Human Evolution and Health (20413)

Qualification/course: Bachelor's Degree in Medicine
Year: 2
Term: 1
Number of ECTS credits: 6 credits
Number of study hours: 100 hours
Course languages: Catalan
Teaching staff: Elena Bosch and David Comas

1. Presentation of the course

The goal of the subject is to show the general evolutionary mechanisms that have acted and are acting within our species and to understand that the current diversity (morphological, genetic) of humanity can be explained by evolutionary processes. Human evolutionary strategy will be demonstrated and the selection factors that have made us, taking disease into account within the adaptive process. There will be a focus on the concept of gene-environment interaction, showing that many human characteristics have a biological, and therefore evolutionary, explanation. All the evolutionary concepts will be applied to understanding health and disease, giving a complementary scientific approach to the mechanistic explanations that predominate in the medical sciences. We will see how human adaptations are related to the evolution of pathogens, their virulence and resistance to antibiotics. The course will explain how human adaptations to ancestral environments affect current humans via different diets, life expectancy, exercise and hygiene. In general, the trade-offs, or biological compensation mechanisms, throughout human evolution will be revealed.

2. Competences to be achieved

2.1. General competences:

2.1.1. Instrumental competences

- Observation of phenomena from biological data.

3. Content

LECTURES

1. Human evolution: definitions and problems

Evolution in a historical context. Darwin's revolution. Evidence for evolution. Models and mechanisms of evolution.

2. The human genome and its variation

Our genome. Types of polymorphisms and their detection. Haplotypes and HapMap. Linkage disequilibrium.

3. Human genetic diversity

Measuring genetic diversity: heterozygosity, number of segregating sites, Watterson's estimator and number of pairwise differences. Populational differentiation: F_{ST}. Mismatch distributions. Genealogy of a gene: coalescence.

4. Genetics of human populations and evolutionary processes

Synthetic theory of evolution. Hardy-Weinberg equilibrium. Evolutionary factors: mutation, migration, genetic drift and selection.

5. The forces of evolution I.

The forces of evolution: selection, mutation, migration and drift. Selection types and models. Selection against the recessive homozygote. Selection in favor of the heterozygote. Codominance and gene dose.

6. Detection of selection in humans

Time scales for the signatures of selection. Detection of selection between species (interspecific level): Ka/Ks ratio. Detection of selection within species variation (intraspecific level): diversity and frequency spectra, population differences and F_{ST},, linkage disequilibrium and haplotype length.

7. The forces of evolution II.

Mutation and migration: changes in allele frequencies. Genetic drift, bottlenecks, and founder effects. The neo-Darwinian view versus the neutral theory of evolution. Gene substitution: probability of fixation, fixation time and rate of gene substitution .

8. Molecular phylogeny.

Terminology and types of data. Methods of phylogenetic tree reconstruction . Distance-based methods: UPGMA and NJ. Maximum parsimony methods. Maximum likelihood methods. The root of a phylogenetic tree. Robustness of a tree: bootstrapping and consensus tree. Applications of phylogenies to the study of humans.

9. Substitution rates and patterns.

Number of substitutions between two DNA sequences. Rates of nucleotide substitution. The molecular evolutionary clock. Amino acid substitution patterns. Non-random usage of synonymous codons.

10. Molecular and species phylogenies

Phylogenetic position of humans in nature. Classical classification and the molecular approach. Phylogeography of uniparental human genomes: diversity of mitochondrial DNA and the Y-chromosome. Other molecular markers.

11. Origin of the human line

Early hominids. Evolutionary adaptations: bipedalism, encephalization. The appearance of the Homo genus.

12. Origin of the human genus and species

The appearance of our species: paleontological and genetic data. Neanderthals and anatomically modern humans. Ancient DNA evidence. The appearance of our species: paleontological and genetic data.

13. Geographical distribution of human diversity

Biological diversity (morphological and genetic) of humanity: from sub-Saharan Africa to colonisation of the Americas and the Pacific. Validity of concepts of ethnicity and race.

14. Human adaptations

Introduction to human adaptations. Classification. Climatic adaptations: temperature, humidity, altitude, solar radiation. Human pigmentation as a paradigm.

15. Evolution of the human life cycle

Comparative models: primates, hunter-gatherers. Human reproduction, mother-child conflict, long-term changes, theories on aging and the menopause.

16. Natural history of human disease

Population differences of disease. Mendelian disease and complex disease.

17. Evolution of infectious diseases

The malaria paradigm. Other infectious diseases. Vaccines and resistance to antibiotics. Phylogeny of pathogens.

18. Changes in diet and implications in disease

Cultural nutritional evolution: the paradigm of lactose intolerance. Diabetes and obesity in an evolutionary context. Theory of energy saving genetic variations.

19. Evolution of chronic diseases.

Evolutionary implications in the development of cardiovascular diseases. Cancer. Behavioural disorders, schizophrenia, addictive behaviours in an evolutionary context.

20. The biological future of humanity

Impact of migrations and cultural practices on human genetic diversity. Dysgenics and eugenics. Ethical, healthcare and adaptive implications.

OTHER ACTIVITIES

1. Seminars (I)

Discussion of articles on human adaptations and evolution.

2. Seminars (II)

Discussion of articles on the evolution of disease.

3. Study of diversity in primates.

Visit to the Parc Zoològic in Barcelona to observe diversity in the order of primates and compare this to the human species.

4. Human geographical diversity.

Video screening "The Journey of Man" National Geographic, to discuss biological and demographic differences on a continental scale

PRACTICALS

1. Analysis of human intraspecific variation (I)

Analysis strategies to confront specific problems: interpretations in demographic and genomic terms. Calculation of measures of diversity: heterozygosity, number of segregating sites, Watterson's estimator. Use of the DNASP program. (2 hours). Computer room

2. Population genetics problems (I) (2 hours)

3. Population genetics problems (II) (2 hours)

4. Analysis of human intraspecific variation (II).

Dynamics of human populations using mitochondrial DNA and SNP data from HapMap. Calculation of the number of pairwise differences, mismatch distributions, F_{ST}

and difference between populations. Use of the DNASP program. Calculation (2 hours) *Computer room*

5. Analysis of human intraspecific variation (III).

Methods of phylogenetic tree reconstruction. Hands-on distance-based methods: UPGMA and NJ. Hands-on maximum parsimony methods. Applications of phylogenies to the study of humans (2 hours). Computer room.

6. Databases and alignments

Introduction to public databases of DNA sequences and proteins. Extraction of information. Structure of the data. Formats. Sequence alignment: CLUSTALW package and Revtrans (2 hours). Computer room

7. Analysis of interspecific variation

Computer resources for studying interspecific variation. Calculation of genetic distances and construction of trees: use of the MEGA package (2 hours). Computer room

8. Approximation to diversity in the human line and comparison with other primates

Analysis of specific, ontogenetic and sexual variation. Human adaptations (2 hours).

4. Assessment

The assessment will use five tools:

1. Multiple choice questions (PEM) in the general end-of-term test. Emphasis is placed on factual knowledge, and this part will contribute 30% to the final mark.

2. Short essay questions and solving problems on the practicals. Reasoning skills and use of acquired knowledge will be assessed. This part will contribute 40% to the final mark.

3. Continuous assessment I. Presentation and discussion of articles will be evaluated during the seminars, which will contribute 15% to the final mark.

4. Continuous assessment II. Delivery of exercises solved during practical sessions 1, 4, 5, and 7. Presentation and discussion of articles will be evaluated during the seminars, which will contribute 15% to the final mark.

5. Formative assessment. Halfway through the term there will be an exam containing MCQ and essay questions. If the student passes, an additional mark will be added to the final mark, which will increase linearly from 0.25 (for a mark of 5 in the formative assessment) to 0.5 points (for a mark of 10).

5. Bibliography and teaching resources

5.1. Basic bibliography

~~BARTON, H. B.; BRIGGS, DEG; EISEN, J. A.; GOLDSTEIN, D. B.; PATEL, N. H. *Evolution*. Nova York: Cold Spring Harbor Laboratory Press, 2007.~~

BOYD, R.; SILK, J. B. *How humans evolved*. WW Norton & Company Inc. Fifth Edition, 2009. [N'hi ha una traducció al castellà de la tercera edició: Cómo evolucionaron los humanos. Editorial Ariel, 2004].

GRAUR, L. I. *Fundamentals of Molecular Evolution.* Sinauer, 2000.

GRIFFITHS AJF, WESSLER SR, LEWONTIN RC, CARROLL. Genética. Mc Graw-Hill, 2008.

JOBLING, M. A.; HURLES, M. E.; TYLER-SMITH, C. *Human Evolutionary Genetics*. Garland Science, 2004.

~~RIDLEY, M. Evolution. Blackwell Science, 1997.~~

~~STEARNS, S. C.; KOELLA, J. C. *Evolution in Health and Disease*. Oxford University Press, 2008.~~

~~TREVATHAN, W. R.; SMITH, E. O.; McKENNA, J. J. *Evolutionary Medicine and Health. New perspectives*. Oxford University Press, 2008.~~

5.2. Complementary bibliography

BARTON, H. B.; BRIGGS, DEG; EISEN, J. A.; GOLDSTEIN, D. B.; PATEL, N. H. *Evolution.* Nova York: Cold Spring Harbor Laboratory Press, 2007.

CAVALLI-SFORZA, L. L.; PIAZZA, A.; MENOZZI, P. *History and geography of human genes*. Princeton, Princeton University Press, 1994.

HARTL, D. L.; CLARK, A. G. *Principles of Population Genetics.* Fourth Edition. Sinauer Associates, Inc. Publishers, Sunderland, Massachusetts, 2007.

NESSE, R. M.; WILLIAMS, G. C. W*hy we get sick: the new science of Darwinian medicine*. Vintage Books, a division of Random House Inc, 1995.

RIDLEY, M. *La evolución y sus problemas*. Pirámide, 1987.

RIDLEY, M. Evolution. Blackwell Science, 1997.

STEARNS, S. C.; KOELLA, J. C. *Evolution in Health and Disease*. Oxford University Press, 2008.

TREVATHAN, W. R.; SMITH, E. O.; McKENNA, J. J. *Evolutionary Medicine and Health. New perspectives*. Oxford University Press, 2008.

5.3. Teaching resources

* Course page in the Moodle Aula Global, where the students will find introductions to each topic, graphical presentations and the complete texts for the seminars.

* External web pages:

http://www.prenhall.com/boaz

http://www.wwnorton.com/college/anthro

http://darwiniana.org/evolution.htm

6. Methodology

The teaching of the subject consists of lectures, other activities (seminars, video screenings, visits to the Zoo) and practicals. The seminars are given in small groups and consist of the discussion of recent articles on topics related to the subject. The articles are found in the Moodle Aula Global, and students have one week to prepare the topics, at the end of which they present and discuss the topic they have worked on to their colleagues, who will partially assess the work done. In the practicals the students will explore resources and analytical tools in anthropology and evolution, including guided solving of population genetics problems and applying the main methodological resources used for exploring human diversity.

7. Programme of activities

Week 1: Lectures 1 & 2.

Week 2: Lectures 3 & 4.

Week 3: Lectures 5 & 6. Practical 1.

Week 4: Lectures 7 & 8. Practical 2.

Week 5: Lectures 9 & 10. Practical 3. Formative assessment.

Week 6: Lectures 11 & 12. Practical 4. Seminar 1.

Week 7: Practical 5.

Week 8: Lectures 13 & 14. Practical 6. Seminar 2.

Week 9: Lectures 15 & 16. Practical 7. Visit to Zoo.

Week 10: Lectures 17 & 18. Practical 8.

Week 11: Lectures 19 & 20. Document-based discussion.

Week 12-13: Study

Week 14: PEM and essay test.