

# Clinical Genomics Research Assignment: Exploring Genetic Variation in a Disease

## Background:

You are a junior researcher in a clinical genomics lab, and your team is studying the genetic basis of a specific disease. The disease has a known genetic component, and you have been tasked with utilizing genomics tools to analyze genetic variations associated with the disease.

## Assignment Task:

- 1. Select a Disease:** Choose a specific disease with a known genetic basis (e.g., cystic fibrosis, sickle cell anemia, familial hypercholesterolemia).
- 2. Identify Relevant Genes:** Research and identify the key genes that are associated with the chosen disease. Provide a brief overview of the functions of these genes and their relevance to the disease.
- 3. Genomic Data Retrieval:** Using publicly available databases (e.g., NCBI Gene, ClinVar), retrieve genetic information related to the identified genes for individuals affected by the disease and healthy controls. Compile a dataset of genetic variations (mutations, single nucleotide polymorphisms, insertions, deletions) associated with the disease.
- 4. Genetic Variation Analysis:** Utilize genomics tools (such as BLAST, UCSC Genome Browser, or Ensembl) to analyze the genetic variations. Compare the genetic sequences of affected individuals and healthy controls. Identify any common or unique variations in the disease group.
- 5. Functional Annotation:** For the identified genetic variations, perform basic functional annotation using tools like Variant Effect Predictor (VEP) or SnpEff. Determine the potential functional consequences (e.g., missense, nonsense, frameshift) of the variations.
- 6. Variant Frequency and Population Analysis:** Investigate the frequency of the identified genetic variations in different populations using tools like ExAC, gnomAD, or 1000 Genomes Project. Discuss any potential population-specific trends in genetic variation associated with the disease.
- 7. Data Interpretation:** Interpret your findings by discussing the potential impact of the identified genetic variations on protein structure and function. Relate these findings to the clinical manifestations of the disease.

**8. Research Report:** Compile your findings into a research report. Your report should include an introduction to the disease, details about the selected genes, methodology used for data retrieval and analysis, results of genetic variation analysis, functional annotation, variant frequency analysis, and data interpretation. Report should be maximum 6 pages, not including tables, figures and references. Arial, 12pt font, double-spaced, with 2cm margins.

**9. Code:** Save a copy of the commands you ran to conduct the analysis with comments and submit it as part of your Supplementary Materials.

**10. References:** Provide a list of references for the genetic information, tools, and databases you used in your research. References should be Nature style, with appropriate in-text citations and bibliography.

### **Assessment Criteria:**

Your assignment will be evaluated based on the following criteria:

**1. Accuracy and Relevance:** The chosen disease and genes are accurately identified and relevant to clinical genomics.

**2. Data Retrieval and Analysis:** Genetic variations are accurately retrieved and analyzed using beginner-level genomics tools.

**3. Functional Annotation:** Genetic variations are functionally annotated, demonstrating an understanding of their potential consequences.

**4. Population Analysis:** Analysis of variant frequency in different populations is conducted and interpreted.

**5. Data Interpretation:** Findings are effectively interpreted and related to the disease's clinical manifestations.

**6. Research Report:** The research report is well-organized, provides clear explanations, and includes all required sections.

**7. References:** Accurate and appropriate references are provided for sources used in the research.

### **Submission:**

Submit your research report along with any supplementary files (e.g., figures, tables) as a single file through Blackboard.