Population and Quantitative Genetics – HS18

v0.1

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Vorwort

This document aims to summarize the lecture Population and Quantitative Genetics as it was taught in the autumn semester of 2018. Unfortunately I can't guarantee that it is complete and free of errors. You can contact me under `glebert@student.ethz.ch` if you have any suggestions for improvement. The newest version of this summary can always be found here: `https://n.ethz.ch/~glebert/`

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1 Introduction

Population genetics apply Mendel's laws and other genetic principles to populations. It studies genetic variation within and between populations and species and the forces that result in evolutionary changes in populations and species through time. Population genetics are useful in studies of evolutionary processes, conservation, medicine, agriculture and other fields.

1.1 Useful Formulas

mean	$\overline{x} = \frac{1}{n}\sum_{i=1}^{n} x_i$
variance	$V_x = \frac{1}{n-1}\sum_{i=1}^{n}(x_i - \overline{x})^2$
standard deviation	$sd = \sqrt{V_x}$

1.2 Genetic Variation

"Nothing in biology makes sense except in the light of evolution"
— Theodosius Dobzhansky

Modern synthesis brought Mendelian genetics together with Darwin's theory of natural selection to help quantifying the genetic variation in natural populations.
The classical view of genome organization was that wild type alleles made up the whole genome with a few mutations in between. The balanced view however said that for each gene there are multiple alleles and that heterozygosity is possible.

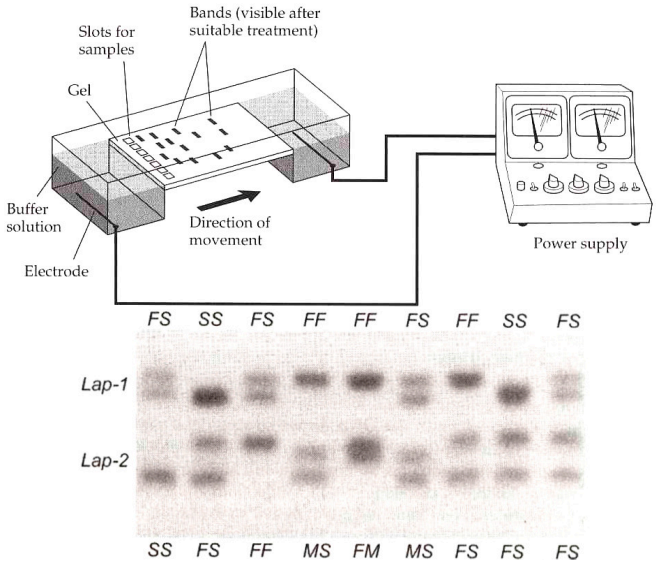
1.3 Genetic variation

Experimental methods for detecting genetic variation (in chronological order):

- Allozyme electrophoresis
- DNA (Sanger) sequencing
- Restriction fragment length polymorphism (RFLP)
- Simple sequence repeats (SSR or microsattelites)
- Amplified fragment length polymorphism (AFLP)
- Single nucleotide polymorphisms (SNPs)
- Next-generation sequencing (NGS)

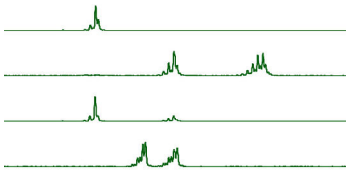
1.3.1 Allozyme electrophoresis

Enzymes that differ in electrophoretic mobility as a result of allelic differences in a single gene are called allozymes. Allozyme electrophoresis separates these. Allozymes underestimate the levels DNA polymorphism because they detect only a subset of existing amino acid replacement and they do not detect synonymous mutations. However, they may also overestimate polymorphisms because they represent mostly groupd 1 enzymes (common in tissues and body fluids) and enzyme polymorphisms may not be neutral and therefore not reflect polymorphism elsewhere in the genome. Allozyme electrophoresis was replaced by DNA electrophoresis.



1.3.2 Microsattelites (SSRs, VNTRs)

Microsattelites are highly polymorphic markers that are widely used in animals, plants and fungi to assess genetic variation (e.g. ...GATCGA$\mathbf{(GC)}_7$TAGCCGAT...). They – like allozymes – are codominant markers. Homozygotes can be distinguished from heterozygotes.



1.3.3 Amplified Fragment Length Polymorphisms (AFLPs)

AFLPs are dominant markers. Homozygotes cannot be distinguished from heterozygotes.

1.3.4 Types of Polymorphisms

A polymorphism that doesn't alter the amino acid sequence is called synonymous. They are a consequence of the redundancy of the genetic code. Non-synonymous or replacement polymorphisms change the amino acid. If a polymorphism is noncoding or silent, it doesn't affect nucleotides in coding regions. Nucleotide polymorphisms can be devided into insertion/deletion polymorphisms (indels) and single-nucleotide polymorphisms (SNPs). A unique combination of linked genetic markers is often called a haplotype.

1.3.5 Genetic Variation in Natural Populations

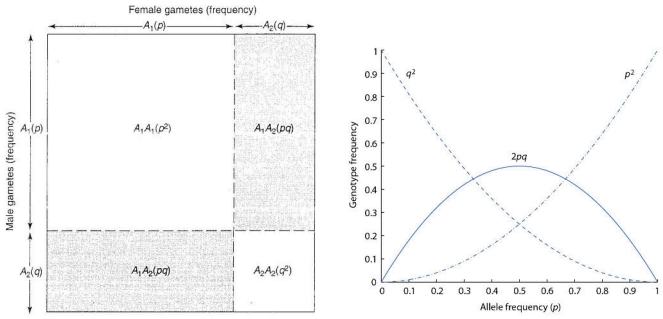
A population is a group of interbreeding, same-species individuals that exist together in time and space. If it is random-mating (panmictic), the probability of mating between individuals of particular genotypes is equal to the product of their individual frequencies in the population.

1.4 The Hardy-Weinberg Principle

The Hardy-Weinberg principle describes allele frequencies in diploid populations. It applies after one generation of random mating. The principle assumes that allele frequency

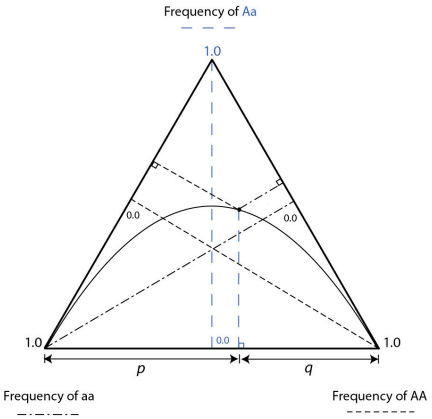
changing factors (natural selection, drift, ...) are absent.

$$(p+q)^2 = p^2 + 2pq + q^2 = 1$$



At low frequencies the majority of a certain allele occurs in heterozygous individuals.

1.5 De Finetti Diagram



De Finetti diagram for one locus with two alleles. The parabola describes HW expected genotype frequencies.