UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Genetica Evolutiva

2627-2-H4103D158-H4103D15802

Aims

The course provides the theoretical and methodological foundations of human population genetics, molecular evolution, and evolutionary medicine. Upon completion of the course, students will be able to:
• understand the evolutionary processes that have shaped human genetic diversity;
• interpret the demographic history of human populations and the phenomena of migration and admixture;
• recognize the main mechanisms of natural selection and the methods used to identify them in genomic data;
• apply concepts from molecular evolution and comparative genomics to the interpretation of genetic variants and human diseases;
• understand the role of genetic conflicts and transposable elements in human evolution and disease;
• analyze the evolutionary processes of viruses and their biomedical relevance, with particular emphasis on host adaptation and infectious diseases.

Contents

1. Population genetics— Hardy–Weinberg equilibrium, inbreeding, human demographic history (Out of Africa and subsequent migrations), admixture with archaic hominins, and genetic diversity in human populations.
2. Natural selection — Types of selection (purifying, balancing, positive, sweeps, polygenic selection), molecular methods for detecting selection, adaptive examples (lactase persistence, malaria resistance) and their medical relevance.
3. Molecular evolution — Neutral theory, comparative genomics, conservation indices (PhyloP, GERP) used in clinical genetics, and dN/dS tests for detecting positive selection.
4. Genetic conflicts — Meiotic drive, imprinting, host–pathogen interactions, sexual selection, and “selfish spermatogonial selection.”
5. Transposable elements — Origin, pathogenic roles, domestication (placentation, immunity), and contribution to genomic plasticity. Germline silencing mechanisms and relevance to fertility.
6. Viral evolution — Phylogenomics, phylogeography, zoonoses, and host adaptation. History and genetic characteristics of Jenner’s smallpox vaccine (1796).

Detailed program

1. Human population genetics. Basic concepts of population genetics (gene pool, allele and genotype frequencies, Hardy–Weinberg equilibrium, genetic drift, gene flow, natural selection). Non-random mating, with attention to phenotypic traits (BMI) and pathological traits (neuropsychiatric disorders). Human demographic history (African origin, Out of Africa migration, bottlenecks, population expansions, founder effects). Admixture with archaic hominins and adaptive introgression, Yamnaya migration. Genetic variability in human populations. Databases of genetic variation (1000 Genomes, gnomAD).
2. Human population genetics. Types of natural selection: purifying, balancing, positive, hard sweep, soft sweep, polygenic adaptation. Molecular methods to detect selection (EHH, iHS, and reconstruction of haplotype history using haplotype networks). Examples of adaptation to environmental factors and relevance to evolutionary medicine (e.g., lactose tolerance, resistance to malaria and other pathogens, high-altitude adaptation, kidney disease and trypanosome resistance).
3. Molecular evolution. Basic mechanisms and comparative genomics (orthologs, paralogs, homologs, multiple sequence alignment). Conservation indices (PhyloP, GERP) and their use in medical genetics. Positive selection, methods of detection (dN/dS), biomedical relevance (Red Queen dynamics). Loss of function as adaptation (CMAH and absence of Neu5Gc in the human species).
4. Genetic conflicts. Intragenomic conflicts (meiotic drive, transposable elements, genomic imprinting) and intergenomic conflicts (host–pathogen interactions). Their biomedical relevance and impact on fertility (Robertsonian translocations and centromere drive). Sexual selection in humans. “Selfish spermatogonial selection” and relevance for the “paternal age effect” (achondroplasia, Apert, Costello, Noonan syndromes).
5. Transposable elements in the human genome. Evolutionary origin and molecular features, pathogenic roles (insertional mutations, chromosomal rearrangements, genomic instability). Domestication: syncytins (envelope proteins of endogenous retroviruses domesticated for formation of the placental syncytiotrophoblast), RAG1/RAG2 (V(D)J recombination machinery derived from a Transib transposon family), and other examples. Contribution to genomic plasticity (regulatory innovation, new enhancers, new promoters). Clinical relevance (cancer and neural plasticity). Their repressors and relevance for fertility (piRNAs and PIWI proteins).
6. Viral evolution: phylogenomics and phylogeography (epidemic tracking, variant identification, geographic spread, migratory pathways). Population structure, host switching (zoonoses), host adaptation. History and genetic characteristics of the first vaccine (against smallpox, Jenner 1796).

Prerequisites

As specified in the Degree Programme Regulations.

Teaching form

Face-to-face lectures, complemented by class discussions.
All teaching activities are delivered on campus in lecture format.
The language of instruction is Italian.

Textbook and teaching resource

Slides and scientific journal articles

Semester

Second year, first semester

Assessment method

Described in the course syllabus.

Office hours

By appointment, writing to manuela.sironi@unimib.it

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING