

HUMAN POPULATION GENETICS

Patterns and Processes



A complete advanced undergraduate/graduate course with:

- 18 online lectures by leading authorities
- Resources for workshops, tutorials, journal clubs, projects and seminars
- Suggested exam questions and model answers
- Multiple choice questions and answers
- Recommended reading: original papers and review articles



View the content of the course on our
website: hstalks.com/HumanPopulationGenetics



View our in-depth HSTalks:
hstalks.com/CoursesBrochure

Course module with video lectures, material for tutorials (case studies, projects, workshops and recommended reading), multiple choice questions and suggested exam questions with model answers. A comprehensive course on a subject of major importance.

The material is especially designed to support research and teaching staff when presenting a comprehensive course at graduate or advanced undergraduate level with seminars, journal clubs, laboratory exercises, data workshops, online tests and end of course examinations.

The course is also suitable for continuing professional development/education programmes.

This brochure provides brief details of the complete module, including the lectures, lecturers and additional learning material.

Who is the course for?

The comprehensive material is especially suitable for teachers and researchers who wish to offer courses on specialist subjects to small groups of students (or even a single student) when it is not possible to justify the time and expense of preparing, internally, a course or there is not the range of expertise available locally to do so. All the lecturers are highly regarded experts in their fields and few institutions are likely to have a comprehensive group of faculty members with a similar range of experience and knowledge of the subject matter.

The course material is designed to be used by local faculty and staff acting as course directors, tutors and mentors.

The material is suitable for flipped classroom, blended, team and distance learning courses.

New courses are time consuming and expensive to create. These modules cut both the cost and the time, enabling a wider range of options to be offered on specialist topics. Graduate students can take the courses, mentored by their supervisors, while pursuing their research.

Ideal for Virtual Learning Environments (VLE)

All course material, including the additional learning material, is arranged in a standard format that allows easy embedding into virtual learning environments such as Moodle, Blackboard or your institute's own system.

Supporting learning and teaching goals

In an age when faculty and staff face ever greater demands on budgets and time, these lectures and additional learning material will be of great help when preparing and delivering graduate and advanced undergraduate courses.

Course Summary

Patterns of genetic variation in the human population represent the outcome of a complex process of genetic descent that has taken place over the course of human evolutionary history. These patterns in turn provide a rich source of information useful for inference of the events and phenomena of human evolution, and for studying evolutionary aspects of heritable disease and other human traits.

This course examines the subject of human population genetics, with a focus both on phenomena of population genetics as examined from the standpoint of human populations, and on the interpretation of human genetic variation and its history.

The first section of the course provides an overview of core concepts in population genetics, as reflected in the study of human genetic variation. The course next proceeds to examine a number of case studies of human population genetics, studying different geographic regions and different stages of human evolution. Next, the course covers a number of cultural, demographic, genomic, and selective phenomena in human population genetics, including a number of topics of current interest. The last section of the course focuses on the relationship of human genetic variation and human evolutionary history to the understanding of the genetic underpinnings of human phenotypes, including disease phenotypes.

Each lecture will be presented by a renowned scientist, providing an account of principles, methods, and results in the field, and leading up to current knowledge. Lecturers will interpret the topics of the suggested lectures in light of their own discoveries, to incorporate both basic principles and recent work from their own laboratories and those of other researchers. Modern developments in genome sequencing and in the theory and statistics of genetic variation have made possible an increasingly deep understanding of human evolutionary history, and this course aims to share these exciting developments.



Editor: Prof. Noah Rosenberg
Stanford University, USA



Editor: Prof. Rasmus Nielsen
University of California, Berkeley, USA

**The course
module is
designed
for:**

- Researchers and graduate students in the fields of Life Science, Human Genetics and Anthropology
- Advanced undergraduate students
- Policy makers and managers in public and private sectors
- Continuing Professional Education / Development

Course Lectures

Introduction to human population genetics

→ An overview of human migrations

Prof. Mark Jobling
University of Leicester, UK



Genetic drift in human evolution

Prof. Sohini Ramachandran
Brown University, USA



Human migration and population structure

Prof. John Novembre
University of Chicago, USA



Human population genetics across time and space

Human molecular evolution since
the human-chimpanzee divergence

Prof. Katherine Pollard
UC San Francisco, USA



Ancient DNA and human evolutionary inference

Prof. Mattias Jakobsson
Uppsala University, SE



The genetic history of Australia, Oceania,
and Southeast Asia

Prof. Mark Stoneking
Max Planck Institute for Evolutionary Anthropology, DE



Genetic variation in North America

Prof. Ripan Malhi
University of Illinois at Urbana-Champaign, USA



Patterns of genetic variation and admixture in Latin America

Dr. Andrés Moreno-Estrada
Stanford Center for Computational Evolutionary and Human Genomics, USA



*Click
the lecture title
to access*

18 specially recorded, animated lectures
by world leading authorities

Cultural, demographic, genomic, and selective phenomena in human
population genetics

> Local adaptations in humans

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the lecture title
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Human population growth and its impact on genetic variation

Dr. Anna Di Rienzo
University of Chicago, USA



Genetic and linguistic evolution and coevolution

Prof. Andrew Clark
Cornell University, USA



Cross-talk between cultural and genetic evolution in humans

Prof. Keith Hunley
University of New Mexico, USA



Human admixture

Prof. Evelyne Heyer
National Museum of Natural History, FR



Consanguinity and genomic sharing in human evolutionary inference

Dr. Paul Verdu
Centre national de la recherche scientifique, FR



Prof. Trevor Pemberton
University of Manitoba, CA



Human population genetics across time and space

Genetics and human skeletal variation

Prof. Timothy Weaver
University of California, USA



Genetic variation in gene regulation

Dr. Jonathan Pritchard
Stanford University, USA



Human pathogen-driven evolution

Dr. Matteo Fumagalli
University College London, UK



Biological and cultural influences on disease

Dr. Connie Mulligan
University of Florida, USA





Examples of Course Materials

For each lecture the course offers tutorials, workshops, recommended reading, multiple-choice questions, and suggested exam questions with model answers.



HST Moodle My Courses ▸ Human Population Genetics

Human molecular evolution since the human-chimpanzee divergence

Prof. Katherine Pollard - University of California, San Francisco, USA

Using the UC Santa Cruz Genome Browser (genome.ucsc.edu), navigate to the "Genomes" page and select the human genome hg19 assembly.


1. Use the "Tools" menu to select "In-Silico PCR". What genomic region (chromosome, start position, end position) would you clone if you used the following PCR primers?
Forward primer: GTGTGTGGCGCAGCCTTGGG
Reverse primer: TGTGCGTTGTCCAGGTTGCA
2. Click through to the cloned sequence in the browser viewing-window. Try zooming in and out, and turning on and off some of the tracks listed below the viewing-window. Using the "UCSC Genes" track, is the cloned sequence coding or non-coding?
3. Turn on and configure the "Conservation track" to show "multiz alignments", "basewise conservation (phyloP)", "conserved elements", and sequences of all species. Across what species - if any - is the cloned sequence evolutionarily conserved? What evidence is there for conservation?
4. Does the cloned sequence include human-chimp differences and did these mutations happen in humans or chimpanzees?




Exam Questions and Model Answers

Question 1

Not yet answered
Marked out of 1.00

 Flag question

 Edit question

What is the overall sequence identity between human and chimpanzee, and what factors do you need to consider when calculating this number?




Multiple-choice questions and answers

Question 4

Not yet answered
Marked out of 1.00

 Flag question

 Edit question

Multiple choice questions and answers:

Negative selection results in:

- ☐ a) Fewer substitutions than under the neutral model
- ☐ b) More substitutions than under the neutral model
- ☐ c) Different patterns of substitutions across species
- ☐ d) Different rates of substitutions across species



Recommended reading supporting each lecture: Original research papers and review articles

Recommended Review Articles

1. A. Siepel, G. Bejerano, J.S. Pedersen, A.S. Hinrichs, M. Hou, K. Rosenbloom, et al. (2005). Evolutionarily conserved elements in vertebrate, insect, worm, and yeast genomes, *Genome Research*, 15: 1034-1050.
2. K.S. Pollard, M.J. Hubisz, K.R. Rosenbloom, A. Siepel (2010). Detection of non-neutral substitution rates on mammalian phylogenies, *Genome Research*, 20: 110-121.
3. M.J. Hubisz, K.S. Pollard, A. Siepel (2011). PHAST and RPHAST: phylogenetic analysis with space/time models, *Briefings in Bioinformatics*, 12: 41-51.
4. M.J. Hubisz, K.S. Pollard (2014). Exploring the genesis and functions of Human Accelerated Regions sheds light on their role in human evolution, *Current Opinion in Genetics and Development*, 29: 15-21.



How to access the course

Extracts of lectures can be viewed at hstalks.com/biosci/. The full length lectures can be viewed by all members of universities, colleges and medical schools currently subscribing to The Biomedical & Life Sciences Collection. Institutions that do not subscribe to The Biomedical & Life Sciences Collection may take annual licenses at US \$2,000 covering an unlimited number of students.

Full supporting material: video lectures, material for tutorials (case studies, projects, workshops and recommended reading), multiple choice questions and suggested exam questions with model answers are provided to faculty members of subscribing institutions.

To subscribe, obtain additional information and/or the additional learning material contact Dr. Eyal Kalie at eyalk@hstalks.com.

Upload to your VLE

The complete course (lectures and additional learning material) can be loaded into Moodle, Blackboard and other virtual learning environments.



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HSTalks

hstalks.com

Email: sales@hstalks.com

Tel: +44 207 164 6721

Corporate Headquarters
Ruskin House, 40/41 Museum Street
London WC1A 1LT, United Kingdom
Company Registration Number: 04833828
(England and Wales)



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