WORKSHOP ON BIOINFORMATICS AND GENOMICS FOR CLINICAL AND PHARMACEUTICAL APPLICATIONS

Annex of Pasteur Institute of Algeria in Sidi Fredj (Algiers)
From April 13 to 15, 2025

Dr. Radja Badji – Qatar Precision Health Institute
Dr. Chadi Saad – Qatar Precision Health Institute
Dr. Rania Abdelatif – Qatar Precision Health Institute

Workshop Description :

This workshop provides a comprehensive introduction to the interdisciplinary field of bioinformatics with a specific focus on its applications in clinical genomics and pharmacogenomics. Attendees will gain a foundational understanding of the principles and tools used to analyze large-scale genomic data, exploring both theoretical and practical aspects. The workshop covers the use of computational techniques for genomic sequence analysis, gene prediction, variant calling, and data visualization. Topics will include DNA sequence alignment, gene annotation, genomic data integration, and the application of machine learning in genomics. Practical exercises will use popular bioinformatics tools and programming languages such as Python, R, and specialized bioinformatics software to analyze real-world genomic datasets, including high-throughput sequencing technologies like next-generation sequencing (NGS).

Key Learning Outcomes :

- Provide participants with an overview of bioinformatics applications in genomics
- Develop proficiency in key bioinformatics tools and algorithms for analyzing genomic data, including sequence alignment, annotation, and variant detection.
- Learn how to analyze large-scale genomic datasets, including whole-genome sequencing (WGS)
- Gain hands-on experience with bioinformatics workflows, including data preprocessing, quality control, and visualization of results.
- Apply computational methods to solve real-world genomics problems and interpret genomic findings in biological and medical contexts.

Prerequisites :

Basic understanding of molecular biology and genetics.

Target Audience :

Researchers, clinicians, and students interested in genomics and bioinformatics.

Agenda: 3-Day Intensive Schedule

Day 1: Introduction to Genomics and Bioinformatics Tools

Dr. Chadi Saad

- **Morning :**
    - Introduction to sequencing technologies and data formats
    - Basics of sequence alignment and assembly
    - Introduction to variant calling
    - Phasing and imputation
    - Variant annotation and filtration

== BREAK==

    - Hands-on with bioinformatics tools and resources
    - Quality control of sequencing data (research vs clinical)
    - Sequence alignment
    - Visualization
    - Variant annotation

- **Afternoon :**
    - Introduction to bioinformatics development framework
    - Containerization techniques
    - Infrastructure
    - Optional: Hands-on

Day 2: Pharmacogenomics

Dr. Rania Abdelatif

- **Morning :** General Principles and Assessment of Knowledge Bases
    - Overview of pharmacogenomic concepts and their role in personalized medicine.
    - Key pharmacogenomic genes and variants (e.g., CYP2D6, CYP2C19, VKORC1).
    - Demonstration of pharmacogenomic databases (e.g., PharmGKB, CPIC, PharmVar).
    - Practical session :
        - How to query PharmGKB and CPIC for drug-gene interactions.
        - Case studies using CPIC guidelines for specific drugs like Clopidogrel or Warfarin.
        - Add hands-on activity if an interactive tool is available.
- **Afternoon :** Pharmacogenomics in Practice and Clinical Trial Development
    - Designing pharmacogenomic clinical trials :
        - Adaptive trials and patient stratification.
        - Selecting pharmacogenomic endpoints.
        - Regulatory and ethical considerations in pharmacogenomics research.
    - Practical session :
        - Decoding pharmacogenomic reports

Day 3: Genomic Data Analysis and Interpretation

Dr. Radja Badji

- **Morning :** Variant Interpretation Tools and Guidelines
    - Detailed introduction to databases for variant interpretation :
        - ClinVar: Searching for variant-disease relationships.
        - GnomAD: Exploring population frequencies.
        - HGMD: Accessing curated pathogenic variants.

- Overview of ACMG/AMP guidelines for variant classification :
  - Pathogenicity criteria and scoring system.
  - Examples of single nucleotide variant (SNV) classifications.

- **Afternoon :**
  - Interactive activity :
    - Classify SNVs using ACMG/AMP guidelines.
  - Best practices for genomic test ordering and reporting
  - Practical session :
    - Decoding clinical reports.

Requirements :

- Laptop with internet access
- Create an account in Galaxy and download testing data before the session