

Exercises in Marine Ecological Genetics

01. Introduction

- General info and course outline
- Connect to the HPC cluster
- Work on the command line
- Test for HWE in R

Martin Helmkamp



General course info

- Suggestion: we start at 14:00 and finish at 15:30
- Course language will be English, but questions can always be asked in German
- There will be no tests or grades
- Slides will be provided, but please do not post them online
- Use of personal laptops is recommended, but workstations are available
- Contact: [martin.helmkamp \(at\) uni-oldenburg.de](mailto:martin.helmkamp@uni-oldenburg.de)

Objectives

- Apply theory and concepts of population genetics, genomics and DNA barcoding in practice
- Learn to analyze, visualize and interpret real world data
- Learn how to work on a high performance computing cluster (using bash) / a scripting environment (using R)
- Become familiar with the most common data types and file formats

Course outline

Preliminary, may be subject to change

Class	Date	Topics	Script
01	Apr 11	Introduction, setup	01_intro.R
–	Apr 18	Good Friday	
02	Apr 25	Hardy-Weinberg equilibrium / N_e	
03	May 02	Population structure and gene flow	
04	May 09	Genome assembly and metrics	
05	May 16	SNPs and population genomics	
06	May 23	Recombination and linkage disequilibrium	
–	May 30	Himmelfahrt break	
07	Jun 06	Measures of genetic diversity	
–	Jun 13	Selection and Mutation	
08	Jun 20	Student presentations – no exercises	
09	Jun 27	DNA barcoding	
10	Jul 04	Metabarcoding / eDNA	
11	Jul 11	Metabarcoding / eDNA II	

Required software

- Browser
- Text editor (e.g. Notepad, TextEdit, [VSCodium](#))
- Terminal / ssh client (e.g. git bash, Terminal)

To connect to the high performance computing cluster ROSA, a ssh client is required

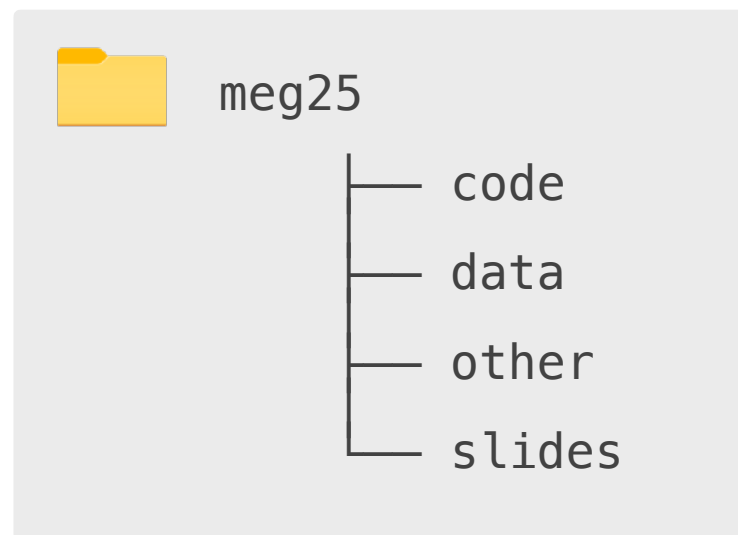
On Windows, a good option is git bash, which is part of Git for Windows. Install from <https://gitforwindows.org> with default settings (alternative: WSL)

On macOS or Linux, a terminal (and git) come preinstalled

Accessing the course materials

All code and data for the course will be provided through a Git repository:

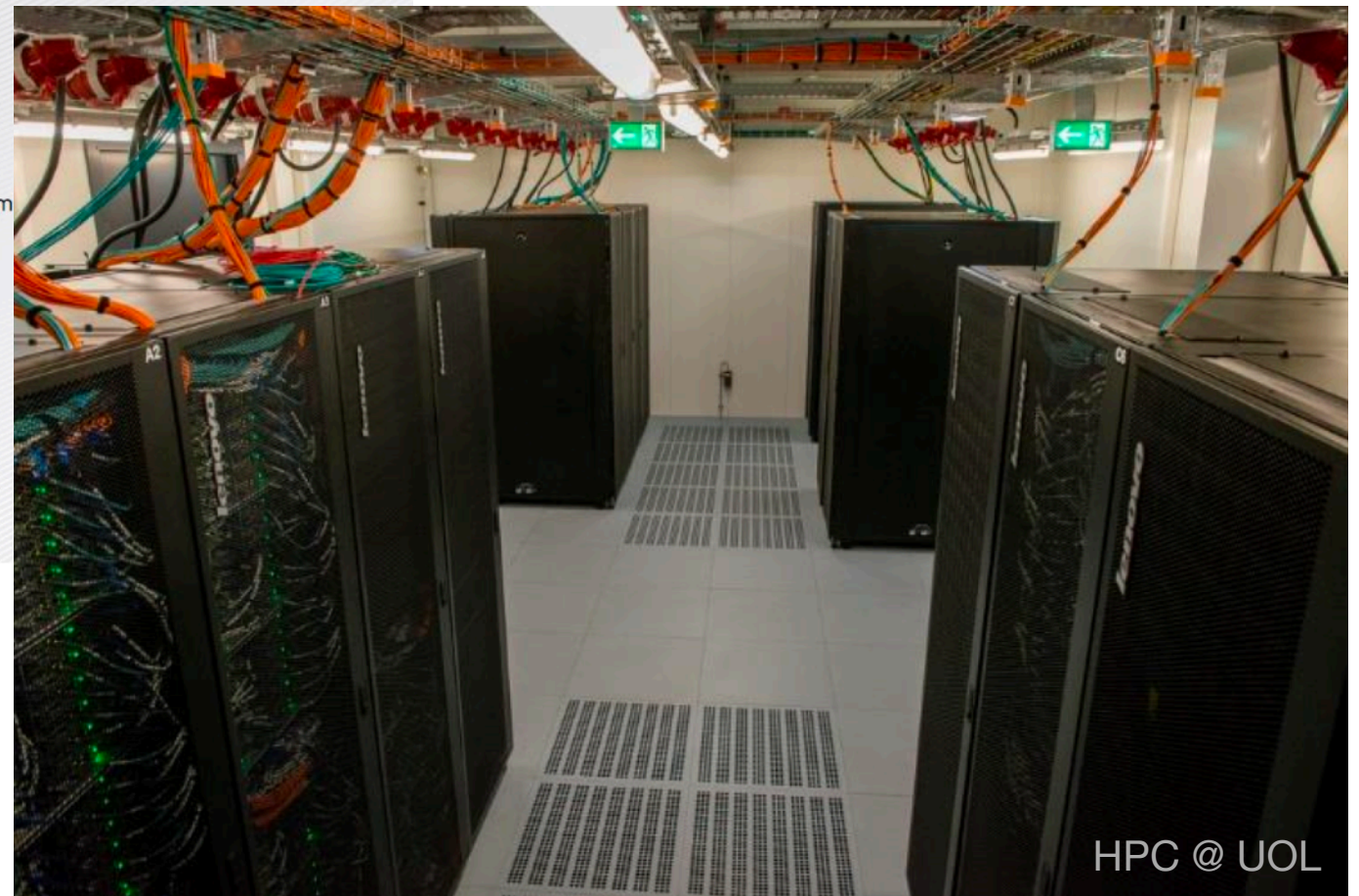
<https://github.com/mhelmkamp/meg25>



High performance computing on ROSA at UOL



91 compute nodes
11,648 cores (CPUs)
91 TB RAM total
625 TFlop/s (R_{\max})



<https://hpcwiki.uni-oldenburg.de/>

Advantages of command line / scripting tools

- Highly flexible
- Can be automated and combined into complex workflows
- Reproducible, easy to document
- Can run on high performance computers

Accessing the cluster from the command line (ssh)

Windows

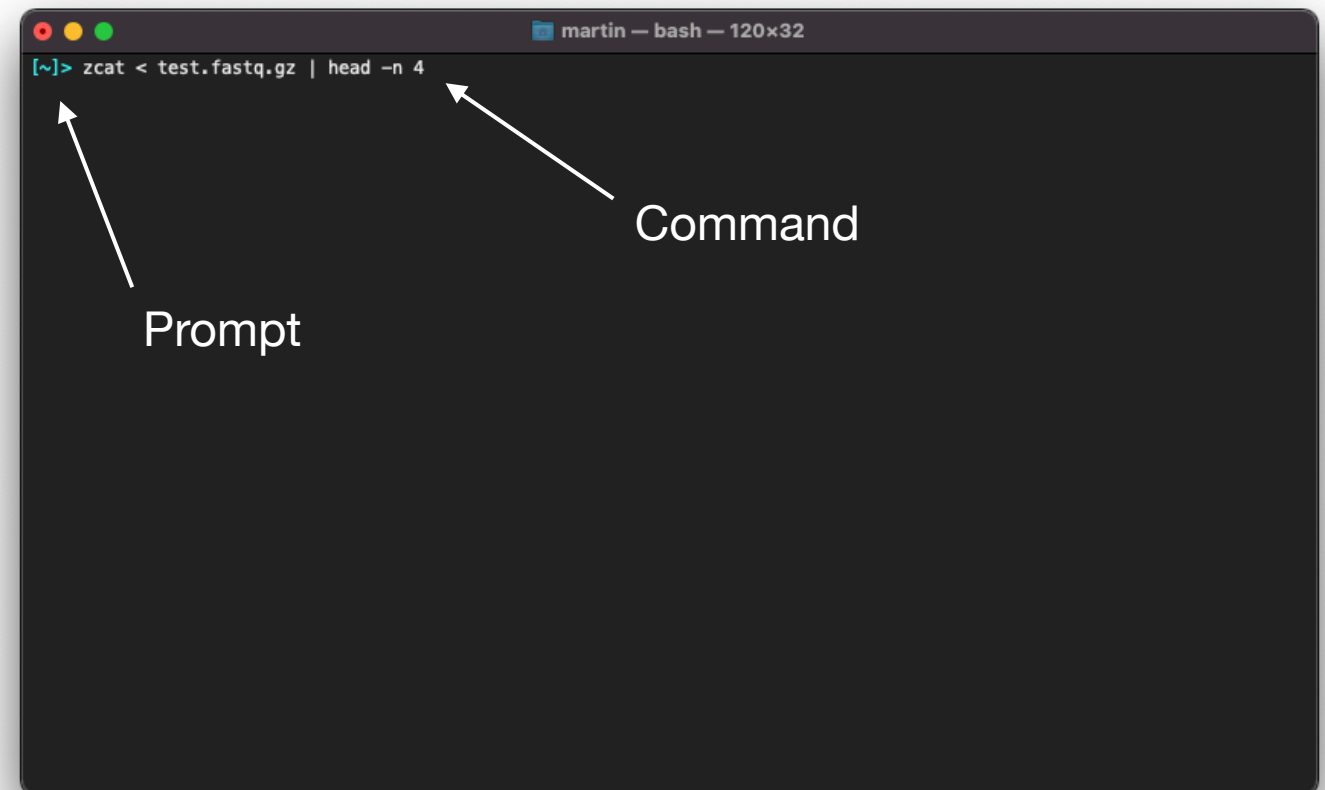
- launch git bash from Start menu
- alternatively, launch terminal in Windows Subsystem for Linux (WSL) on Windows 10 or above (see <https://learn.microsoft.com/en-us/windows/wsl/install>)

macOS

- open Terminal app
in /Applications/Utilities,
type and execute “bash”

Typical usage

`command [-options] [file]`



Get set up on the HPC cluster

Connect to login node

```
# Pick and write down a course account user name and password (passed around)  
ssh user1234@rosa.hpc.uni-oldenburg.de
```

Download course materials to cluster account using git

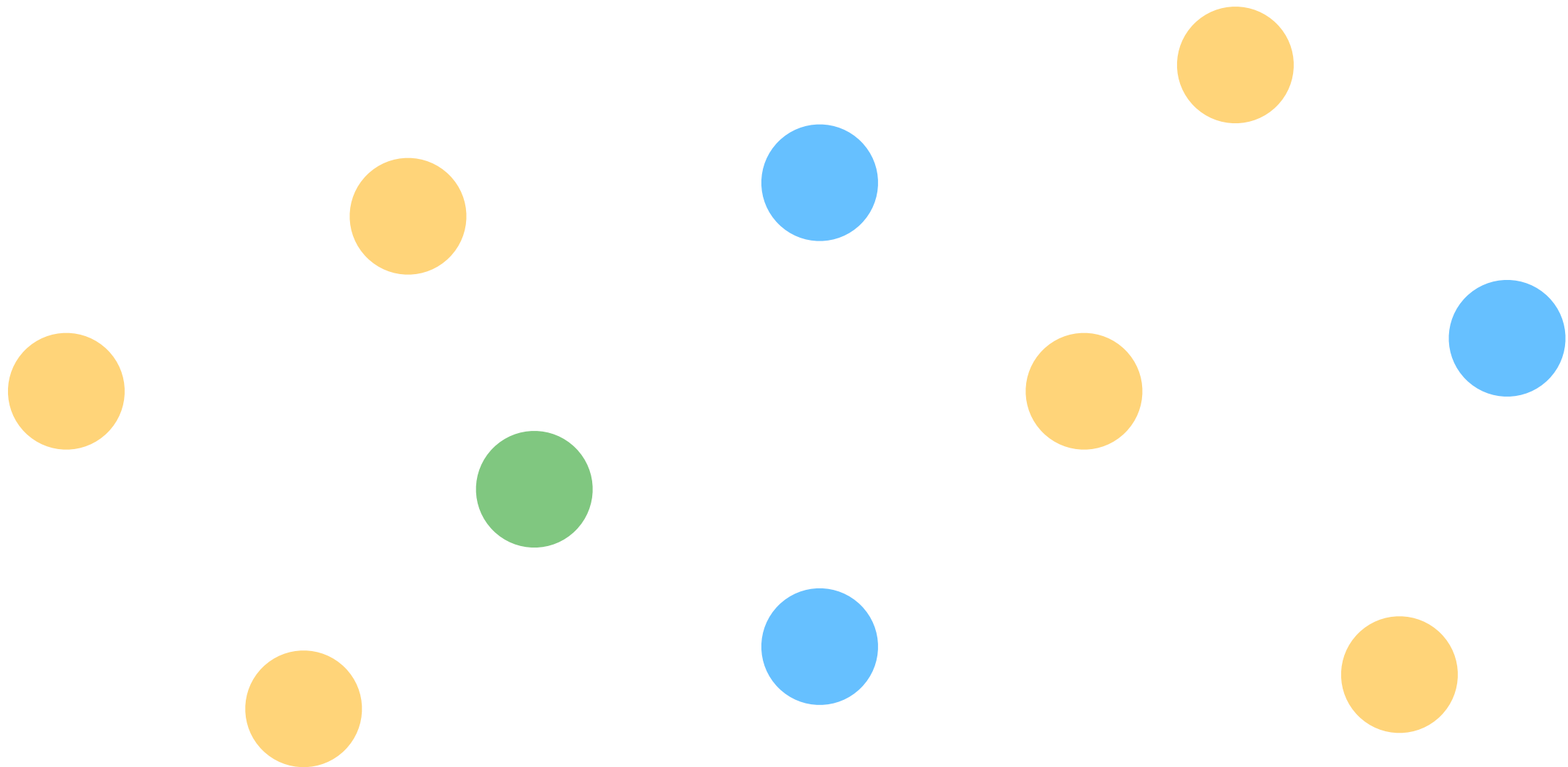
```
git clone https://github.com/mhelmkampf/meg25.git
```

Hardy-Weinberg Equilibrium

Exercise 1

Is this population in HWE?

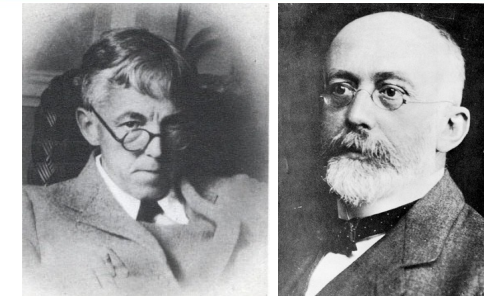
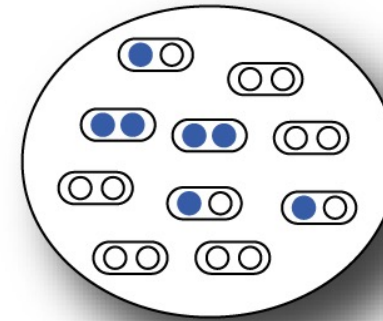
Diploid, 1 locus, 2 co-dominant alleles: yellow, blue



HARDY-WEINBERG (1908)

Godfrey H. Hardy (1877-1947)

Wilhelm Weinberg (1862-1937)

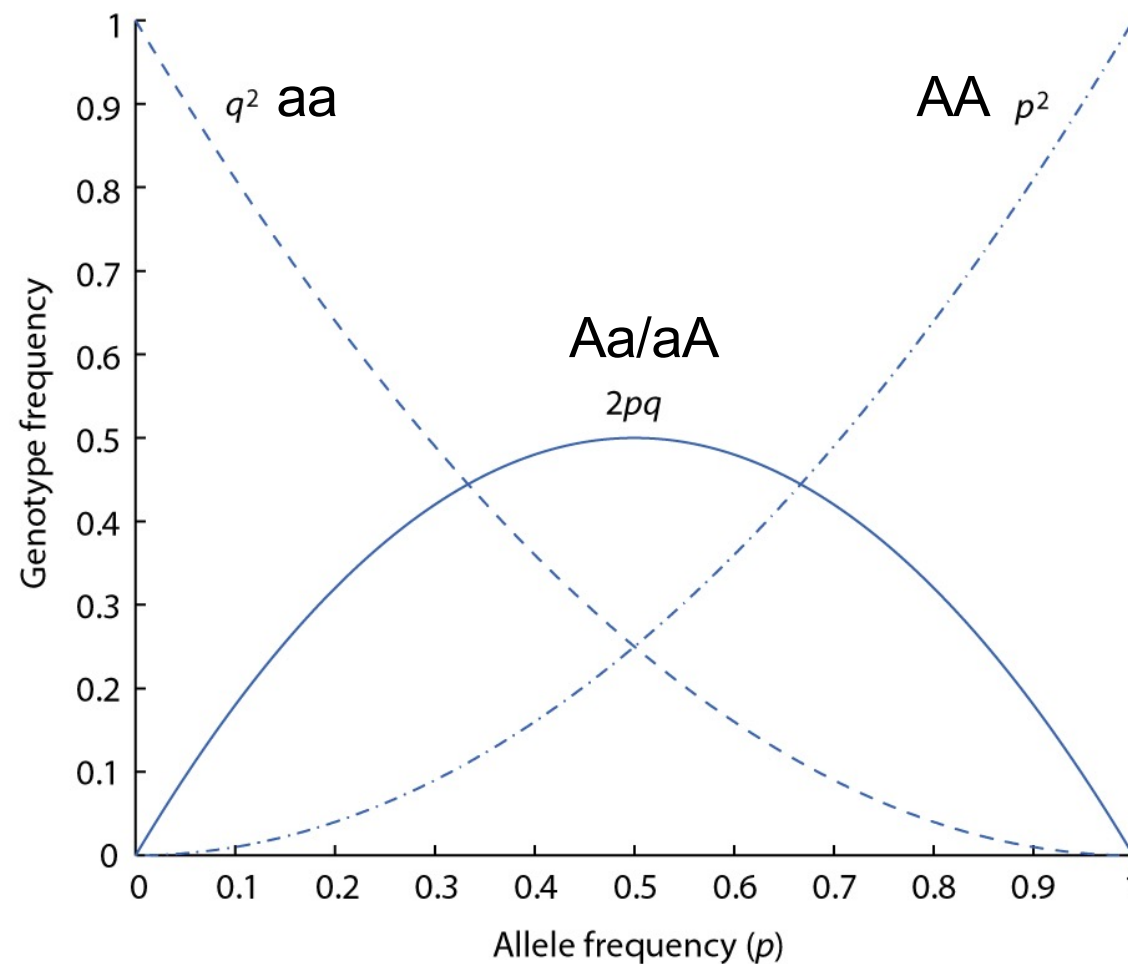


Establish the relationship between allele frequencies and genotype frequencies in a population

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

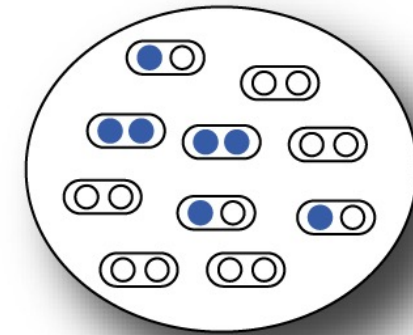
AA Aa/aA aa

p and q : allele frequencies for a locus with two alleles (A and a)
($p + q = 1$)



HETEROZYGOSITY

In one population



H_o = proportion of heterozygote individuals, observed heterozygosity

$H_e = 2pq = 1 - p^2 - q^2$, expected heterozygosity (assuming HW equilibrium)

$$F = \frac{H_e - H_o}{H_e}$$

Fixation index: *proportion by which heterozygosity is reduced or increased relative to the heterozygosity of a population at HW equilibrium with the same allele frequencies.*

Divided by $H_e \rightarrow$ *proportion* (of expected heterozygosity)

Varies between -1 and 1

$F < 0$: heterozygote excess

$F > 0$ heterozygote deficit (homozygote excess)

May be averaged over several loci \rightarrow reduces bias

May be extended to k alleles

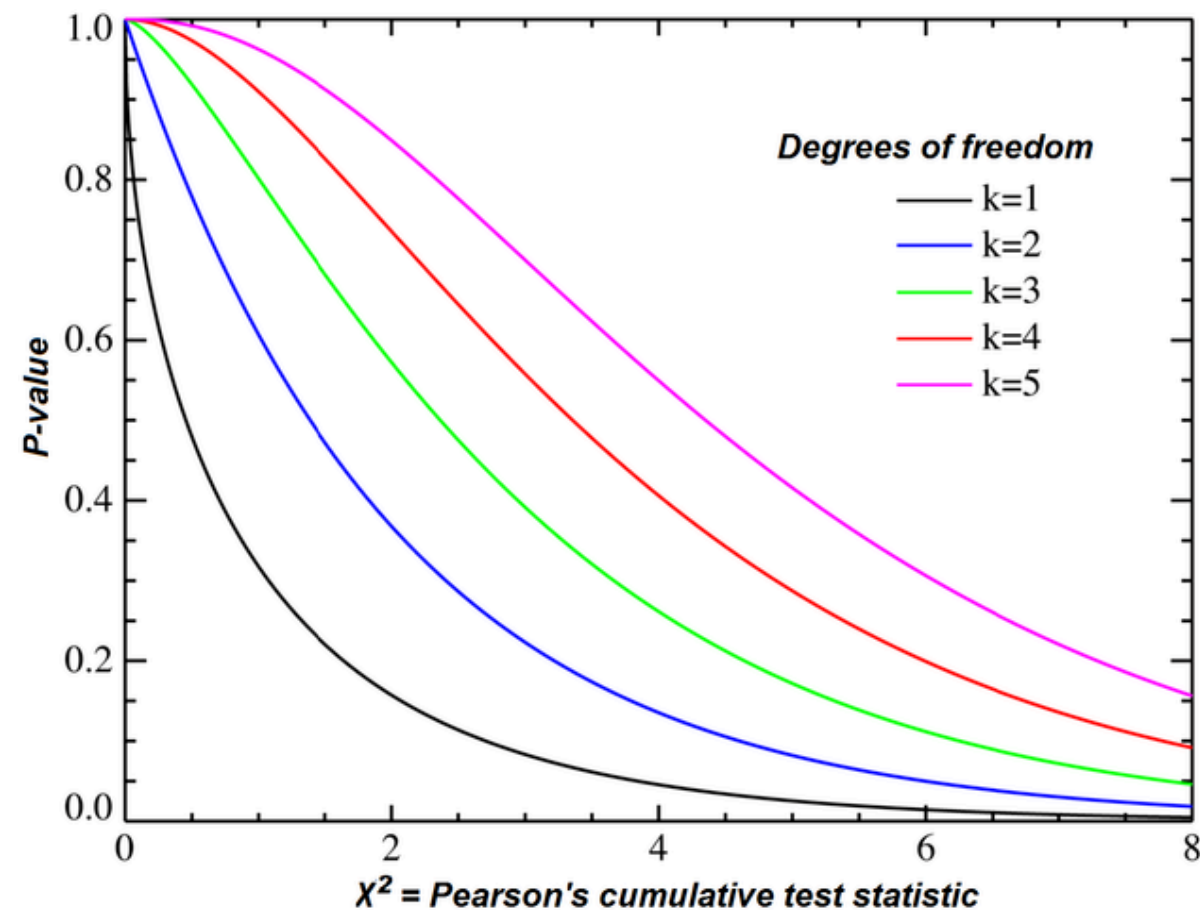
Pearson's chi-squared test

Exercise 1

Chi-square statistic:

$$\chi^2 = \sum_{i=1}^n \frac{(O_i - E_i)^2}{E_i}$$

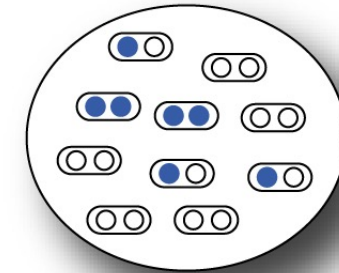
Chi-square distribution:



A single generation of reproduction will result in a population that meets the expected Hardy-Weinberg frequencies, i.e. is at Hardy-Weinberg (HW) equilibrium

Assuming an “ideal” population, i.e. :

- Diploid organisms
- Sexual reproduction (as opposed to clonal)
- Random mating (as opposed to e.g. assortative) with respect to genotype
- Random union of gametes
- Discrete, non-overlapping generations
- Very large (infinite) population
- No migration
- No population structure
- No natural selection
- Two alleles
- Identical allele frequencies in both sexes



-> Departures from HW equilibrium may indicate:

- Inbreeding
- Assortative mating
- Self-fertilization
- Natural selection
- Population structure
- ...