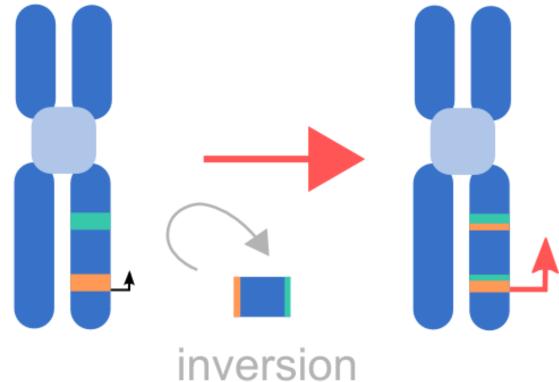
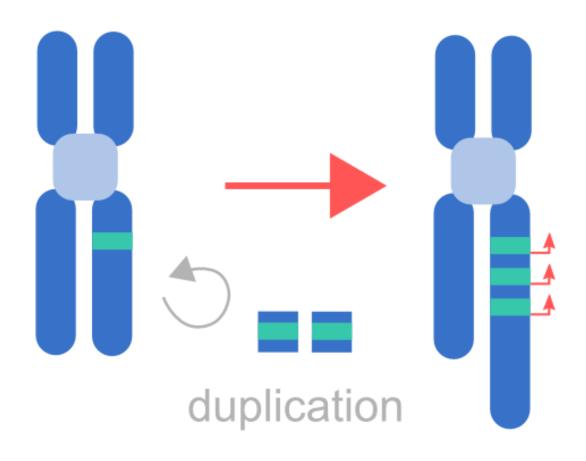
THE FUNCTIONAL CONSEQUENCES OF STRUCTURAL VARIATION

Oncogenic fusion

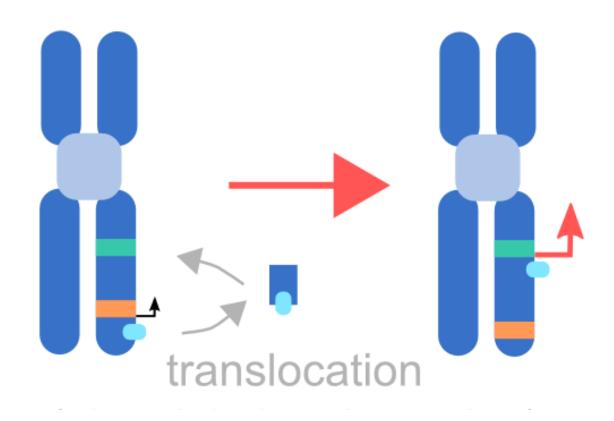


(also deletion, translocation)

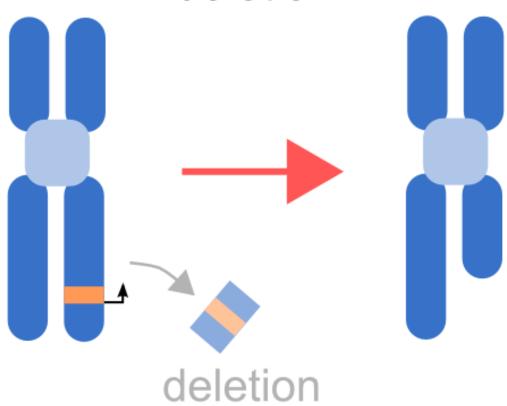
Oncogene amplification



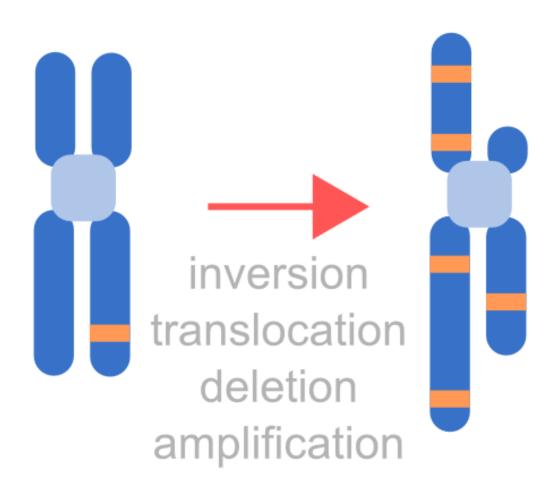
Enhancer hijacking



Tumour suppressor deletion



Genomic instability



Examples of methods for predicting function

- Oncogenic fusions: GRASS
- Amplification/deletion: GISTIC (multisample)
- Enhancer hijacking: ?
- Genomic instability: Complex Arm Aberration Index (CAAI) and Genomic Index (GI)

EXERCISE 4