

Aa

Sample genome

Deletion



Reference genome

**b**

Sample genome

Insertion



Reference genome

**c**

Sample genome

Duplication



Reference genome

**d**

Sample genome

Inversion



Reference genome

**e**

Sample genome

Translocation



Reference genome



Normal read pair



Read depth



Splitread

Abnormal distance
(intrachromosomal)

Unmapped read in a pair

Abnormal read
orientationAbnormal distance
(interchromosomal)