Genetics

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Genome Assignment

1. Chromosome 20
2. Over 900 genes
3. Contains over 60 million base pairs, of which over 90% have been determined
4. Alagille syndrome
5. Provides instructions for making a protein called Jagged-1, which is involved in an important pathway by which cells can signal to each other
6. Homo sapiens CFTR promoter region (LOC111674463) on chromosome 7
7. This region represents the promoter and proximal regulatory regions of the cystic fibrosis transmembrane conductance regulator (CFTR) gene. It includes multiple DNase I hypersensitive sites (DHSs), and enhancer, silencer and transcriptional cis-regulatory elements.
8. CF transmembrane conductance regulator
9. The role of the CFTR protein is an ion channel transporting chloride and bicarbonate

AM;, B.C. (2019) Cystic fibrosis: Pathophysiology of lung disease, Seminars in respiratory and critical care medicine. Available at: https://pubmed.ncbi.nlm.nih.gov/31659725/ (Accessed: 21 June 2023).

1. Cystic Fibrosis
2. Chromosome 7
3. Hylobates lar, Nomascus leucogenys, Pan troglodytes, Gorilla gorilla, Pongo abelii
4. No because of our related ancestry between species
5. Mus musculus TAR DNA binding protein (Tardbp), transcript variant 1, mRNA.
6. Homo sapiens ubiquilin 2 (UBQLN2), mRNA
7. Homo sapiens fusion (involved in t(12;16) in malignant liposarcoma) (FUS), transcript variant 2, mRNA
8. Homo sapiens superoxide dismutase-1 (SOD-1) gene, exon 1 and partial cds
9. 14- parkinsons, 15- ALS and dementia, 16- myxoid liposarcoma (MRCLS) , acute myeloid leukemia (AML), angiomatoid fibrous histiocytoma (AFH), amyotrophic lateral sclerosis (ALS) type 6, 17- familial amyotrophic lateral sclerosis (familial ALS)
10. GenBank is a database of sequences that is open access. It is an annotated collection of nucleotide sequences and their protein translations that are available to the public. Its maintained by the NCBI.
11. National Institutes of Health- 27 different components called institutes and centers. Each component has a specific research agenda. All compmnents except for 3 get funding directly from congress and dicate their own budgets. The National library of medicine is a branch of the NIH. The National Center for Biotechnology is a branch of the National Library of Medicine. GenBank is a part of NCBI, lastly BLAST is also hosted by NCBI.
12. This assignment really opened my eyes to how much open source data is available in the genetics field. I had no idea that previously that you could have access to that level of detailed information on genetics work. I’m extremely happy that this level of collaboration exists within the scientific community and hope to see the level of organization across different discipline sin the future.

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