

NST Part II BBS Bioinformatics 2017-18

Course overview

The NST Part II BBS Bioinformatics will provide an introduction to the field of bioinformatics, focusing on bioinformatics applications related to the study of complex disease genetics and the recent advances made in this field since the introduction of next-generation sequencing (NGS) technologies.

We will first introduce fundamental concepts in human genetics and bioinformatics and then how NGS technologies can be applied to the study of human population genetics, genomics and its clinical applications. Fundamental statistical concepts that are crucial for designing a population study and are required to carry out statistical analysis of genomic data will be covered.

Then we will focus on functional analysis at the genomic level. Strategies for the identification of genomic variants using NGS will be explored, providing an introduction to the basic workflows for variant identification. Emphasis will be put on variants' annotation to infer a variant's biological relevance and consequently its potential diagnostic and therapeutic value. The challenges associated with the analysis and interpretation of genomic variants will be discussed. We will also introduce relevant public databases and the outcomes of large sequencing projects, which have provided new insights into the landscape of functional variation and genetic association.

Students will also learn about bioinformatics methods for RNA sequencing as well as network analysis and how the latter is used to acquire a functional understanding of the deregulation of signalling networks in diseases. In addition, drug developments based on the knowledge acquired through genomics approaches will be discussed as well as fundamental principles of image analysis, computational neuroscience and modelling of biological systems.

The course will consist of 15 lectures and 8 computer-based practical sessions. During the practical sessions, students will use the Unix command-line environment and the R project for statistical computing.

After attending this module, students will not be independent in the analysis of genomic data but will have acquired the critical thinking needed to understand what the statistical analysis of genomic data entails, what are the strengths and weaknesses of different analysis strategies, and will be equipped with a basic set of bioinformatics skills that will enable them to explore and interpret the genomic data available in the public domain.