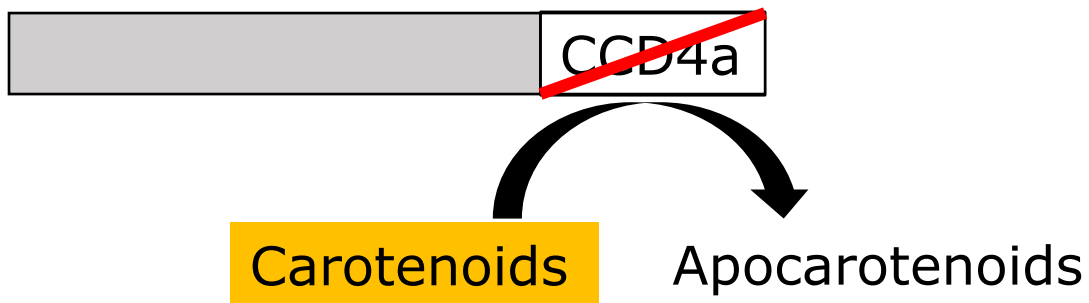


# NGS – variant analysis

Introduction to variant analysis

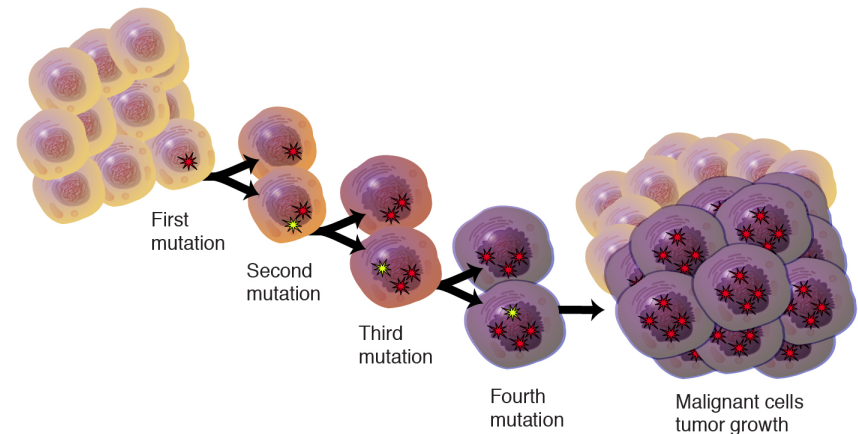
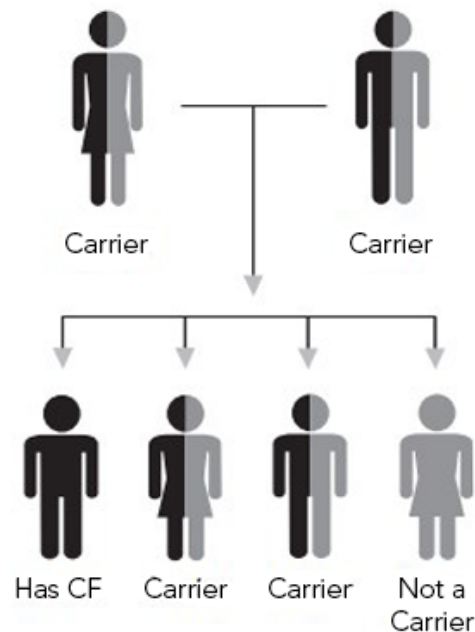
# Mutation

Change in DNA sequence



# Genomic variation

- inherited – germline mutation
- cells – somatic mutation



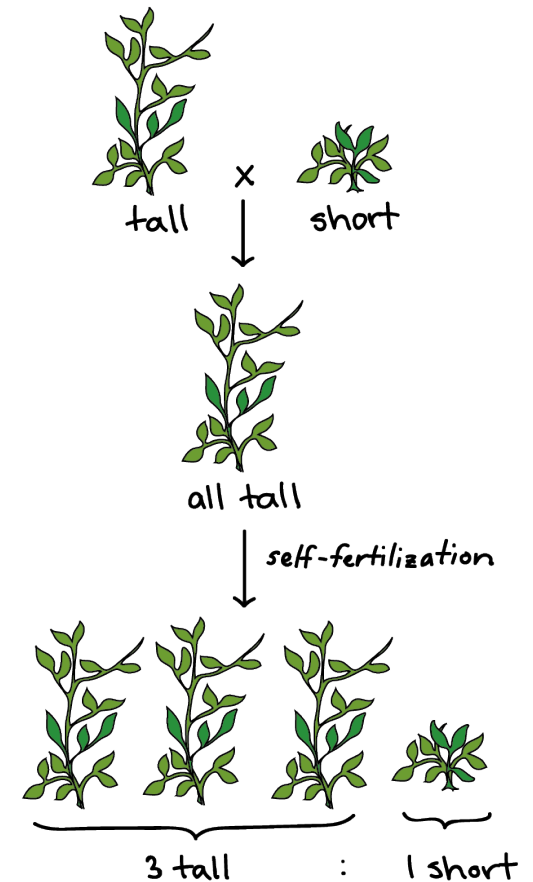
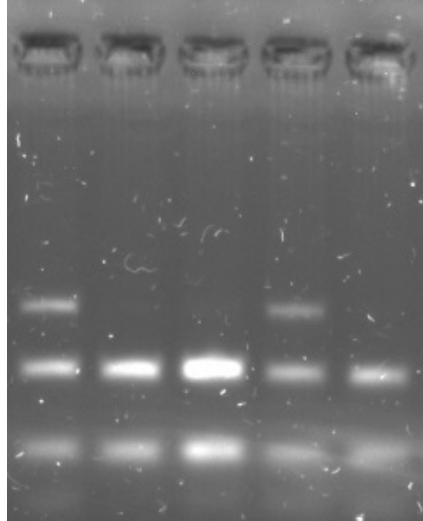
# Question 1

What kind of mutation has caused the flower to turn yellow?

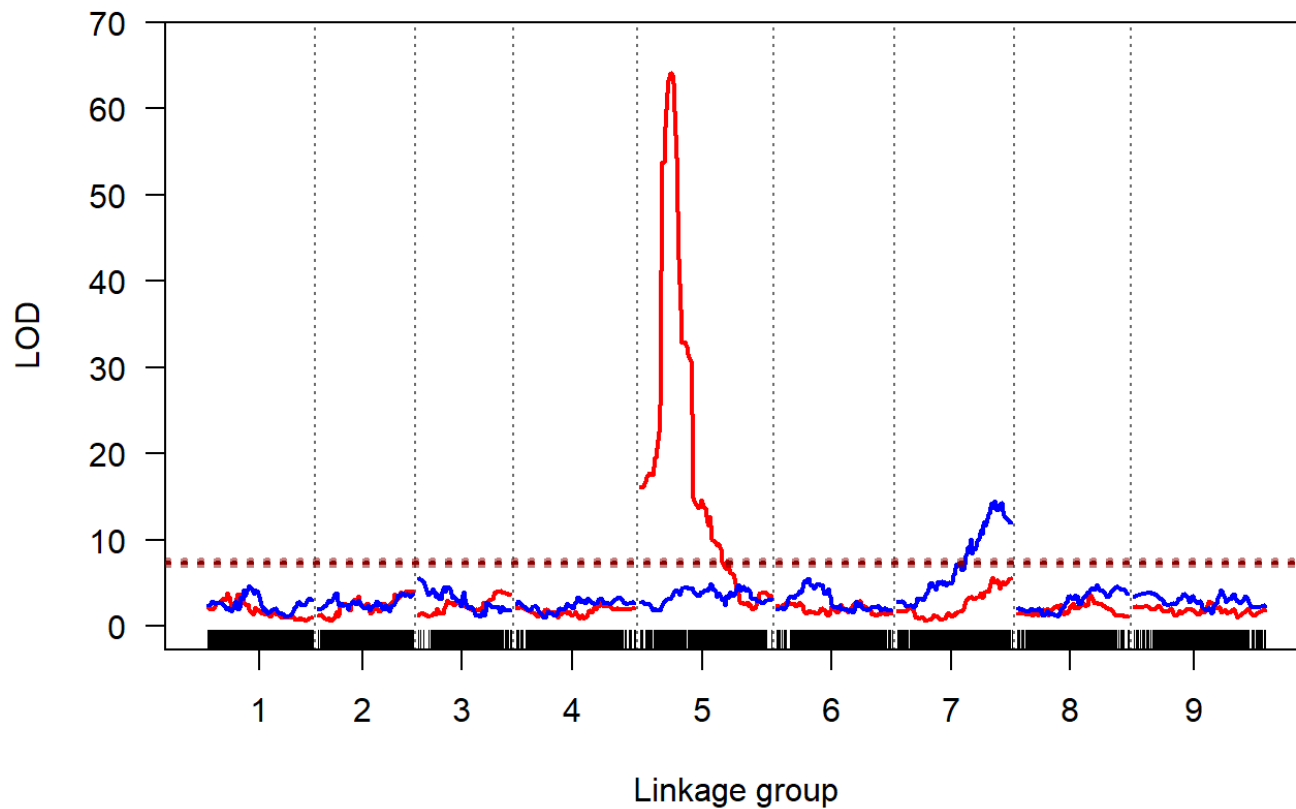
- A. Somatic mutation
- B. Germline mutation
- C. Both

# Detecting mutations

- Phenotypic analysis
- Molecular analysis (DNA)



# Genetic association



# Mutations

- Single nucleotide polymorphism (SNP)
- Insertion/deletion (INDEL)
- Structural variance ( $> 1,000$  base pairs)
  - Copy number variation
  - Transversions
  - Inversions
- Chromosomal aberration



# This course

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



