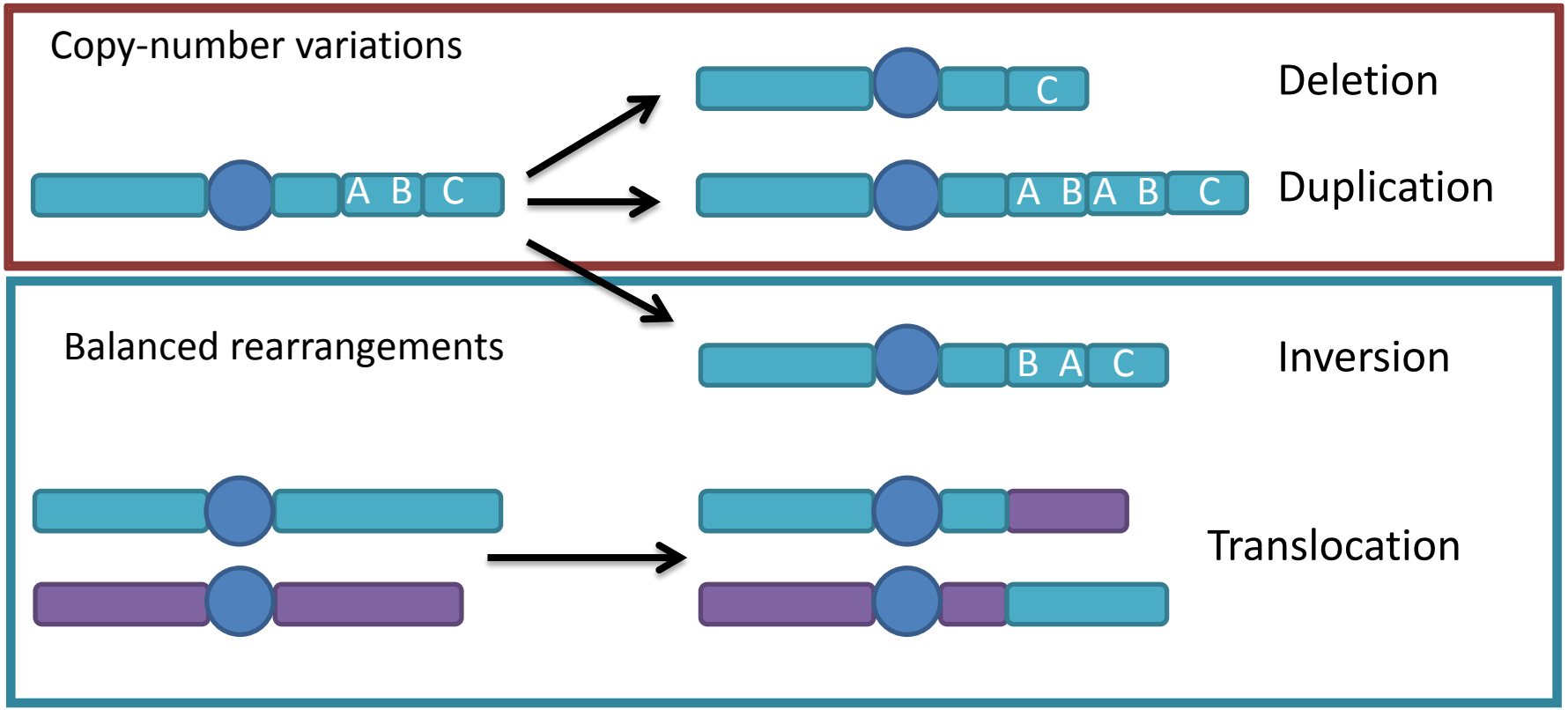
The biology behind structural variation (in short)

Balanced versus unbalanced

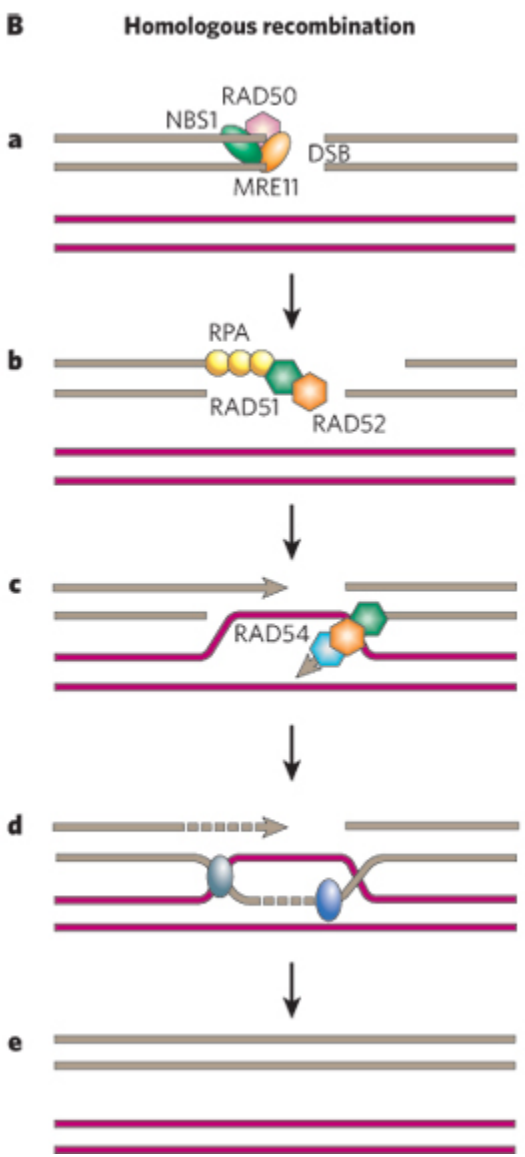


Repair mechanisms underlying SVs

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

NAHR: non-allelic homologous recombination

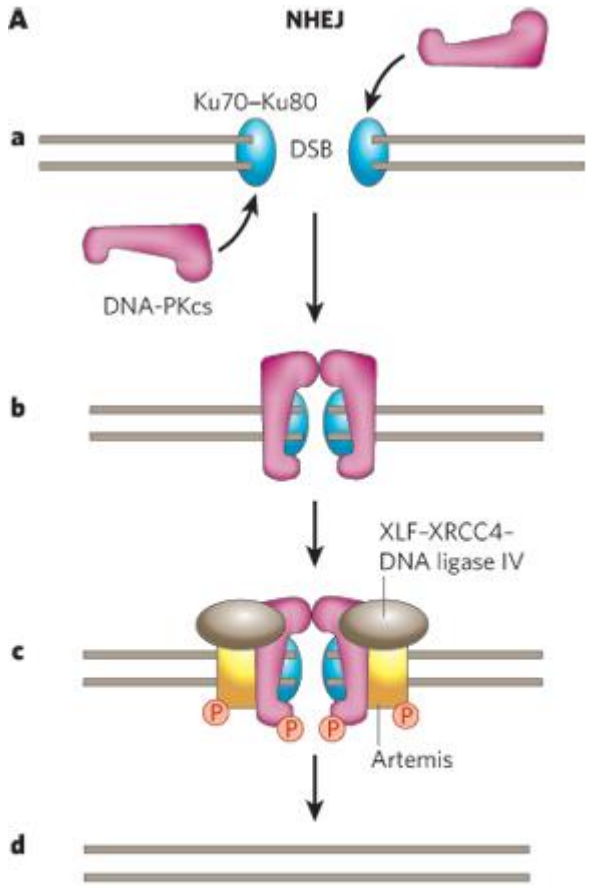
- Mostly results in segmental duplications
- Common repair mechanism in recurrent SVs
- Relatively error free around break



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

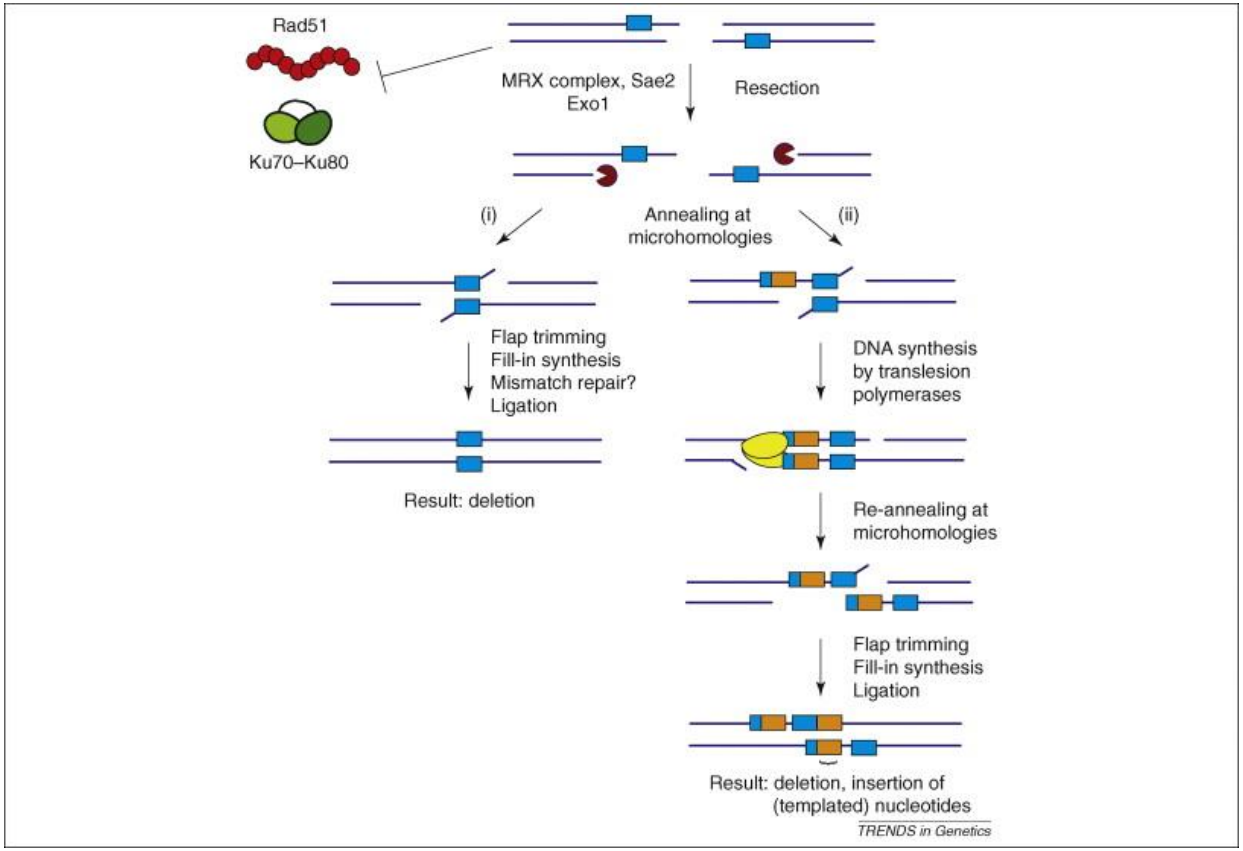
NHEJ: non-homologous end joining

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



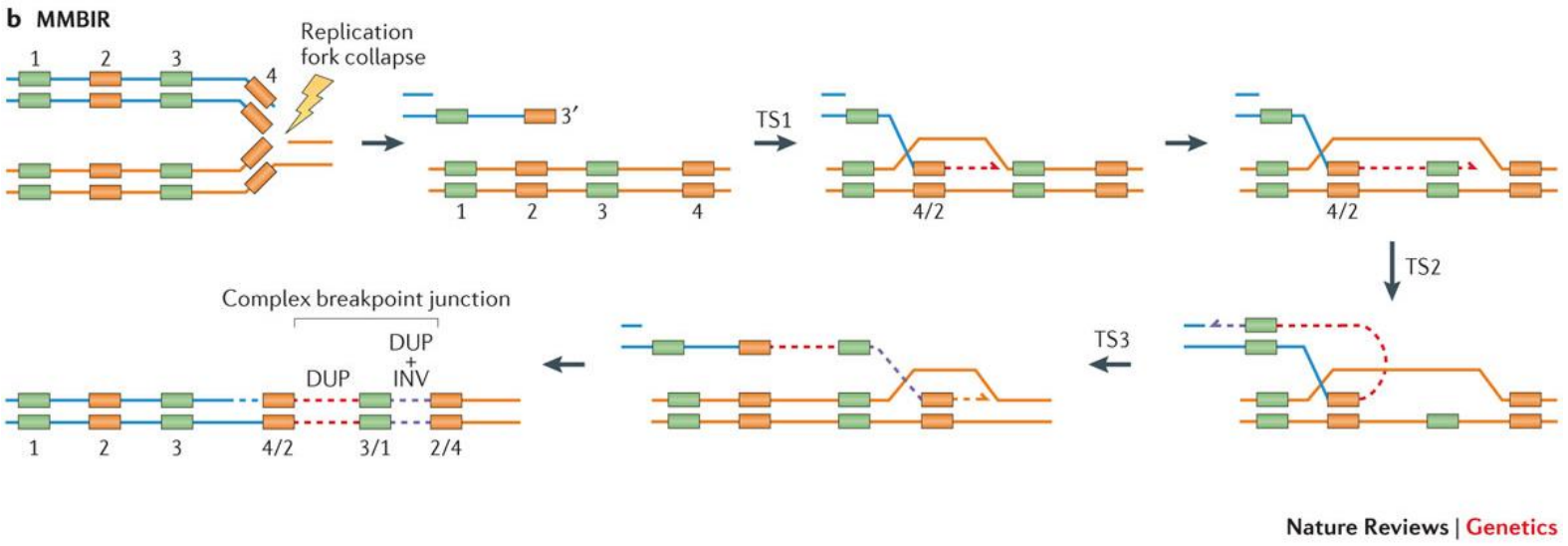
MMEJ: microhomology-mediated end joining

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



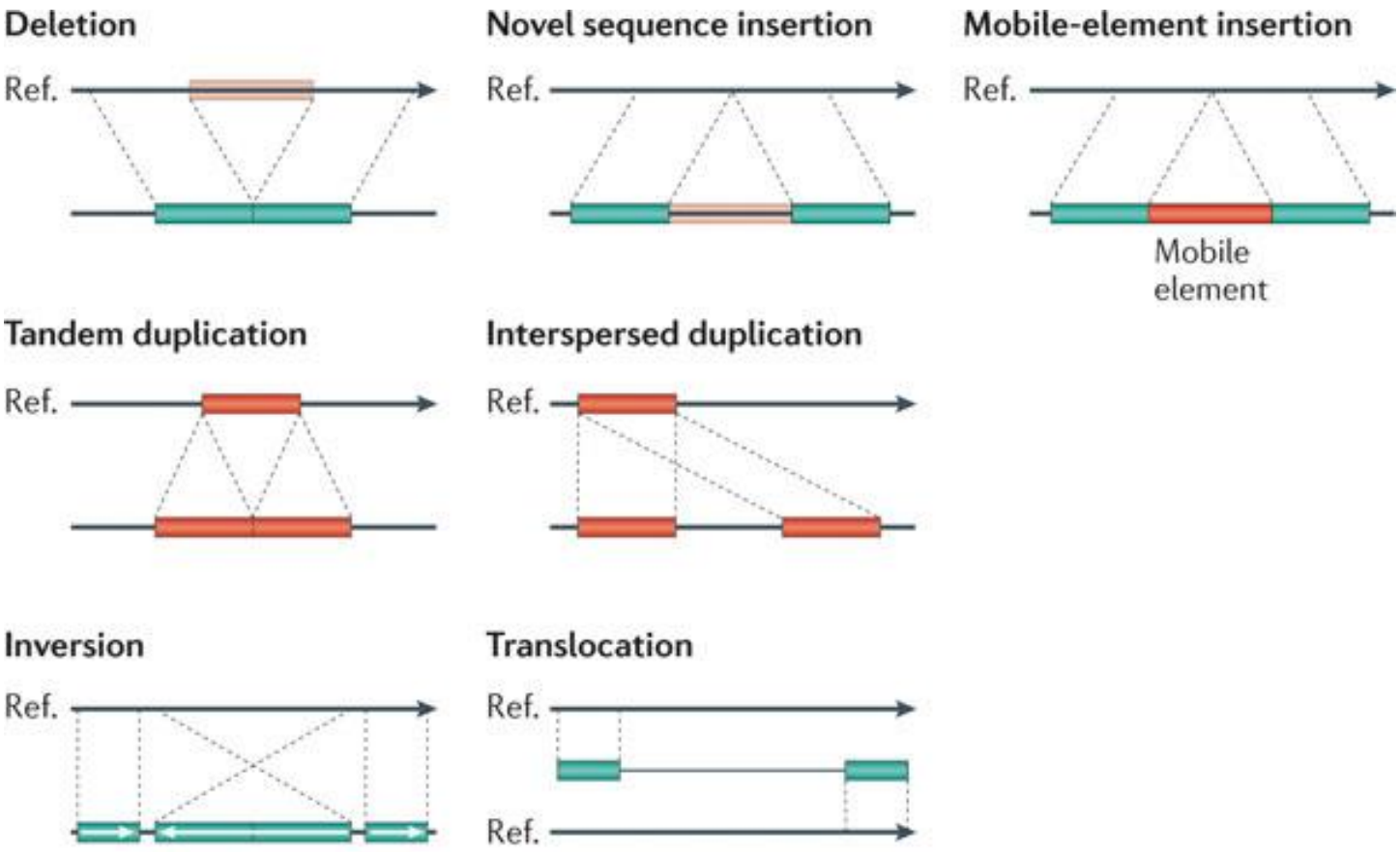
FoSTes: fork stalling and template switching and MMBIR: microhomology-mediated break-induced replication

- Generates complex rearrangements
- Causes microhomology at breakpoints



EXERCISE 1.1

Common types of structural variation



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EXERCISE 1.2