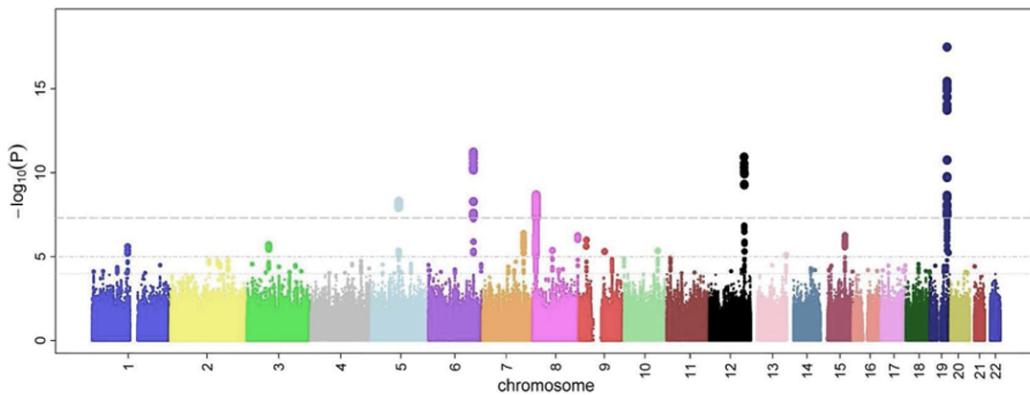


NGS – variant analysis

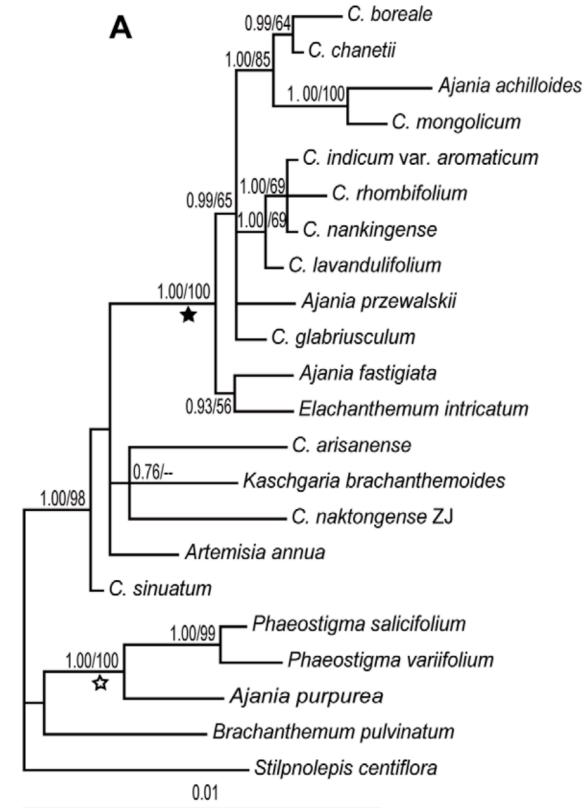
Introduction to variant analysis

Why study variants?

- Find causes for phenotypic variation
- Understand relatedness

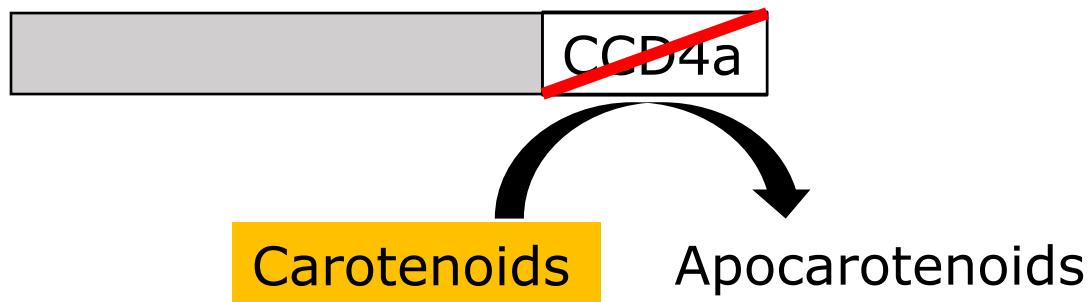


https://en.wikipedia.org/wiki/Genome-wide_association_study



Mutation

Change in DNA sequence



Mutations - causes

- Repair mistakes
- Unbalanced cell division
- Transposable elements



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

Mutations - types

- cells – somatic mutation
- inherited – germline mutation

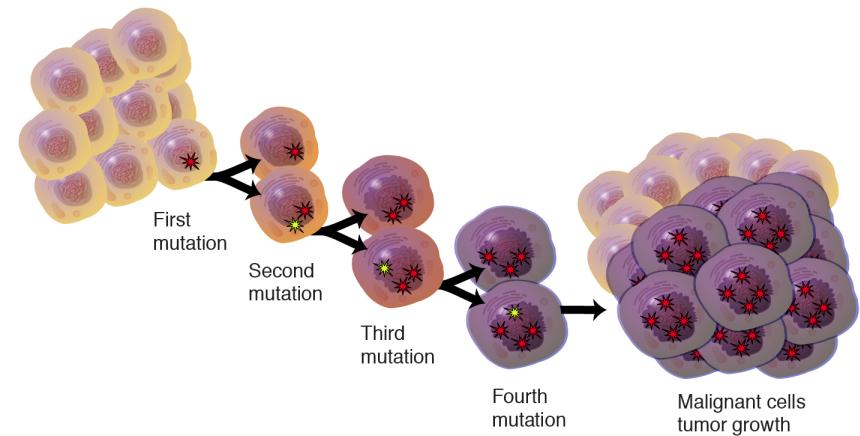
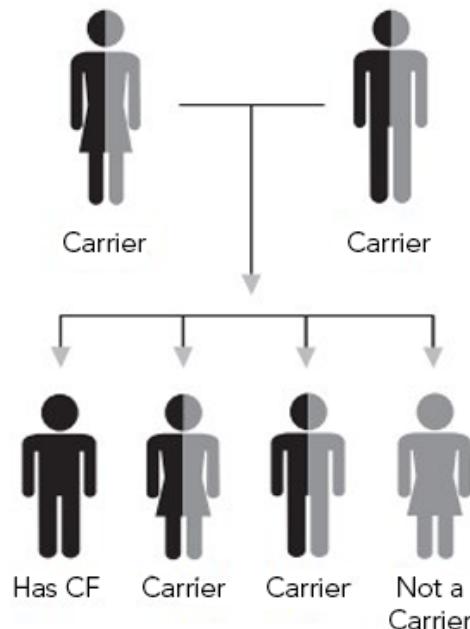
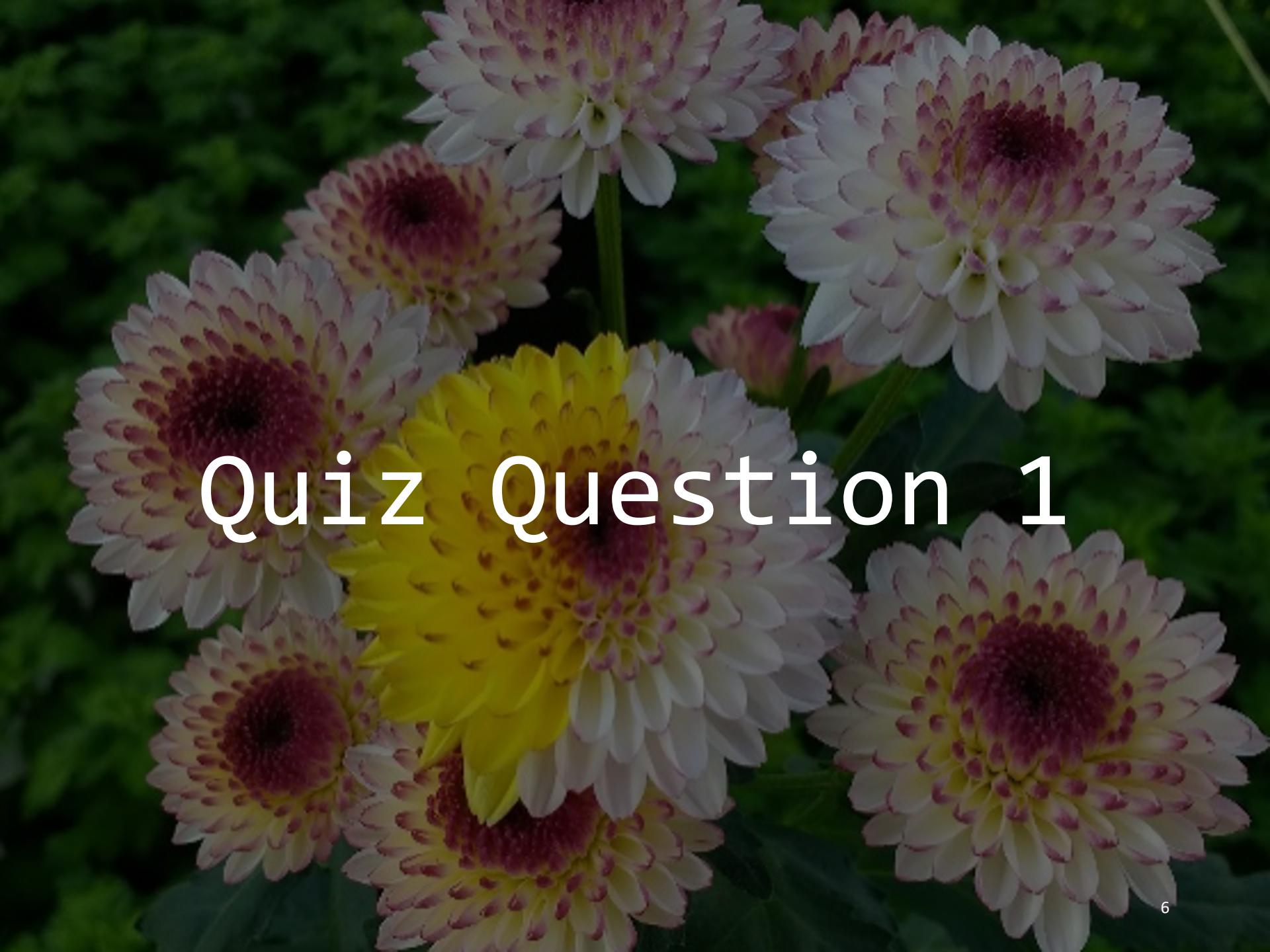


image sources

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

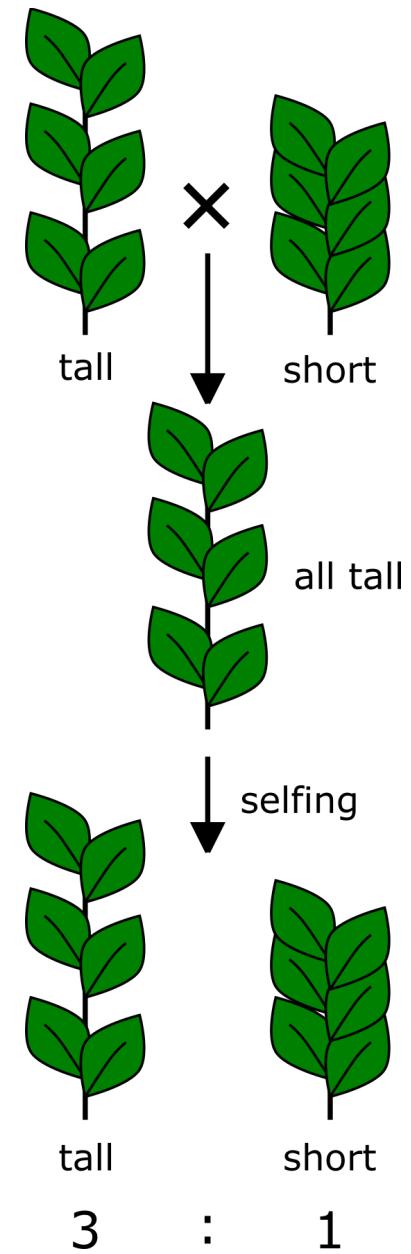
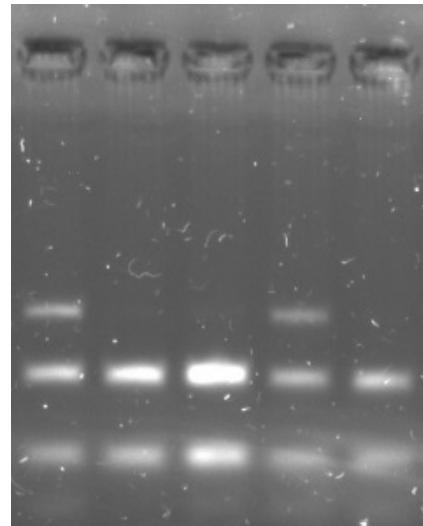
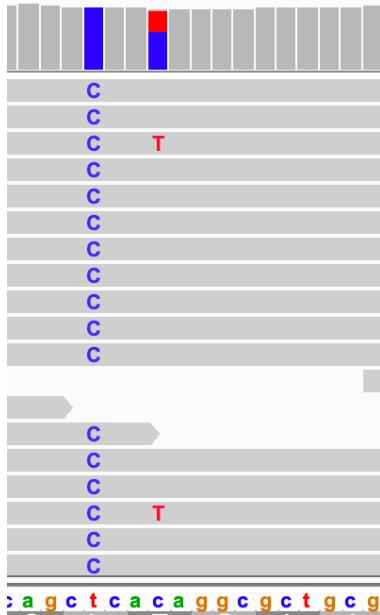
<https://www.genome.gov/genetics-glossary/Cancer>

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 white or purple edges. The centers of the flowers are dark purple and textured. The background is a soft-focus green, suggesting a garden setting.

Quiz Question 1

Detecting variants

- Phenotypic analysis
- Molecular analysis
 - Sequencing



Small variants

- Single nucleotide polymorphism (SNP)

ATCATG**A**CCGTCA

ATCATG**T**CCGTCA

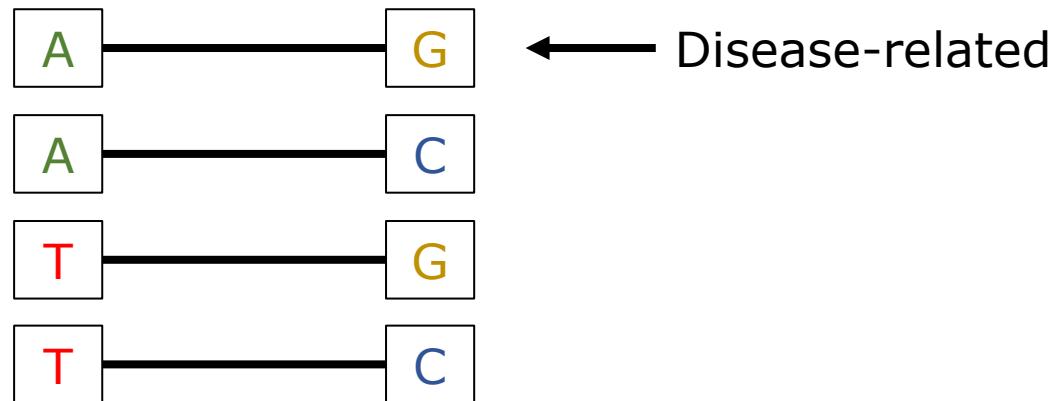
- Insertion/deletion (INDEL)

ATCATG**A**CCGTCA

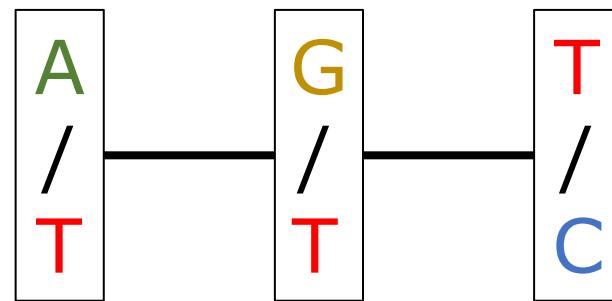
ATCATG---GTCA

Haplotypes

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

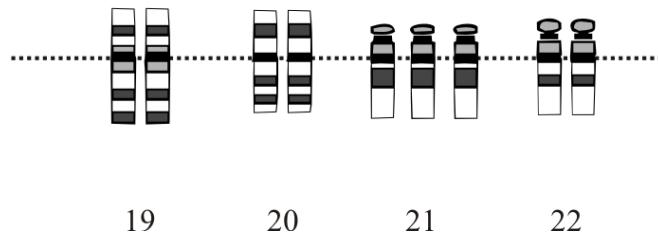


Quiz Question 2



Large variants

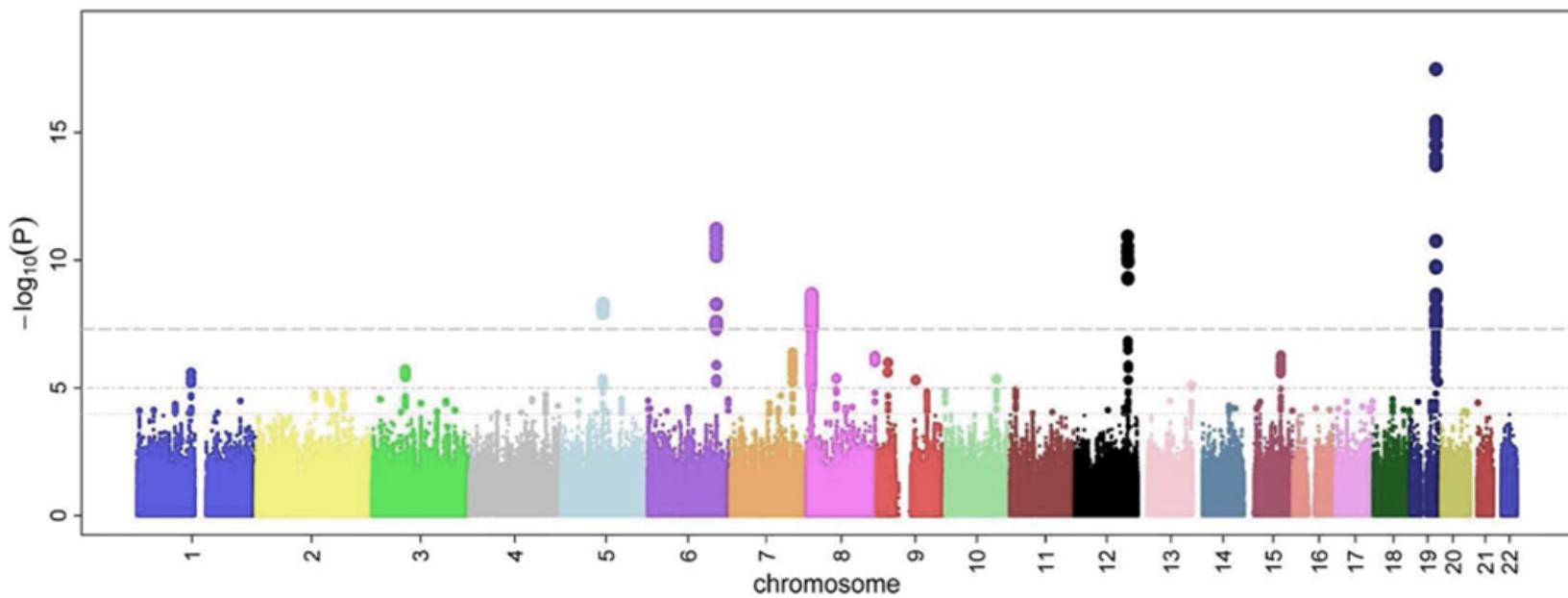
- Structural variance (> 1,000 base pairs)
 - Copy number variation
 - Translocations
 - Inversions
 - Deletions/insertions
- Chromosomal aberration



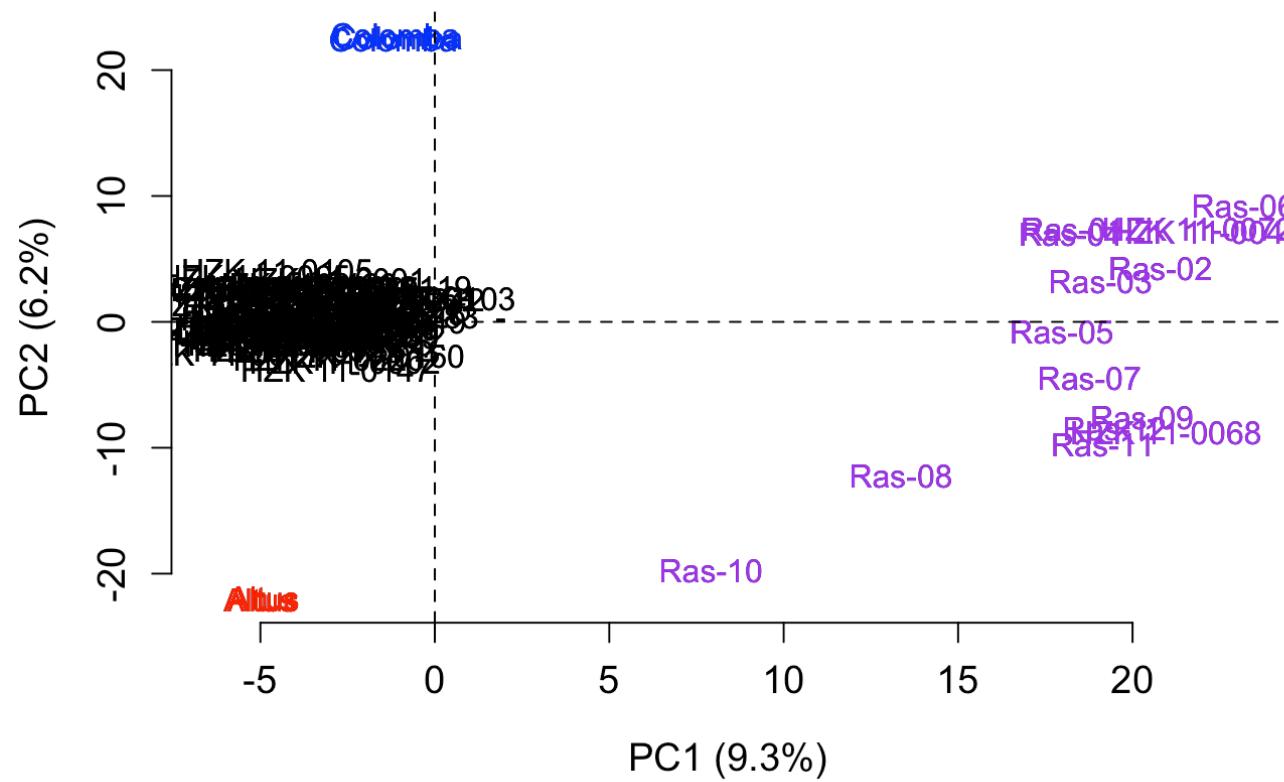
<https://en.wikipedia.org/wiki/Aneuploidy>



Genetic association



Relatedness



This course

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

