Project 1: Epigenomics

Introduction

Genetic epidemiology accommodates different viewpoints to look at "disease". Unraveling important functional determinants to or causal factors for complex diseases requires a systems biology view, combining evidences from different data sources, involving the genome, the transcriptome, and epigenome, amongst others. The latter has been given increasing attention over the last 5 years. This is not surprising, given the trends to generate tons of data with reducing costs and trends to move towards a personalized medicine.

In this project (Project 1), you will investigate what the epigenome is and how it may change "traditional" human complex disease analysis. You may use any resource you can find to build up your story. As an aid, I have provided some guiding questions. As the course progresses, it will become clear how "epigenome analysis" relates to "family-based genomic analyses", "genome-wide association analysis, and "interaction analysis". Subsequent Projects will elaborate on "genetic association" studies and "gene-gene and gene-environment interaction" analyses, in particular. Hence, Projects 1-3 combined will provide a reflection of key learning outcomes for the "Genetic Epidemiology" course as a whole.

Specific questions

- What is the relationship between genomics, statistical genetics, genetic epidemiology, and epidemiology?
- Is your DNA your destiny? Why? Why not?
- How does epigenetics fit into the "genetic epidemiology" picture? Has our understanding about epigenetics and its potential contribution to human disease management evolved over time?
- Do several types of epigenetic mechanisms exist?
- Can you give some examples of environmental epigenetics? What is the relative contribution of environment? (e.g., prenatal stress, nutrition, sun exposure, …)
- What is the relation between epigenetic mechanisms and "time" (i.e., does it persist over time, can it change over time, may it be age-related)?
- Can epigenetic mechanisms link to complex diseases or affect phenotypes? Give some examples.
- Study design (involving subjects and measurements) and research question largely determine the selection of analysis technique. What are some of the study designs currently being adopted when studying the epigenome and its relation to disease phenotypes? Pay extra attention to experimental versus non-experimental designs, sample sizes, power, epigenetic measurement (scales) [beta-values, M-values], relevant measures of association.

Guidelines on Project Reports

- Restrict the written group reports for Projects 1-2 to maximally the equivalent of five single-spaced typed pages of text, excluding figures, tables and bibliography. The report ideally contains an introduction, a description of what the project aims to achieve and it fits into the broader scientific context or the genetic epidemiology course, as well as a conclusions section. If citations are made to other papers, there should also be a bibliography. The body of the report provides answers to guiding questions provided in the project assignment.
  The final due date of the written reports: discussed in class
- Projects 1-2 will be presented in the department/class: Tue 26/3 (30' per assignment, including discussion). The presentation slides will be made available to course participants via the course website.
- The individual-based report for Project 3 can be organized in the same way as the reports for Projects 1-2. It will be presented on an individual basis on the day of the exam and will serve as a starting point for the oral exam. The best way to prepare for this exam is to combine Projects 1-3 and to condense it into a single story, highlighting how one topic is related to the other. More information about how to prepare for the exam will be given as the course progresses.
- The nature of Projects 1-3 facilitates using these projects as a tool to evaluate the described learning outcomes of the course.