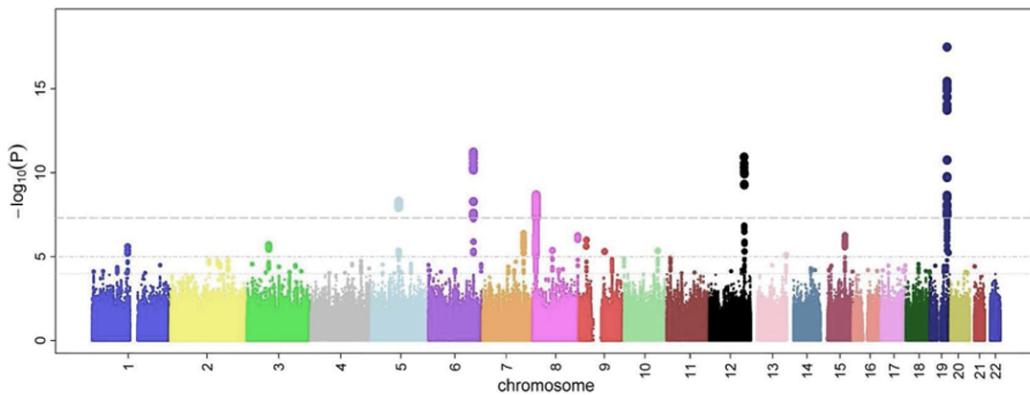


NGS – variant analysis

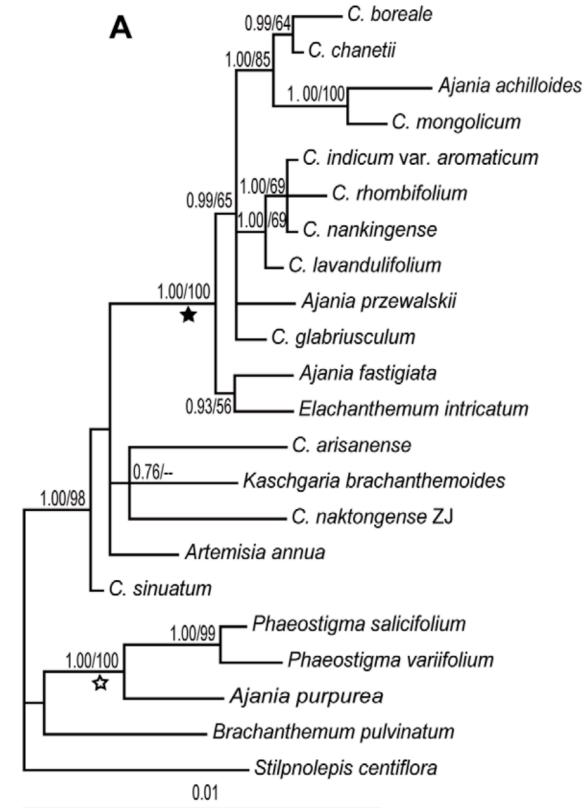
Introduction to variant analysis

Why study variants?

- Find causes for phenotypic variation
- Understand relatedness

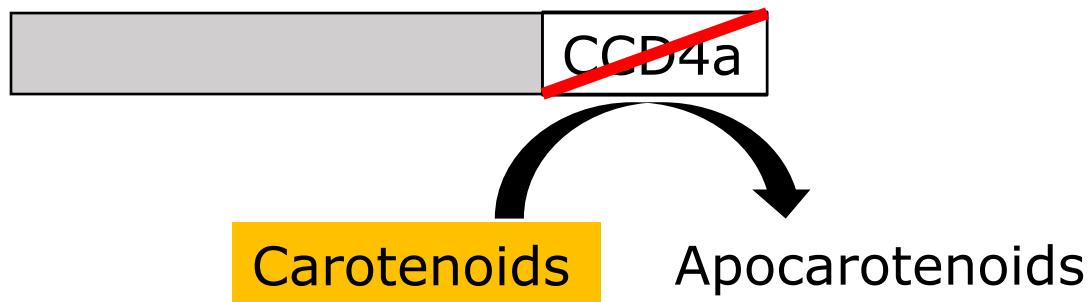


https://en.wikipedia.org/wiki/Genome-wide_association_study



Mutation

Change in DNA sequence



Mutations - causes

Change in DNA sequence

- Repair mistakes
- Unbalanced mitosis
- Transposable elements



https://nl.wikipedia.org/wiki/Springend_gen

Genomic variation

- cells – somatic mutation
- inherited – germline mutation

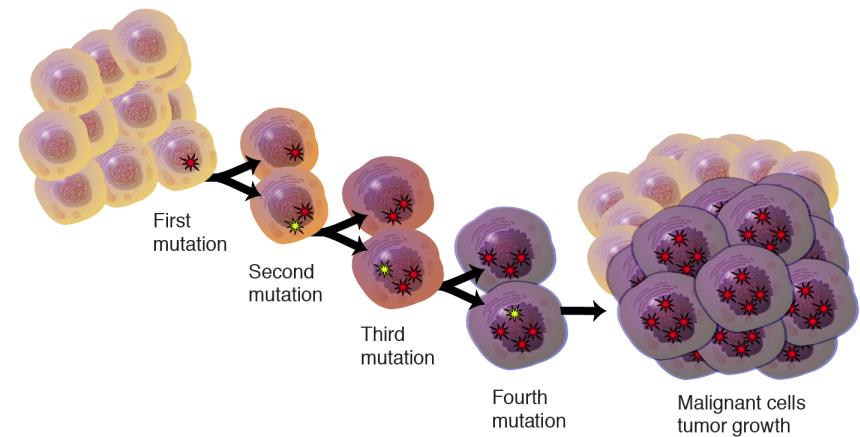
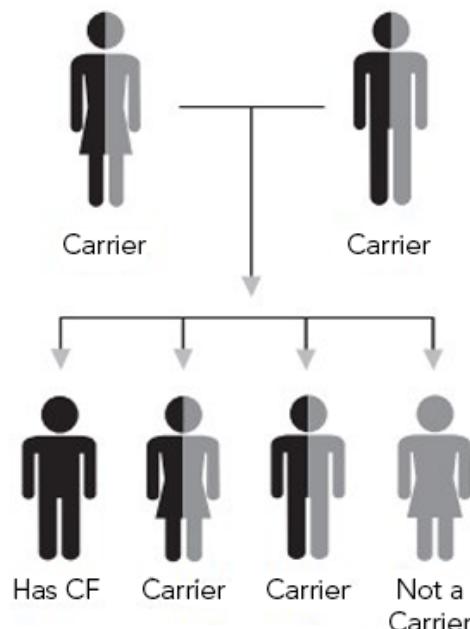
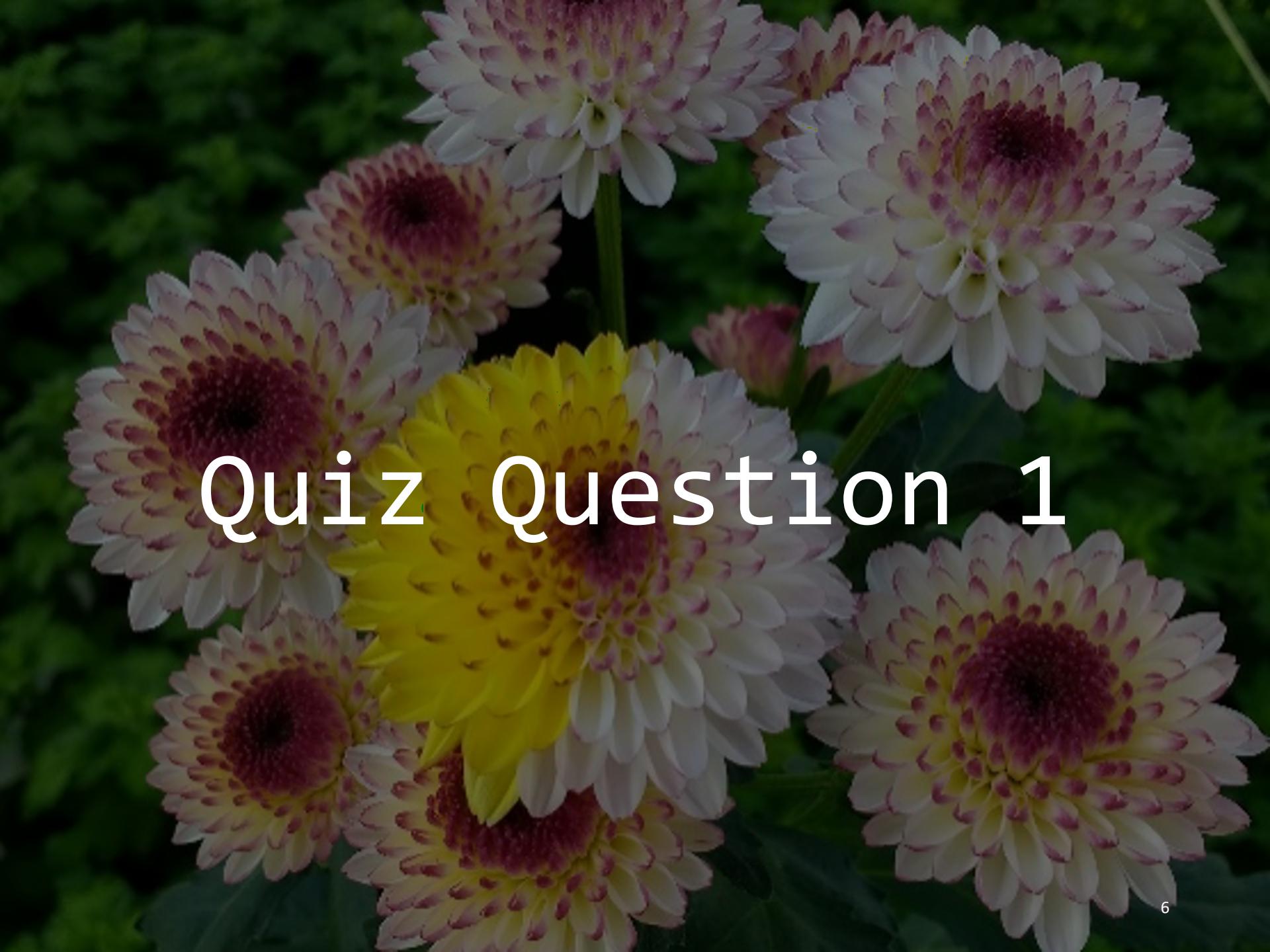


image sources

<https://www.cff.org/>

<https://www.genome.gov/genetics-glossary/Cancer>

A close-up photograph of several chrysanthemum flowers. The flowers are in various stages of bloom, with some showing bright yellow petals and others showing more pinkish-purple or white petals with dark centers. The background is a soft-focus green, suggesting a garden setting.

Quiz Question 1

Detecting mutations

- Phenotypic analysis
- Molecular analysis
 - Sequencing

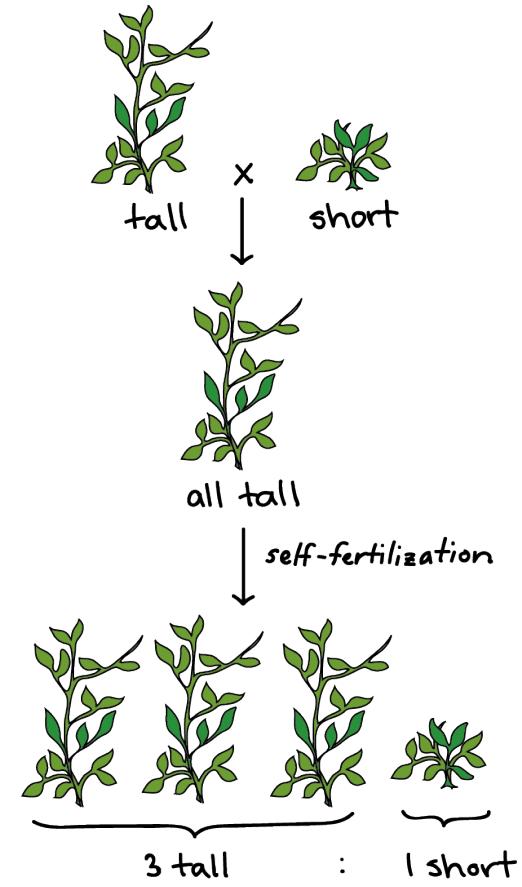
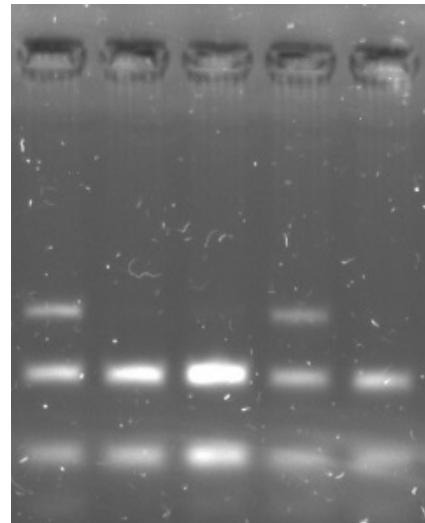
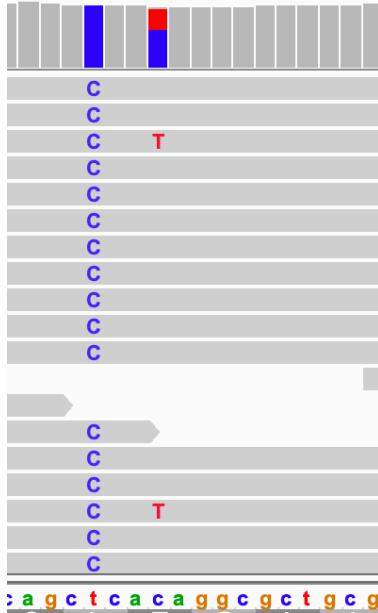


image: <https://www.khanacademy.org>

Small mutations

- Single nucleotide polymorphism (SNP)

ATCATG**A**CCGTCA

ATCATG**T**CCGTCA

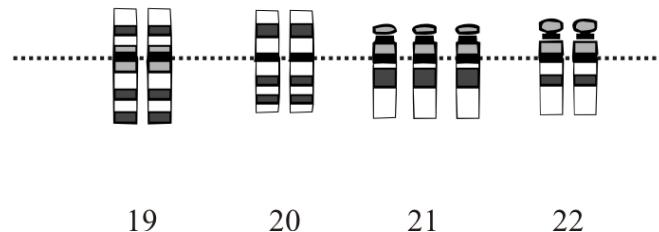
- Insertion/deletion (INDEL)

ATCATG**A**CCGTCA

ATCATG---GTCA

Large mutations

- Structural variance (> 1,000 base pairs)
 - Copy number variation
 - Translocations
 - Inversions
- Chromosomal aberration

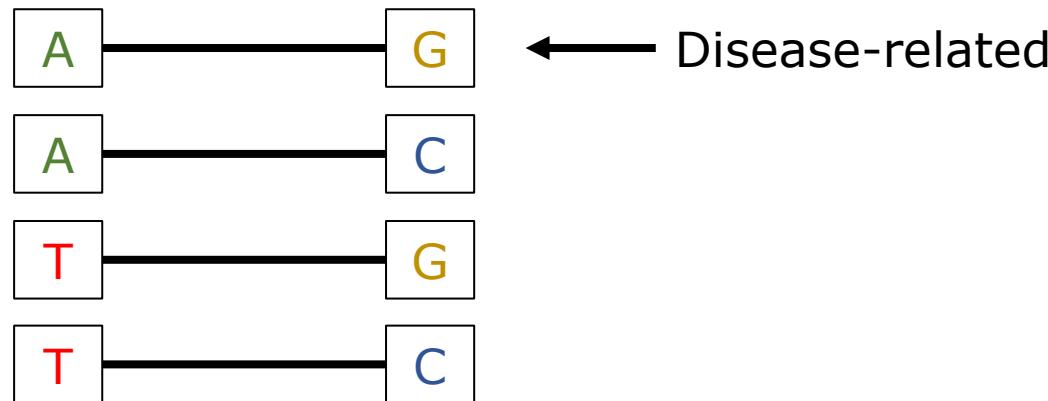


<https://en.wikipedia.org/wiki/Aneuploidy>

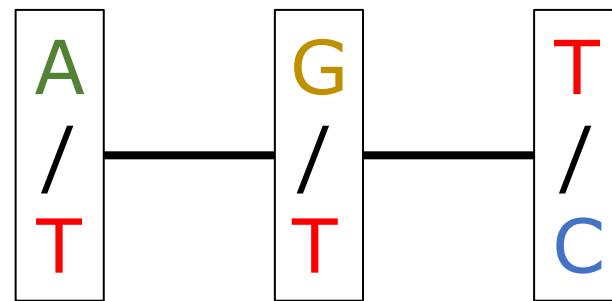


Haplotypes

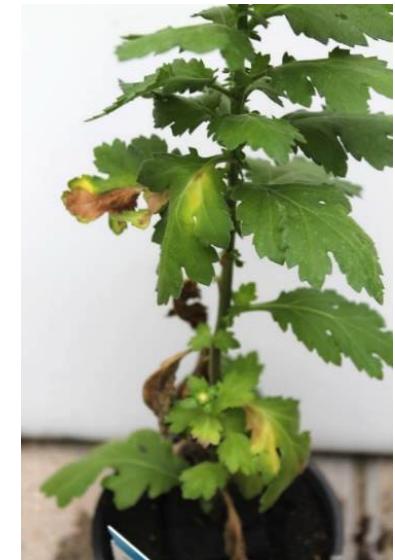
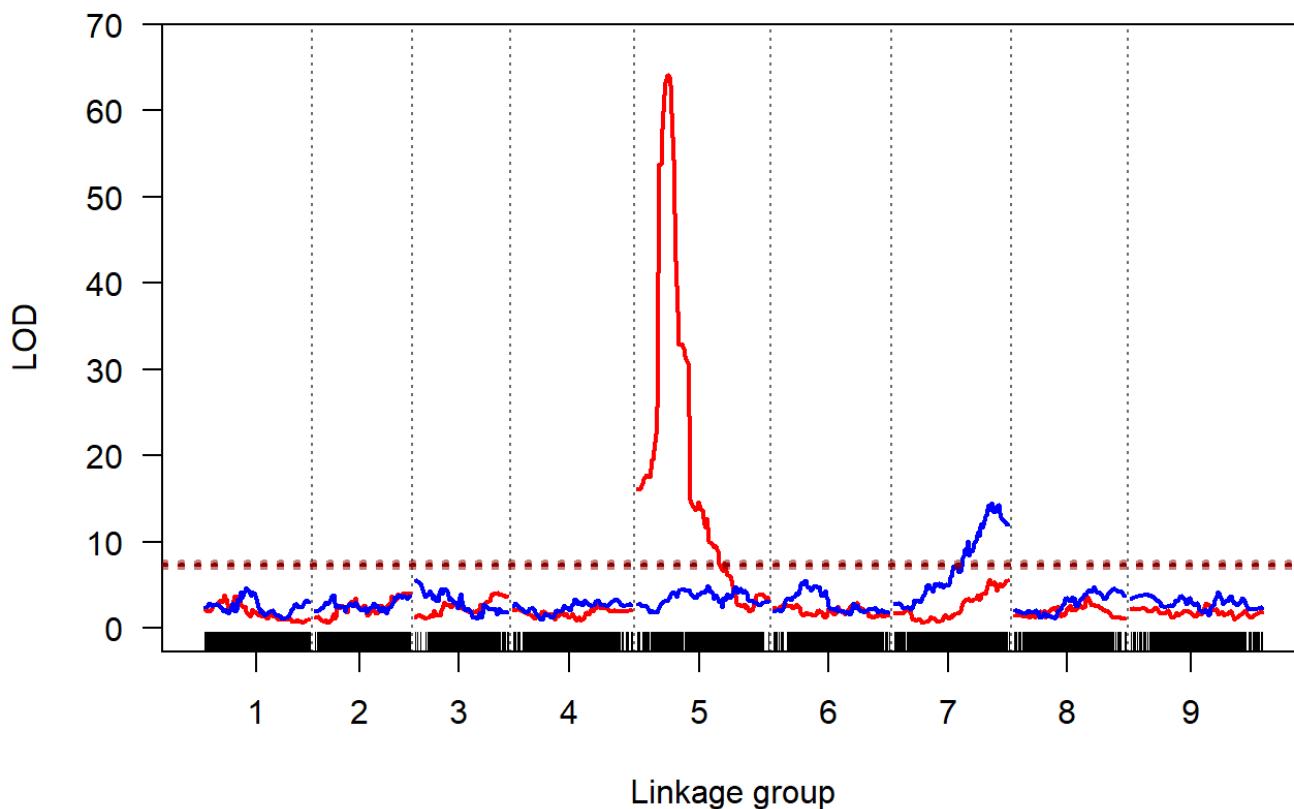
- NGS variants: mostly SNP
- Most SNPs are bi-allelic e.g. [A/T], [G/C]
- Genetic variation is often multi-allelic



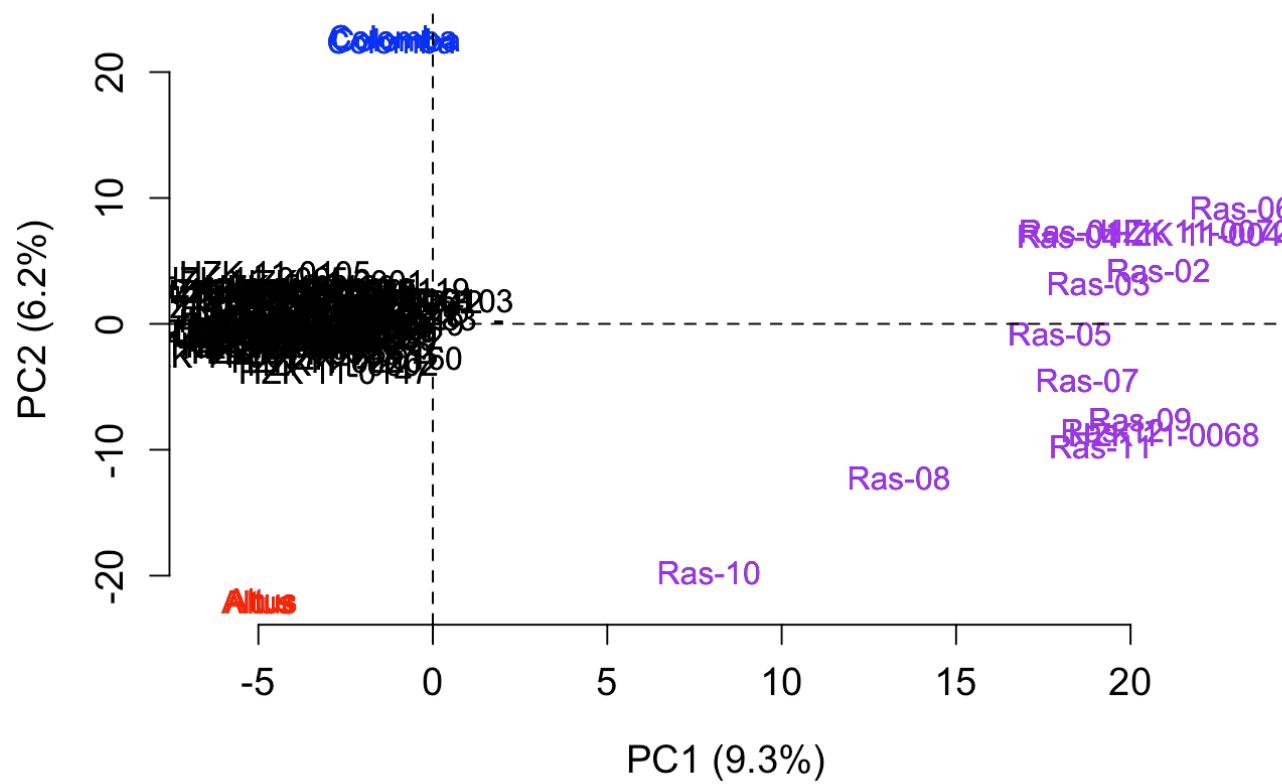
Quiz Question 2



Genetic association



Relatedness



This course

- Inherited (germline) small mutations
- Detection by next generation sequencing (NGS)

