frontotemporal lobar degeneration and amyotrophic lateral sclerosis. Common name for amyotrophic lateral sclerosis is Lou Gehrig's disease.

20. BLAST is possible because of the submission of DNA sequences to GenBank. WHAT IS GENBANK? **NIH genetic sequence database. An annotated collection of all publicly available DNA sequences.**

21. First, answer this question: WHAT IS cDNA? **Copy DNA also called complementary DNA is synthetic DNA that has been transcribed from mRNA through a reaction using the enzyme reverse transcriptase. cDNA contains only coding sequences**

22. WHAT IS THE SEQUENCE MATCH? **Beta Globin**

23. DO YOU SEE ANY DIFFERENCES BETWEEN THE TWO AMINO ACID SEQUENCES? **Yes**
24. IF YOU SAW DIFFERENCES, WHAT WERE THEY? **On query and subject 721 subject has two s at the very end but the query only has one**
25. ARE THERE ANY GAPS IN THE SEQUENCE ALIGNMENT? **Yes 1**
26. WHAT GENE ENCODES FOR THE POLYPEPTIDE YOU WERE ANALYZING? **Fibroblast growth factor receptor 3**
27. WHAT IS THE FUNCTION OF THIS PROTEIN **encodes a member of the fibroblast growth factor receptor family with its amino acid sequence being highly conserved between members among divergent species.**
28. WHAT HUMAN DISEASE IS CAUSED BY A MUTATION IN THIS GENE? **craniosynostosis and multiple types of skeletal dysplasia** Again,
29. WHAT IS THE CONNECTION AMONG THE FOLLOWING: NIH, NLM, NCBI, and HHS? **These are all government institutes for health and biology. NCBI is part of NLM and a branch of NIH**
30. REFLECT ON ONE THING THAT YOU LEARNED FROM DOING THIS ASSIGNMENT. **I already knew that more than one gene and mutation of the genes can cause the same disease, but doing the questions where the different genes cause ALS really put that into perspective for me. Not only that but I also learned the similarities between the genes of primates and what gap sequence alignments are.**