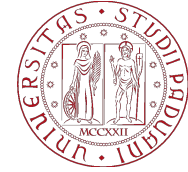




VENETO
INSTITUTE OF
MOLECULAR
MEDICINE

1222 · 2022
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UNIVERSITÀ
DEGLI STUDI
DI PADOVA

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The course of Human Genetics at BHEH

Prerequisites

Knowledge deriving from the topics of the first two years of the Degree, in particular: biology, biostatistics, molecular biology and genetics, chemistry and biochemistry

Objectives

The course provides students with a basic knowledge in human genetics and genetic disease mechanisms, as well as critical assessment and presentation of genetic research in both oral and written form.

On completion of the course, the student should be able to:

Regarding knowledge and understanding

- describe and explain the human genome organization, regulation and expression.
- explain how genetic variation occurs and its impact on health.
- determine different modes of inheritance of genes and traits
- identify suitable approaches to disease gene identification and disease mechanisms in the research field of human genetics
- discuss pros and cons of large sequencing projects and precision medicine

Regarding skills and ability

- critically review relevant scientific literature and discuss the results and conclusions
- search for, collect, evaluate, interpret and discuss (in writing and orally) research data in relation to topics within the course

Regarding judgement and approach

- reflect on ethical aspects of research involving human and animal material
- take responsibility for his/her own learning

The course plan

- the fundamentals of DNA, chromosomes, the cell cycle, human genome organization, and gene expression (including epigenetic regulation).
- basics of three core molecular genetic approaches used to manipulate DNA: DNA amplification (by DNA cloning or PCR), nucleic acid hybridization, and DNA sequencing,
- general principles of genetic variation, including DNA repair mechanisms.
- how genes are transmitted in families and how this is related to allele frequencies in populations.
- how genes are regulated by a wide range of proteins and noncoding RNA regulators,
- clinical applications, including: how chromosome abnormalities arise and their consequences, and how mutations and large-scale DNA changes can directly cause disease, how genes underlying single-gene disorders are identified, and how genetic variants that confer susceptibility to complex diseases are identified.
- How genetic variants, epigenetic dysregulation, and environmental factors all make important contributions to complex diseases.
- approaches for treating genetic disorders: From one-size-fits-all approach to disease treatment toward personalized and precision medicine. Ethical implications.
- Cancer genetics: cancer as a combination of abnormal genetic variants and epigenetic dysregulation.

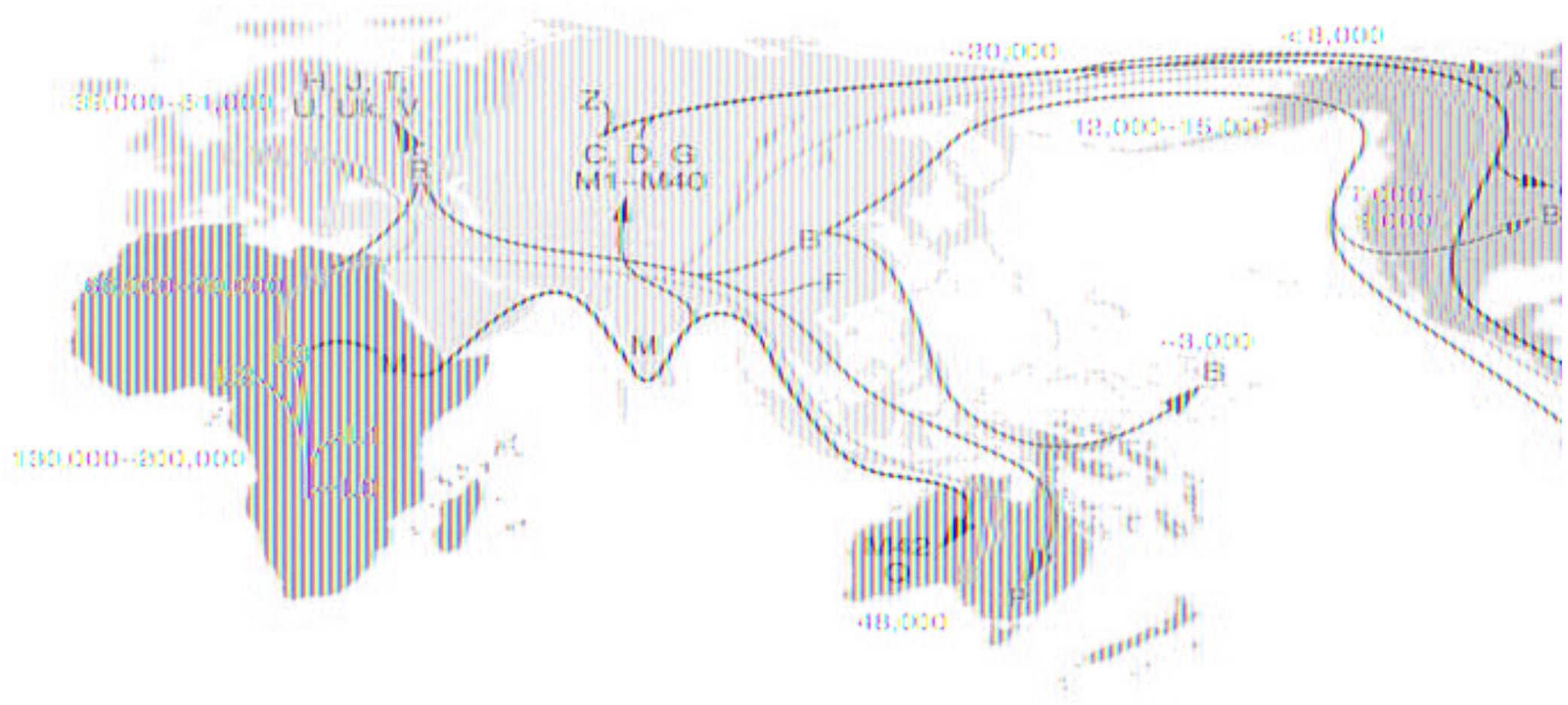
Teaching methods

During this course, the learning will be an active process with integrated feedback. The course will include elements of "flipped classroom" learning in which course material are provided before teacher-moderated discussions with the class. Particular emphasis is placed on peer-learning and self-studies in groups and at the individual level.

Teaching material

- Strachan and Read, Human Molecular Genetics 5^o edition, CRC Press Taylor & Francis Group
- Strachan, Goodship and Chinnery, Genes and Genomes, Garland Science, Taylor & Francis Group, LLC
- Snustad and Simmons, Principles of Genetics, *John Wiley & Sons, Inc.*
- Articles and reviews provided during the course

mtDNA variation in human evolution



Examination

- Written exam with open questions

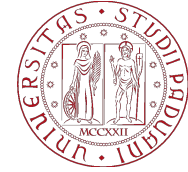
Lab activities (Prof Chemello, UNIBO) will include:

- Genotyping (mouse) families
- Analysis of human pedigrees
- Analysis of mtDNA variants, quantity and quality
- Elements of NGS by long reads
- Elements of analysis of DNA variants using bioinformatic tools



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THANK YOU!

Human Genetics at BHEH