

UW

Introduction

- Two primary existing methods for mutation rate estimation:
 - Counting de novo mutations in parent-offspring trios
 - Difficult to control for genotype error
 - Small numbers of observed mutation events
- Inter-species comparison?
 - Biased by selection, incorrect values for average generation length, uncertainty in the fossil record and changes in mutation rate over time
- Recently, a new approach to mutation rate estimation based on identity by descent (IBD) segments was proposed?
 - We propose to use multi-individuals, which control for phasing uncertainty and genotyping errors

Identity by Descent

- A DNA segment is identical by descent (IBD) in two or more individuals if they have inherited it from a common ancestor without recombination
- The advantages of our approach are:
 - In large population samples, millions of IBD segments are present, and each provides information on multiple meioses
 - Long segments of IBD look back thousands rather than millions of years, which avoids difficulties present with inter-species comparison
 - IBD sharing of the rare variant increases the confidence that the variant is not a genotype artifact

Method

- The estimation of mutation rate is based on IBD trios where three individuals share a single haplotype IBD (Fig. 1)
- We use the haplotype sharing pattern (Fig. 2) and the recombination rates consistent with the rare variant sharing (Fig. 3) to obtain the likelihood of the mutation rate

Fig. 2. Example of haplotype sharing among A, B and C three individuals.

Fig. 3. Relationship between recombination rate and the length of IBD segments.

TOPMed Project

Study data from TOPMed project

DEPARTMENT OF BIostatISTICS

Revolutionizing public health by turning data into knowledge

The Department of Biostatistics is a recognized leader in the statistical sciences. As technological advances generate huge quantities of complex data and create exciting opportunities to develop innovative statistical methods, we provide rigorous training and research experience to equip the next generation of biostatisticians.

FAST FACTS

#1 BIostatISTICS GRADUATE PROGRAM
- IN THE U.S.

GLOBAL LEADER IN RESEARCH
- BIG DATA - STATISTICAL GENETICS - CLINICAL TRIALS

OUR RESEARCH

Wide Ranging Impact

- We are statistical scientists who tackle real problems in public health, biology and medicine.
- We are communicators who speak the language of our clinical colleagues and understand key statistical issues as well as underlying biological and medical contexts.
- We are innovators who develop and apply new statistical methods to solve some of today's most pressing questions: Is a new drug safe? How does air pollution impact public health?
- We provide critical insights into data analysis and help research teams:
 - Design studies
 - Identify data to collect
 - Develop statistical techniques
 - Monitor and analyze data
 - Interpret findings
 - Communicate results and recommendations

Research Areas

Biomarkers * Causal Inference * Clinical Trials * Environmental and Spatial Statistics * Epidemiological Methods * Foundations of Statistics * High Dimensional Data and Statistical Learning * Longitudinal and Multilevel Data * Statistical Genetics and Genomics * Survival Analysis



OUR PARTNERS

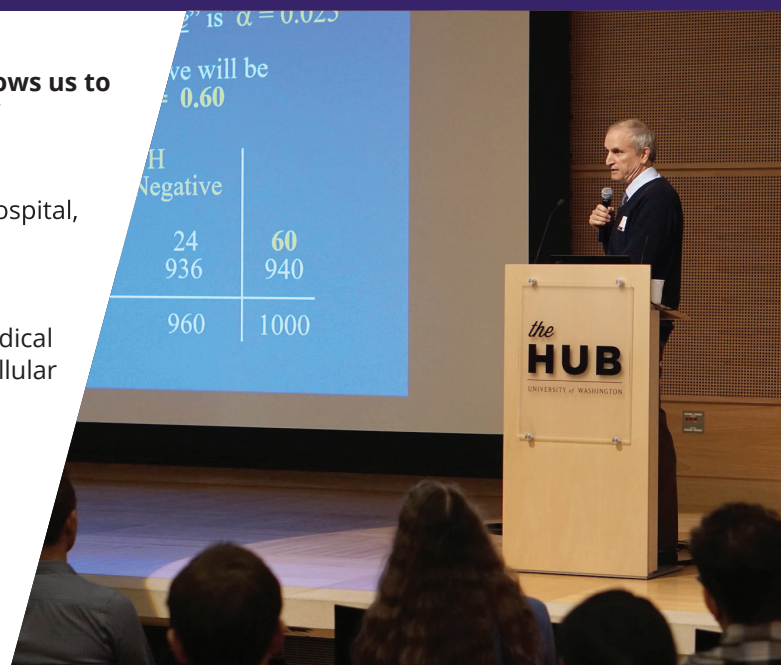
The collaborative, interdisciplinary nature of our work allows us to leverage the resources and expertise of a diverse group of organizations and UW partners including:

Fred Hutchinson Cancer Research Center, Seattle Children's Hospital, Kaiser Permanente, Veterans Administration

UW partners: School of Medicine, Statistics, Data Science, Epidemiology, Health Services, Biomedical Informatics and Medical Education, Genome Sciences, Neuroscience, Molecular and Cellular Biology, University of Washington DISCOVER Center

Affiliate Research Organizations

Alzheimer's Disease Research Center * Center for Ecogenetics and Environmental Health * Center for Statistical Consulting * Northwest PRECEDENT * Statistical Center for HIV/AIDS Research & Prevention (SCHARP) * Vaccine and Infectious Disease Institute



EDUCATION

3 Academic Degree Programs

- MS Capstone: Specializations in Data Science, Statistical Genetics, and Modeling and Methods
- MS Thesis: Foundation in the science of biostatistics as well as practical application leading to research position or pursuit of PhD
- PhD: Standard Pathway and Statistical Genetics Pathway
- 2 Certificate Programs

676 Alumni

Our graduates play leadership roles in academia, government and industry at local, national, and global levels. **99%** of graduates are in jobs or postgraduate programs within 1 year of graduation.

89 Faculty

37 Core
5 Adjunct
30 Clinical & Affiliate
15 Emeritus

91 Current Grad Students

46 Masters students
95 PhD students

2500+ Trained Annually

Number of national and international researchers we train annually through service teaching and programs such as the Summer Institutes and Seattle Symposiums

Contact Information