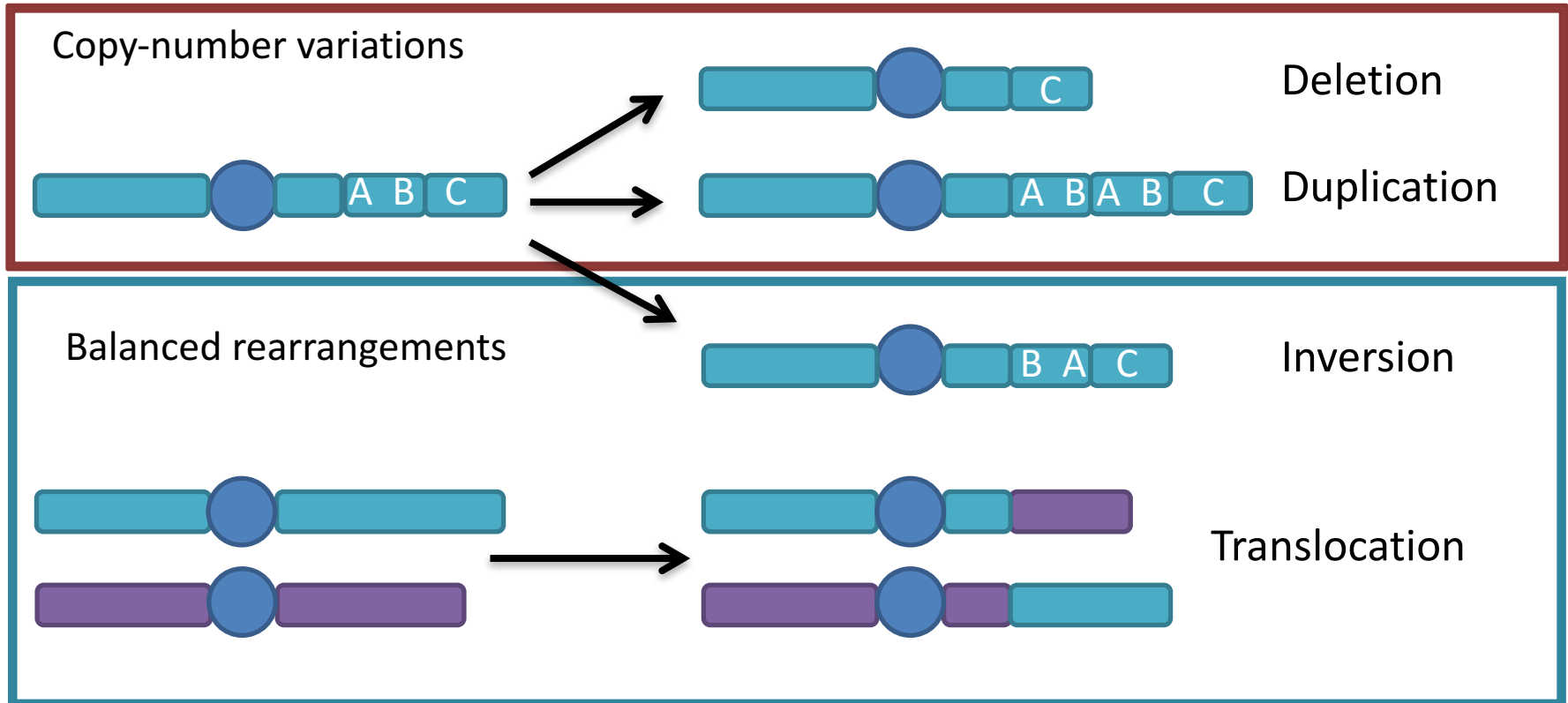
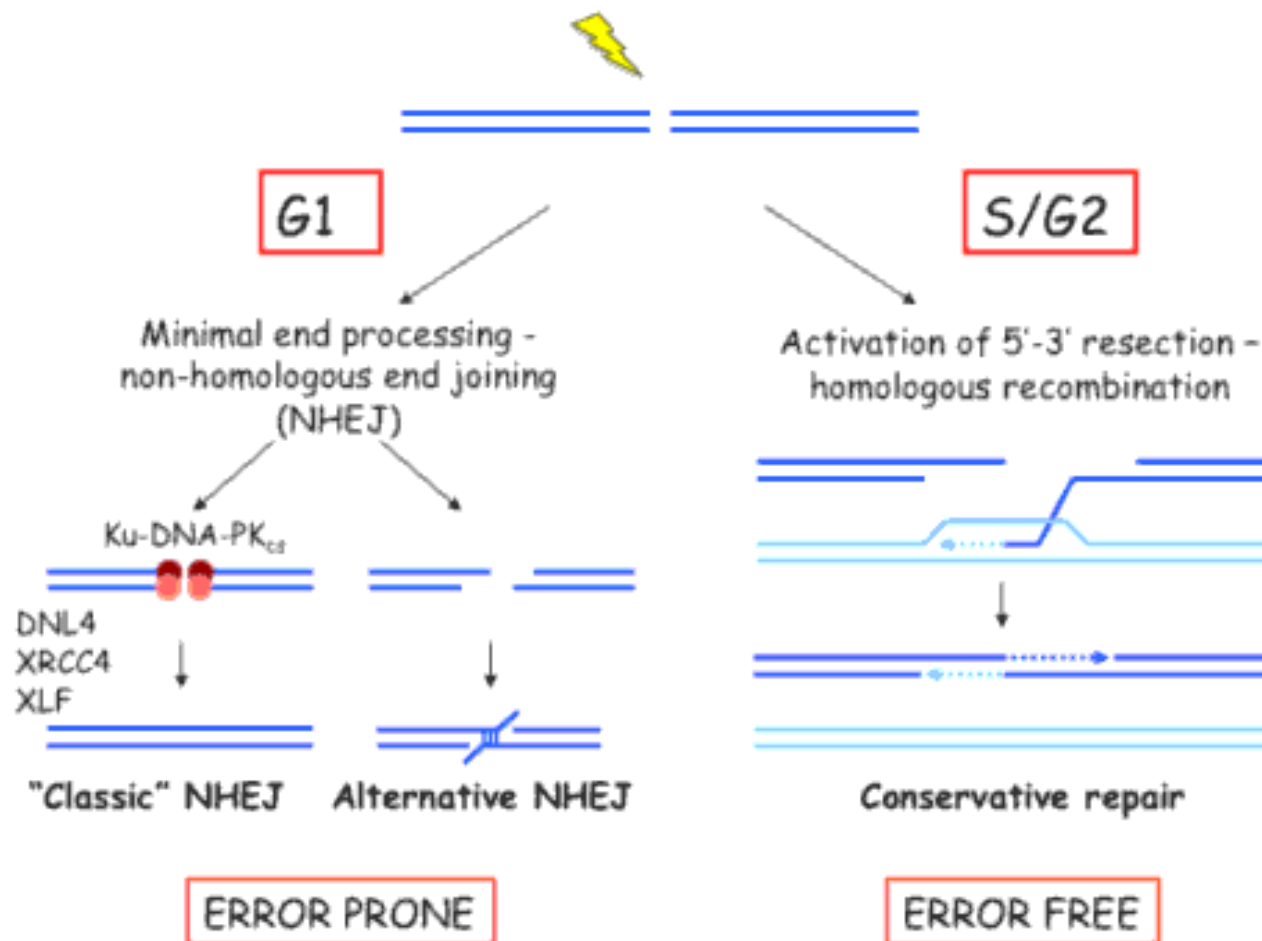


# Somatic structural variation (in short)

# Balanced versus unbalanced



## Two mechanisms to repair DSBs

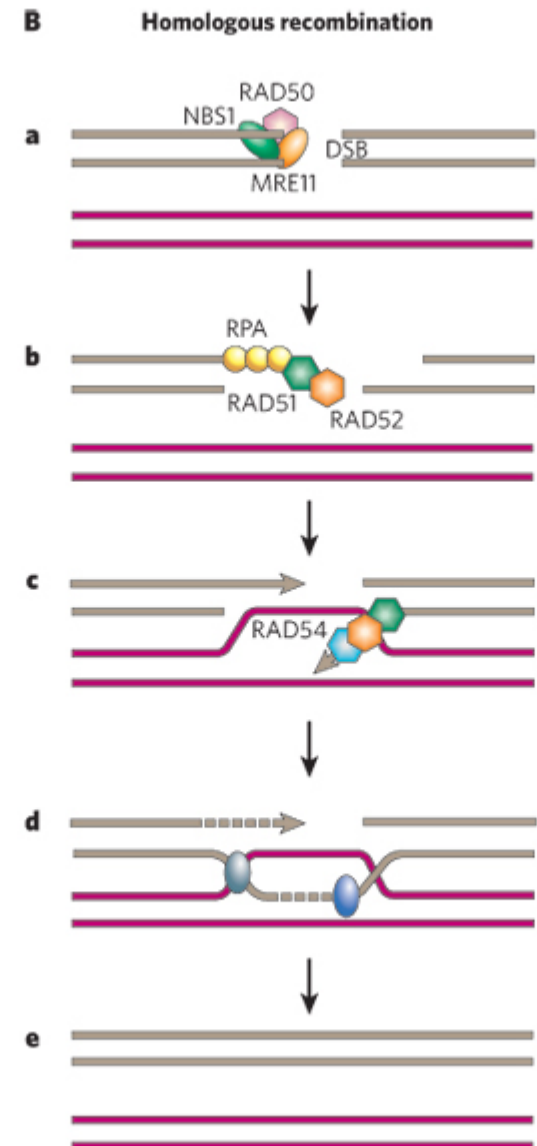


# Repair mechanisms underlying SVs

- HR: homologous recombination
- NHEJ: non-homologous end joining
- NAHR: non-allelic homologous recombination
- MMEJ: microhomology-mediated end joining
- FoSTes: fork stalling and template switching
- MMBIR: microhomology-mediated break-induced replication

# NAHR: non-allelic homologous recombination

- Mostly results in segmental duplications
- Common repair mechanism in recurrent SVs
- Relatively error free around break



From the following article:

[Chromatin dynamics and the preservation of genetic information](#)

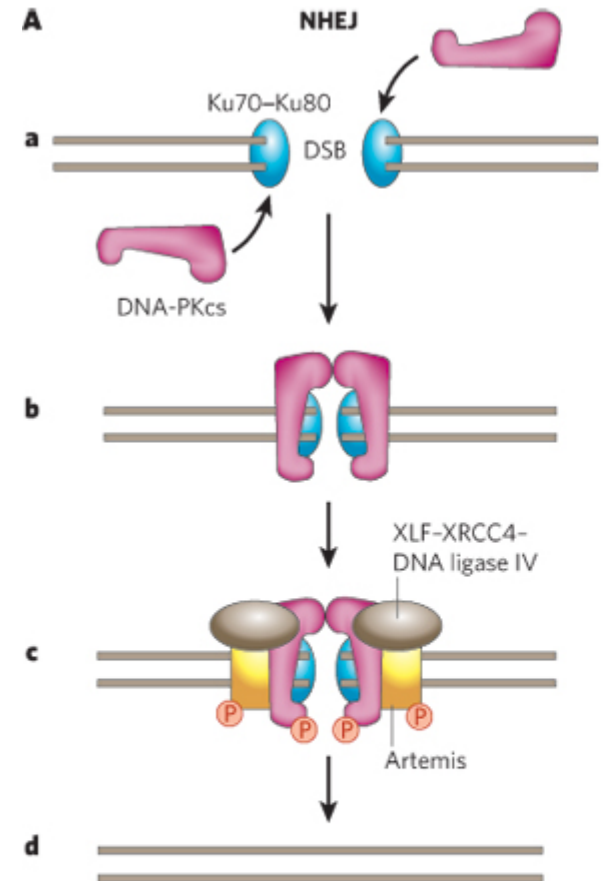
Jessica A. Downs, Michel C. Nussenzweig & André Nussenzweig

*Nature* **447**, 951-958 (21 June 2007)

doi:10.1038/nature05980

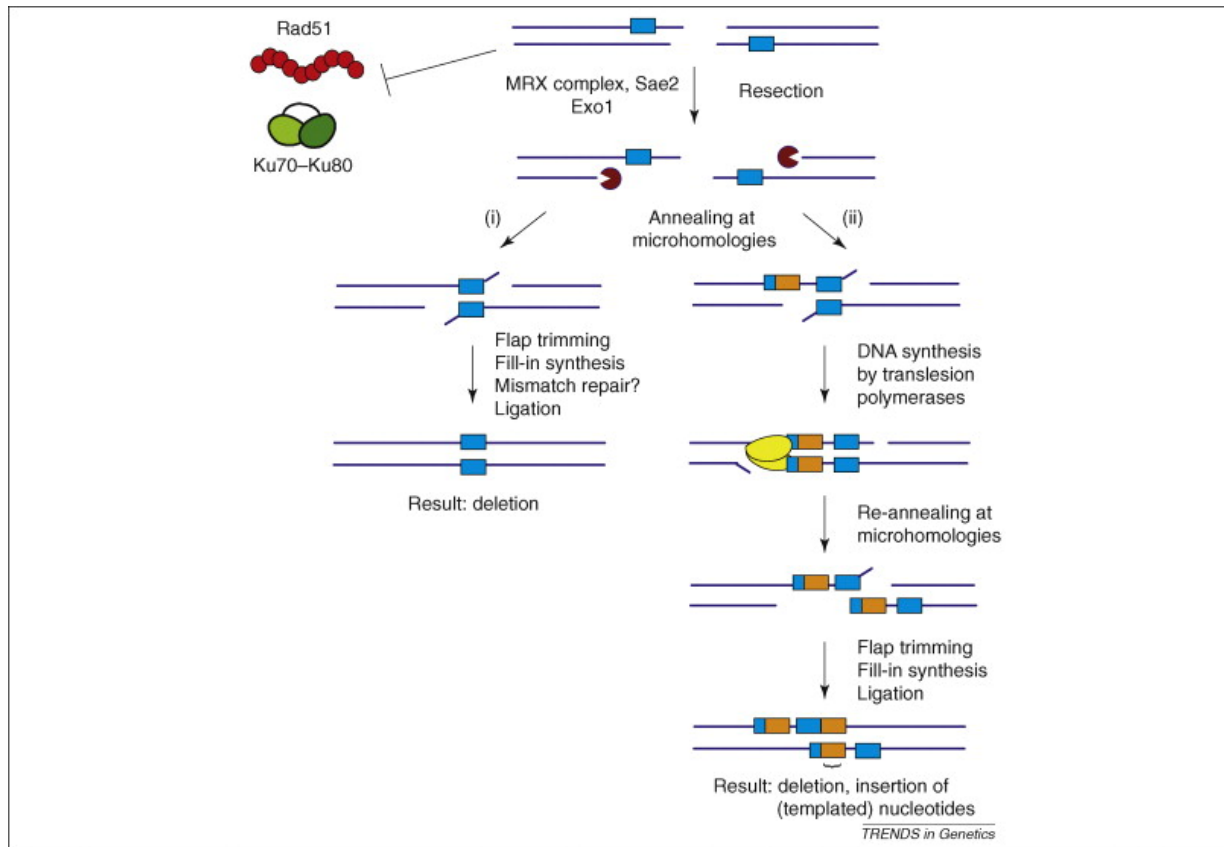
# NHEJ: non-homologous end joining

- Preferred method of break repair
- Can result in small insertions and deletions at breakpoint



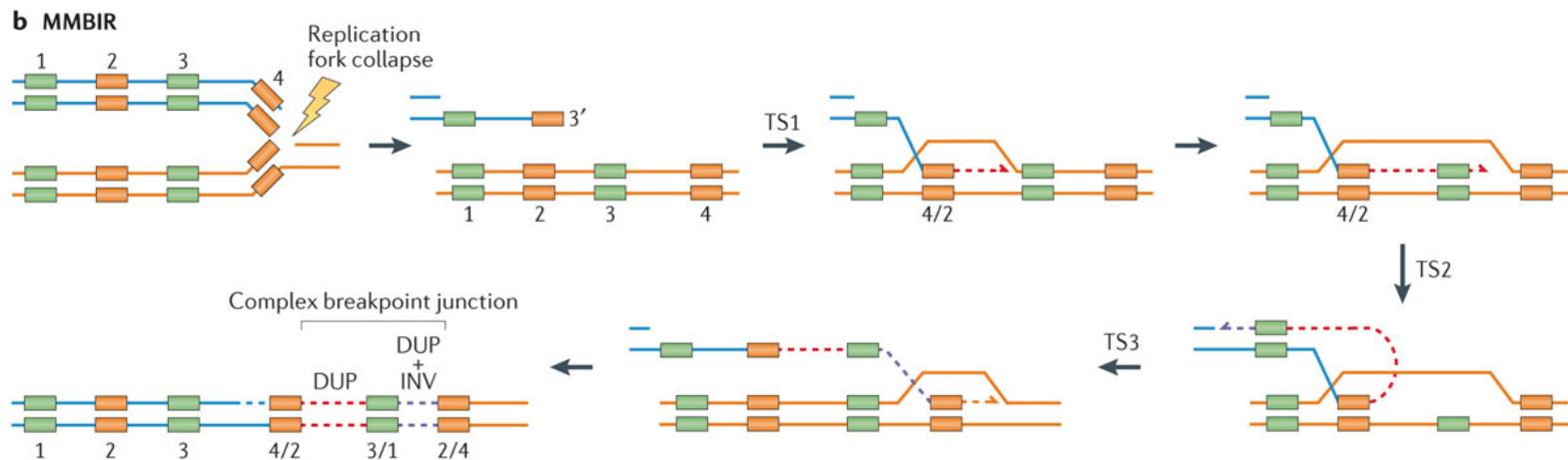
# MMEJ: microhomology-mediated end joining

- Backup to NHEJ
- Similar but more error prone than NHEJ
- Commonly has small deletions at breakpoints



# FoSTes: fork stalling and template switching and MMBIR: microhomology-mediated break- induced replication

- Generates complex rearrangements
- Causes microhomology at breakpoints

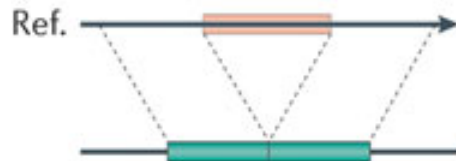




## **EXERCISE 1.1**

# Common types of structural variation

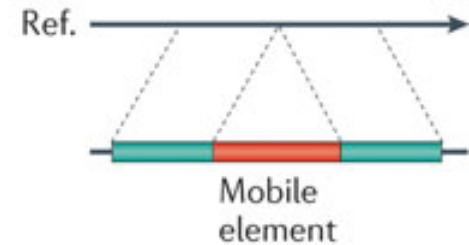
**Deletion**



**Novel sequence insertion**



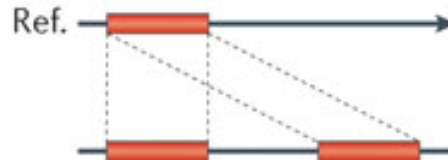
**Mobile-element insertion**



**Tandem duplication**



**Interspersed duplication**



**Inversion**



**Translocation**



## **EXERCISE 1.2**