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PRINCIPLES OF POPULATION GENETICS

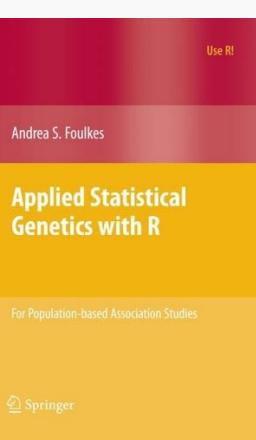
FOURTH EDITION

Daniel L. Hartl & Andrew G. Clark

PRINCIPLES OF POPULATION GENETICS

Second Edition

Daniel L. Hartl
Andrew G. Clark



Einführung in die Genetische Epidemiologie

H. Bockeböller
C. Fischer

Springer

ECOLOGY

Second Edition



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This course has been added to the BU HUB General Education Curriculum as CAS AN/BI 333. There are several HUB-related Learning Outcomes for this course, including: Scientific Inquiry II (SI2) Learning Outcome 1: Human genome data is a relatively new resource with its own challenges, both scientific and ethical. Students will apply population genetic and genomic principles to real human genomic data from the 1000 Genomes Project to assess the influences of population history and natural selection on human genomic variation. Methods will be learned as a class on an example gene of interest, followed by the independent application of these methods on a gene of each student's choosing. We will discuss the relative merits of each test, when they are best used and under what population conditions, and then use this information to assess the appropriateness of tests for student-led projects. Hypotheses and analyses in a peer-review format. Learning Outcome 2: While understanding the scientific merits of each analysis is critical, equally important is understanding the ethical merits of how the data was collected and results disseminated. Through a series of in-lecture discussions, we will interrogate the ethics of genomic data collection from a number of perspectives, including the use of genomic data to assess belonging and population history in indigenous North America (and in contemporary US politics), the proliferation of online personal genomics testing and health reports, and the ways in which genomic sampling continues to be biased. Finally, we will look at the contemporary misuse and misunderstanding of genomic data and studies, including race-based nationalist dialogues, gene-editing and eugenics debates, and public understandings of precision rehtehw ,elpmaxe roff snoitseuq cificeps-enlipicid yb demarf eb liiw seirtuqni IIA .atad cimoneg namuh elbaliava ylcilbup ni noitairav level-ecneueqs terpretni dna ssesa ot R ni segakcap citeneg noitalupop gnisu dehsilpmocca eb liiw siHT ,devlove ytisrevid namuh woh no srotcaf rehto dna , snrettap gnitam ,noitatum ,tfrid ,noitceles fo selor laitnetop eht setadicule taht yaw a ni snamuh gnoma secnereffid puorg neewteb dna nihtiw htoB no thgil dehs nac emoneg eht ni noitairav woh ylevitatitnauq etalulave ot stneduts hcaet liiw sohtem esehT .scimoneg dna sciteneq noitalupop fo tæxtnoc eht ni demarf eb liiw yrotsih noitalupop dna yranoitulove namuh fo snoitseuQ :2 emoctuO gniaraE .secruos latnemmorivne dna cimoneg otni stiat detaler-ytisebo ni noitairav esopmocoed ot sisylana desab-eergidep esu ot ro ,lgninosiop cinesra ot detaler seneg dnuora noitceles wohs snoitulupop naivureP od , .e nrael senilepip dna sloot scitamrofnoib gnisu eruqer stcejorP elpmaxE .eciohc rieht fo)s(noitalupop ni eciohc rieht fo noiger eneg a gnidrager sisehtopyh yranoitulove na gnitset yduts dediuq-fles a tcudnoc ot rotcurtsni eht htiw noitatlusnoc ni seuqinhct laciitylana eseht esu liiw stneduts .sesylana cimoneg elacs-egral ot pu delacs eb neht liiw skrowemarf relpmis esehT .krowemarf evitatitnauq lautpeccoc elbisecca na ni meht nrael ot stneduts gnititnrep ,ssalc ni selpmaxe relpmis ni hguorht dekrow dna dessucsid eb liiw sciteneq noitalupop ot nommoc stset lacitistatS .yrotsih yranoitulove dna noitalupop namuh tuoba snoitseuq ksa ot redro ni seirotsoper enilno morf dedaolnwod atad cimoneg noitalupop namuh ezylana ot ,reitsicJ gnitupmoc detahs eht aiv dessecca ,jslootFCV ,slootMASI sloot sclamrofnoib dna jemoneGpoP ,Rtvc ,sagepI segakcap lacitistatS desab-R esu liiw ssalc siHT :1 emoctuO gniaraE .J2rQ (li gminosær evitatitnauq variation around the ACE2 gene, the primary receptor for SARS-CoV-2, shows differential signs of selection between populations). The diversity found in humans, on a genomic level, is often misinterpreted or misused in the public sphere. We will also assess how public representations of this human phenotypic variation may or may not accurately represent population genetic histories. Learning Outcome 3: After learning background and methods using a combination of lecture and increasingly complex online coding modules, each student will select a gene region associated with a human trait of interest. They will then use their understanding of human evolutionary and/or cultural history to form hypotheses regarding how that gene sequence is expected to vary within and across contemporary human populations. These hypotheses will be tested using the population genomic methods and tools learned in class. Most of these tools must be used in combination in order to provide adequate interpretive power (for example, students must test a sequence region for Hardy-Weinberg equilibrium, and then follow up those hits with further testing for deviations from neutrality using Tajima's D, and if that is indicative of positive selection use a test of Extended Haplotype Homozygosity to confirm a recent selective sweep in that region of the genome. Ultimately, a combination of literature review and their own quantitative analyses of genomic variation will be used to present their own argument for the history of this trait in their human populations of choice. Students will be encouraged to peer-review each other's presentations on the basis of the quality of their quantitative methods, and the submission of their final paper requires a thorough answering of peer questions regarding their methods, and modification of analyses if recommended by the class. Learning Outcome 4: Students will communicate their quantitative analyses and outcomes both visually and verbally The course. Each module comes with homework tasks required that are presented through Google Docs. Each requires explanations of statistical symbols associated with genomic analysis, as well as short essays that clearly describe what statistical evidence they perform and how this translates into biological meaning. This process culminates in an American Association of Physical Anthropologists-formatted presentation at the end of the course. These 15-minute presentations are intended to include graphical representations and analysis boards built in R packages (also known in online modules and classroom work) and results that are verbally interpreted in relation to their assumptions on human evolutionary history related to their choice gene. In this way, quantitative information will be symbolically correlated (students must present equations below their analytical methods in their documents, and demonstrate an in-depth understanding of what these symbols translate as regards the genomic sequence variation), visually (through graphs and figures that must also be interpreted, showing an understanding of what these figures are relating and how this reflects the meaning regarding the variation in the genome), and verbally in the presentation. The written paper combines these three communication modes. Learning Outcome 5: Critical to this course is transmitting an understanding of the underlying hypotheses and models every genomic test of the population. We discuss these assumptions and limits of these tests widely at the conference, and this is also an important aspect of the online modules that is underlined in homework tasks associated with the module. Integrate with the final presentation and the paper is a clear understanding of the hypotheses of each test, and how much - or little - these assumptions can limit Each test can tell us about the history and processes of the population data and populations used. Finally, each student must also discuss how these assumptions and models do or do not take into account important potential covariates and cultural aspects of modern human populations and population stories; This is an aspect of human genomics notoriously neglected even in highly impacted publications of human genomic and quantitative genetic analyses. research and information literacy (ri) learning result 1: students will learn and compare more methods to access free online repository of contemporary and ancient human genomic data, also evaluating the quality of those samples through a rigorous interpretation of literature published on their origins and analysis. to this end, students will read primary literature regarding the creation of repositories and types of genomic data, which will discuss the relative merits of accessing different types of genomic data (for example, construction of the assembly of the genome de novo from raw sequence data than the pre-built ombo of call formats variants and other more elaborate methods to store genomic data) and even reference axis (where through the process that downloads and analyzes its chosen genomic region, students must also repeatedly evaluate the quality of their data and critically reflect on how their processing techniques may have modified the content of the information of their genomic dataset (in fact, in past courses, data passed corruption often leads to results that students must detect and remedy using the tools provided in their modules). These studies will be integrated by discussions on social and ethical issues relating to data collection, data, and unique presentation for human genomic data sets, including discussions on the data of indigenous and non-Western populations and how the results of the analyses of these populations are presented and interpreted. This includes reflections on how they choose their own population to question, what cultural or biological hypotheses can they make to distort such choices and how their choice of population can also alter the way their analytical results can adequately address their demand. Learning result 2: å€ € Online exercises with problem solving sessions in class drive students through the genomic research process of the human population. This process follows a sequence from assignments relative to low works that familiarize students with data sources, interfaces and processing, gradually leading to a complete project in which students choose a gene to analyze comparatively between human populations, with an eye to understanding how the available data and resources necessarily limit their scope of investigation. This process is mediated by online modules that guide students through the initial process of data acquisition through increasingly complex methods of data processing and manipulation for population genetic evaluations. Students will learn quickly to ensure that, with each modification of the data set, the quality of their data remains high and has not been compromised by the faulty coding (a very common result) or by not meeting analytical hypotheses. After successfully completing the training modules, students then work with the instructor to independently develop a question related to human genomic variation which is limited to the resources provided (the project data set of 1000 genomes available). In itnedeuts itnedeuts ilg iuc ni enoizatneserp id ossecorp nu edeverp elanif otnemucod li .oroval orol led airatrap enoisiver anu onognetto iuc ni ehcisif igoloporna ad attamrof id inoizatneserp id anacirema enoizaicossa'nu ni essalc ni oroval orol li onatneserp itnedeuts ilg attach a cover letter directly addressing critiques raised by their peers during their presentation. This process prepares students for the entirety of the scientific process, from literature review and hypothesis formation through peer-review and presentation. presentation.

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