

Genetic Epidemiology

*May 9-13 * Alda Sousa (ICBAS/IBMC)

Discipline: Genetic Epidemiology

Objectives: The students should to become acquainted with some basic aspects of human genetics and with some of the different strategies in genetic epidemiology (that will be dealt with in depth in the advanced course)

Program:

1. Genetic Epidemiology and its relationship with other disciplines (Epidemiology, Genetic of Populations, Formal Genetics, Statistical Genetics and Molecular Genetics).
2. Mendelian modes of inheritance and risk calculation in simple situations. Application of the H & W equilibrium to estimate allelic and genotypic frequencies in case of complete dominance.
3. Studying the familial aggregation of a disease or trait and assessing its possible genetic component: population studies (case-control or “follow-up”) and in families (in twins, in relatives with several kinship degrees)
4. Some epidemiological concepts; measuring a disease’s frequency; relative risk and odds-ratio
5. Linkage analysis: basic concepts. The *lodscores* method and its application to simple situations in mendelian diseases
6. Introduction to association studies

Bibliography:

- T. Strachan and A.P. Read. *Human Molecular Genetics 2* (2nd ed.). BIOS Sci. Publ., Oxford, 1999
- M. Khoury, T. Beatty and B. Cohen. *Fundamentals of Genetic Epidemiology*. Oxford University Press, 1993
- M. Khoury, J. Little and W. Burke. *Human Genome Epidemiology. A Scientific Foundation for Using Genetic Information to Improve Health and Prevent Disease*. Oxford University Press, 2003
- R. A. King, J.I. Rotter, A. G. Motulsky. *The Genetic Basis of Common Diseases*. Oxford University Press, 2002
- J. H. Haines and M.A. Pericack-Vance (editors). *Approaches to Gene Mapping in Complex Human Diseases*. Wiley-Liss, 1998
- I.H. Pawlowitzki, J.H. Edwards and E.A. Thompson (editors). *Genetic Mapping of Disease Genes*. Academic Press, 1997
- J. Ott. *Analysis of Human Genetics Linkage*. Johns Hopkins University Press, Baltimore, 1991
- J. D. Terwilliger and J. Ott. *Handbook of Human Genetic Linkage*. Johns Hopkins University Press, Baltimore,

Teaching and Learning Methods: Lectures, exercises, paper discussion

Evaluation: At the end of advanced course students will be presenting and discussing a design study

Site: IBMC

Schedule: 10h-13h