NYU DEPARTMENT OF BIOLOGY

# BIOL-UA 45 Biostatistics and Human Genetics

**Instructor:**
David Gresham

**Course Description:**
Deciphering the information encoded in the human genome is one of the greatest (and most exciting) challenges of the 21st century. This course will provide an introduction to studying and interpreting the human genome with a focus on the statistical methods required for its study. Fundamental concepts in human genetics will be introduced including inheritance of mendelian disease, population genetics, multifactorial disease and functional genomics. Accompanying each topic will be an introduction to the statistical concepts and tools that are required to study inheritance, genes and gene function. These include probability, hypothesis testing, ANOVA, regression, correlation and likelihood. Hands on experience will be provided through weekly assignments using the statistical programming language, R. Prior experience with statistics and genetics is not required

**Pre-requisites:**
Principles of Biology II (BIOL-UA 12).
Molecular and Cell Biology I (BIOL-UA 21)
Molecular and Cell Biology II (BIOL-UA 22)

**Textbook and Required Materials:**
Introduction to Genetic Analysis, 10th Edition
Griffiths, Wessler, Carrol, Doebley
The Analysis of Biological Data
Whitlock & Schluter

**Grading:**

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| Assignments: | 50% |
| Quizzes: | 10% |
| Midterm: | 15% |
| Final Exam: | 20% |
| Attendance/Participation: | 5% |

**Topics:**
An introduction to using R and Rstudio
Using Rmarkdown for reproducible research
Genetics: Distributions of human phenotypes
Statistics: Descriptive Statistics
Genetics: Samples from populations
Statistics: Uncertainty, sampling distributions, standard error
Genetics: Mendel's experiments and expected phenotypic proportions
Statistics: Probability
Genetics: Independent assortment, introduction to linkage
Statistics: chi square, contingency tables, hypothesis testing
Genetics: Mendelian inheritance in humans I, penetrance
Statistics: Conditional probability, Relative risk
Genetics: Mendelian inheritance in humans II, Segregation ratios
Statistics: Binomial distribution
Genetics: Linkage, recombination, three factor cross
Statistics: Probability

Genetics: Interference, Genetic variation in humans, Mapping functions
Statistics: Poisson distribution
Genetics: Linkage analysis in human pedigrees
Statistics: Likelihood
Genetics: Human Linkage Analysis, Refined genetic mapping
Statistics: Likelihood, LOD scores
Genetics: Genetic testing
Statistics: Law of total probability, conditional probability; Bayes Theorem
Genetics: Gene frequencies in populations
Statistics: Hardy-Weinberg equilibrium
Genetics: Inbreeding
Statistics: Recursive calculations
Genetics: Genetic drift, Selection
Statistics: Binomial sampling, simulation
Genetics: Genetic diversity, Linkage disequilibrium, mutation-drift equilibrium
Statistics: Computational simulation
Genetics: Distribution of quantitative traits
Statistics: Normal distribution, z-scores, Central limit theorem
Genetics: Sampling quantitative phenotypes
Statistics: Student's t-test, confidence intervals
Genetics: Comparison of quantitative phenotypes between two populations
Statistics: Two sample t-test
Genetics: Comparison of quantitative traits in more than two populations
Statistics: ANOVA
Genetics: Broad sense heritability
Statistics: Covariance, correlation
Genetics: Narrow sense heritability
Statistics: Linear regression I
Genetics: Narrow sense heritability and prediction
Statistics: Linear regression II: significance and variance explained
Genetics: Genome-wide expression analysis
Statistics: Non-parametric methods
Genetics: eQTL mapping
Statistics: Randomization and Bootstrapping
Genetics: Genome-wide association studies
Statistics: Odds ratio, multiple hypothesis testing