

## Short graduate-level course

# Population Genetics

July 20 to 31

Escuela de Postgrado

Facultad de Medicina, Universidad de Chile

Target public: Graduate students in genetics and related fields

Language: English

Duration: 38 hrs of lecture, 76 hrs take-home work (4 academic credits)

Schedule: 9:00-17:00: 20/7, 22/7, 27/7, 31/7

9:00-14:30: 21/7, 23/7, 24/7, 28/7, 29/7, 30/7

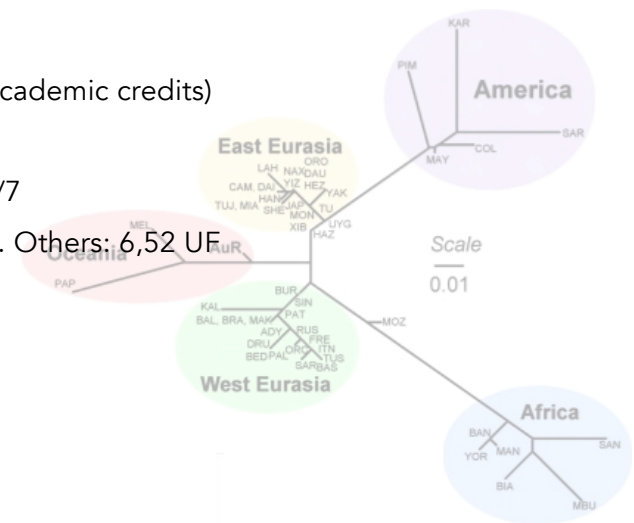
Admission: Graduate student enrolled in U. Chile: free. Others: 6,52 UF

### Topics

Module 1: Genetic population structure

Module 2: Predictive populations genetics

Module 3: Retrospective population genetics



Responsible instructor: **Ricardo Verdugo**, PhD.

Programa de Genética Humana, ICBM

Facultad de Medicina

Universidad de Chile



Invited instructor: **Michael Miller**, PhD.

Department of Animal Science

Integrative Genetics and

Genomics Graduate Group

University of California Davis, EE.UU.



Principles and practical application of population genetics, coalescence, and genomic-population studies. The course will be theoretical / practical and it will cover concepts of population genetics and coalescence to answer questions about population structure, demographic history and its effect on emergence and prevalence of diseases from genomic information.

**More information and registration: <http://genomed.med.uchile.cl/cursos/>**

*Funded by UC Davis and Proyecto Mecsup de Internacionalización del PDCBM.*

**Workshop weekly schedule**

	20-Jul	21-Jul	22-Jul	23-Jul	24-Jul
<b>9:00-10:00</b>	M1.1	M1.6	M1.9	M2.1	M2.4
<b>Break</b>					
<b>10:15-11:15</b>	M1.2	M1.7	M1.10	M2.2	M2.5
<b>Break</b>					
<b>11:30-12:30</b>	M1.3	M1.8	Discussion	M2.3	M2.6
<b>Lunch</b>					
<b>14:30-15:30</b>	M1.4		Test 1		
<b>Break</b>					
<b>16:00-17:00</b>	M1.5				

27-Jul	28-Jul	29-Jul	30-Jul	31-Jul
M2.7	Test 2	M3.2	M3.5	M3.8
M2.8		M3.3	M3.6	M3.9
M2.9	M3.1	M3.4	M3.7	Discussion
Discussion				Test 3



## CURSO DE POSTGRADO

*Curso Intensivo de Genética de Poblaciones*

Nombre Curso

SEMESTRE

2

AÑO

2015

PROF. ENCARGADO

*Ricardo Alejandro Verdugo Salgado*

13.199.074-K

Nombre Completo

RUT

*Programa de Genética Humana, ICBM, Facultad de Medicina, U-Chile*

UNIDAD ACADÉMICA

TELÉFONO

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E-MAIL

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TIPO DE CURSO

*Avanzado*

(Básico, Avanzado, Complementario, Seminarios Bibliográficos, Formación General)

CLASES	20 HRS.
SEMINARIOS	03:20
PRUEBAS	2 HRS.
TRABAJOS	06:40 HRS. (TRABAJOS PRÁCTICOS)

Nº HORAS PRESENCIALES	38
Nº HORAS NO PRESENCIALES	76
Nº HORAS TOTALES	114

CRÉDITOS

4

(1 Crédito Equivale a 30 Horas Semestrales)

CUPO ALUMNOS

4

(Nº mínimo)

25

(Nº máximo)

PRE-REQUISITOS

*Genética de poblaciones*

INICIO

*Ver Calendario de Actividades*

TERMINO

*Ver Calendario de Actividades*

DIA/HORARIO  
POR SESION

*Ver Calendario de Actividades*

DIA / HORARIO  
POR SESION

LUGAR

*A determinar*

Escuela De Postgrado (Sala a determinar) u otro lugar

## METODOLOGÍA

Las **clases teóricas** serán de carácter expositivo, basando en el desarrollo de problemas y metodologías en el pizarrón. **Todas las charlas y material del curso serán en idioma Inglés.** Al final de cada clase, el profesor asignará problemas a ser resueltos por los estudiantes para la clase siguiente.

Luego de cada clase, los estudiantes recibirán un problema a resolver diariamente. Al final de cada módulo tendremos una sesión de discusión donde los estudiantes podrán proponer problemas con los que hayan tenido dificultad, para que sean resueltos en conjunto.

(Clases, Seminarios, Prácticos)

## EVALUACIÓN (INDICAR % DE CADA EVALUACIÓN)

Habrà una prueba de desarrollo al final de cada módulo, basada en la resolución de problemas. La participación en clase será evaluada mediante un registro de la asistencia. El cálculo de la nota por participación será: (% de asistencia) x 6 + 1.

Ponderación de notas:

<i>Participación en clase</i>	25%
<i>Prueba Módulo 1</i>	25%
<i>Prueba Módulo 2</i>	25%
<i>Prueba Módulo 3</i>	25%

## PROFESORES PARTICIPANTES (INDICAR UNIDADES ACADEMICAS)

**Facultad de Medicina (FMed)**

*Programa de Genética Humana (PGH)*

*Instituto de Ciencias Biomédicas (ICBM)*

*Ricardo Verdugo (RV) – PGH, ICBM, raverdugo@u.uchile.cl*

**University of California Davis, EE.UU.**

*Department of Animal Science (ANS)*

*Integrative Genetics and Genomics Graduate Group (IGGG)*

*Dr. Michael Miller (MM) – ANS, IGGG, micmiller@ucdavis.edu*

## DESCRIPCIÓN / OBJETIVOS

Curso intensivo en Genética de Poblaciones, clases todos los días por dos semanas. **Todas las clases serán en Inglés.**

El curso cubriría los conceptos básicos de genética de poblaciones, genómica, coalescencia, y estudios genómico-poblacionales. El curso será teórico/práctico y entregará las herramientas para que los estudiantes rápidamente puedan hacer uso de información genómica para responder preguntas sobre estructura poblacional, historia demográfica y su efecto en aparición y prevalencia enfermedades.

## CALENDARIO DE ACTIVIDADES

(A continuación señalar : Descripción de la actividad, fechas, horas presenciales y no presenciales y Profesores a cargo)

Todas las clases se realizan en la Escuela de Posgrado: Bloque F, 2° piso, Facultad de Medicina. Independencia 1027.

Fecha y ubicación	Horas Presenciales	Horas no presenciales	Descripción Actividad	Profesor
<b>Module 1: Genetic population structure: principles and applications</b>				
M1.1 07/20 9:00-10:00	1	2	CT: Course introduction. How to summarize data from one SNP in one population Genetic variation, allele and genotype frequencies	RV, MM
M1.2 07/20 10:15-11:15	1	2	CT: How accurate are my allele and genotype frequency estimates? Parameters vs statistics, confidence intervals	MM
M1.3 07/20 11:30-12:30	1	2	CT: What is the expected relationship between allele and genotype frequencies? Setting up model to determine expectations, HWE, comparing data to expectation using chi-square test	MM
M1.4 07/20 14:30-15:30	1	2	CT: How to summarize data from two SNPs in one population Concept of haplotype, haplotype frequency, expected haplotype frequency, $D$ , $D'$ , $r^2$ , $r^2*n$	MM
M1.5 07/20 16:00-17:00	1	2	CT: Relationship between mutation, recombination rate and LD New mutation arise on background of pre-existing variation	MM
M1.6 07/21 9:00-10:00	1	2	CT: Genome-wide association studies (GWAS) Decay in LD, hapmap projects, Correction for multiple tests	MM
M1.7 07/21 10:15-11:15	1	2	CT: How to summarize data from more than than one population (allele frequency differences, heterozygosity, F-statistics)	MM
M1.8 07/21 11:30-12:30	1	2	CT: Relationship between populations (based on allele frequencies from groups of individuals)	MM
M1.9 07/22 9:00-10:00	1	2	CT: Genetic assignment Maximum likelihood and Bayesian analysis	MM

M1.10 07/22 10:15-11:15	1	2	CT: Methods for determining population structure from individuals genotypes PCA, model-based clustering	MM
M1.D 07/22 11:30-12:30	1	2	Discussion: Module 1 discussion Time to answer questions and about material covered in Module 1. We can solve together problems assigned in each lecture of Module 1.	MM, RV
M1.T 07/22 14:30-17:00			Examination: Test 1 The students will solve problems related to material covered in Module 1 individually.	RV
<b>Module 2: Predictive populations genetics: principles and applications</b>				
M2.1 07/23 9:00-10:00	1	2	CT: Genetic drift in idealized population Wright-Fisher Model, binomial random variable, expectation, performing simulations	MM
M2.2 07/23 10:15-11:15	1	2	CT: Genetic drift in real populations Effective population size, relationship between $N_c$ and $N_e$	MM
M2.3 07/23 11:30-12:30	1	2	CT: Genetic drift and mutation $P(\text{fixation}) = \text{allele frequency}$ , rate of substitution is independent of population size	MM
M2.4 07/24 9:00-10:00	1	2	CT: Molecular clock and phylogenetic inference	MM
M2.5 07/24 10:15-11:15	1	2	CT: Selection in infinite populations Selection coefficient, fitness	MM
M2.6 07/24 11:30-12:30	1	2	CT: Genetic hitchhiking	MM
M2.7 07/27 9:00-10:00	1	2	CT: $F_{st}$ and LD-based approaches to detect selection: $F_{st}$ outliers, $p$ -excess	MM
M2.8 07/27 10:15-11:15	1	2	CT: Selection in finite populations Mutation, selection, and drift combined	MM
M2.9 07/27 11:30-12:30	1	2	CT: Migration migration rate ( $m$ ), number of migrants ( $M$ ), relationship to $F_{st}$	MM
M2.D 07/27 14:30-15:30	1	2	Discussion: Module 2 discussion Time to answer questions and about material covered in Module 2. We can solve together problems assigned in each lecture of Module 2.	MM, RV
M2.T 07/28 9:00-10:00	1	2	Examination: Test 2 The students will solve problems related to material covered in Module 2 individually.	MM

<b>Module 3: Retrospective population genetics: principles and applications</b>				
M3.1 07/28 11:30-12:30	1	2	CT: How to summarize sequence data segregating site (s), average pairwise nucleotide divergence (pie), site frequency spectrum (sfs), ancestral vs derived	MM
M3.2 07/29 9:00-10:00	1	2	CT: What is the expectation for pie under the Wright-Fisher Model (coalescent process for n=2) dice example and simulation, exponential distribution approximation, theta (4Nu), infinite sites model, Tjima's estimator	MM
M3.3 07/29 10:15-11:15	1	2	CT: What is the expectation for s under the Wright-Fisher Model (coalescent process for n>2 part I) TMRCA, TTL, Watterson's estimator	MM
M3.4 07/29 11:30-12:30	1	2	CT: What is the expectation for sfs under the Wright-Fisher Model (coalescent process for n>2 part II) Tree topology and site frequency spectrum (SFS)	MM
M3.5 07/30 9:00-10:00	1	2	CT: This population's sfs looks strange: the effect of population size change on the sfs The effect of population size change (bottleneck and expansion) on tree topology and SFS	MM
M3.6 07/30 10:15-11:15	1	2	CT: How can I estimate historic demography (population size changes) based on an sfs? Intro to coalescent simulations and ABC	MM
M3.7 07/30 11:30-12:30	1	2	CT: 2d-sfs and subpopulation demography Hey NRG review, distinct populations with migration vs divergence with no migration (different SFSs)	MM
M3.8 07/31 9:00-10:00	1	2	CT: Testing complex demographic models with coalescent simulations and MCMC Read a paper	MM
M3.9 07/31 10:15-11:15	1	2	CT: Selection also influences the sfs: the effect of positive and negative selection Genomic region SFS, negative selection, positive selection, Tjima's D	MM
M3.D 07/31 11:30-12:30	1	2	Discussion: We will review the mayor topics covered and answer questions from the students	MM, RV
M3.T 07/31 14:30-17:00	1	2	Examination: Test 3	RV