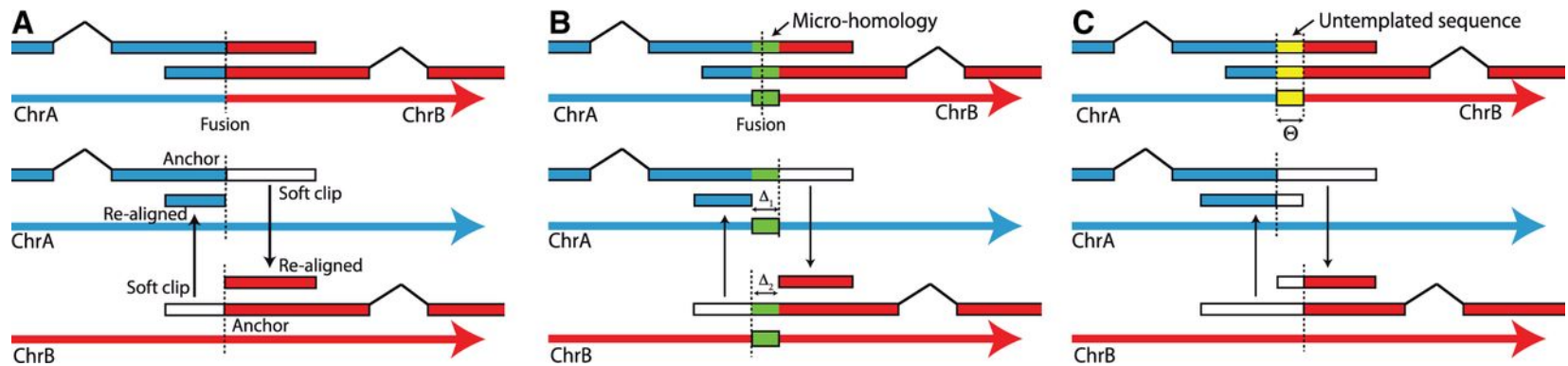


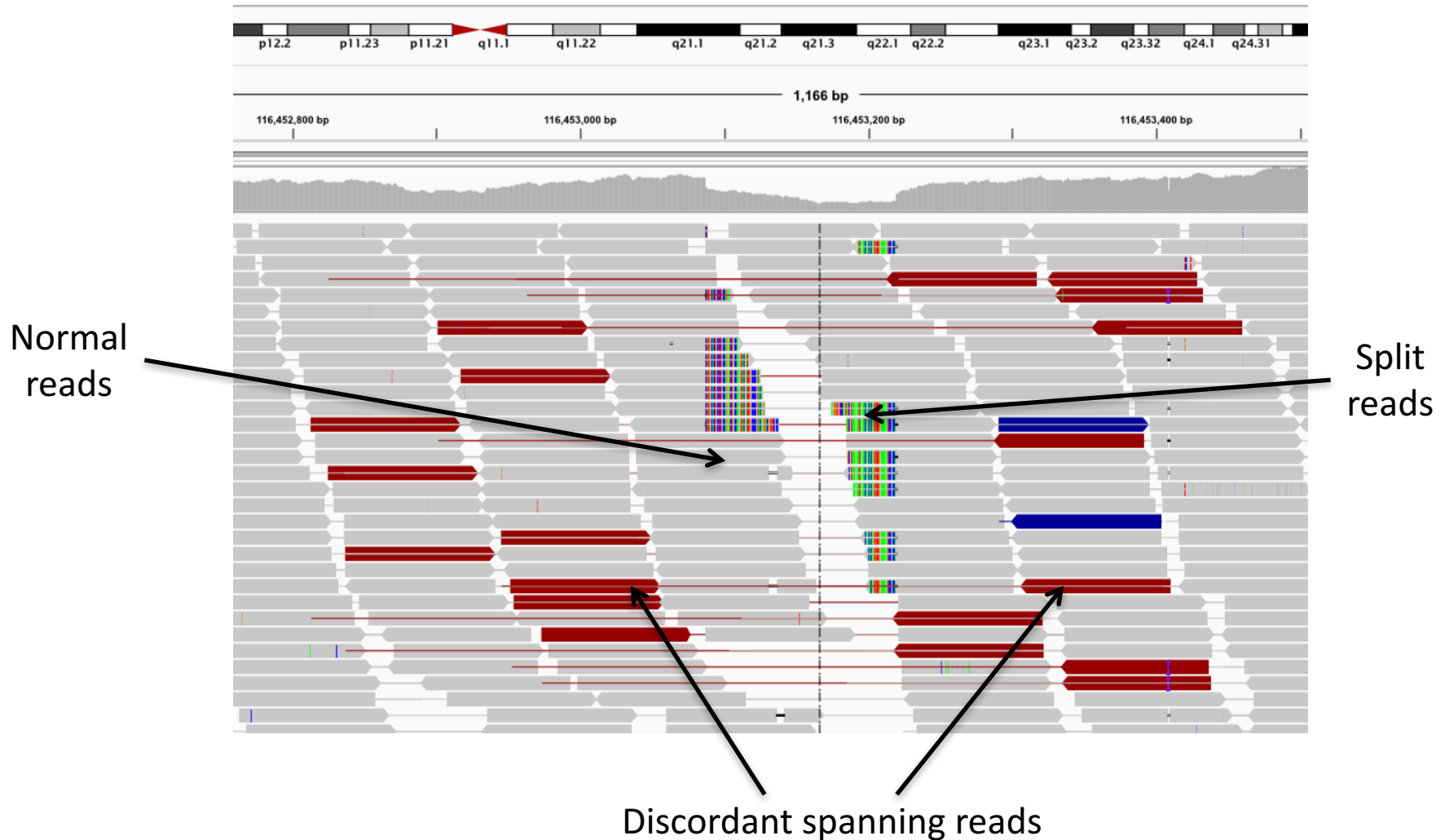
METHODS FOR CALLING SVS

Reads overlapping the breakpoint

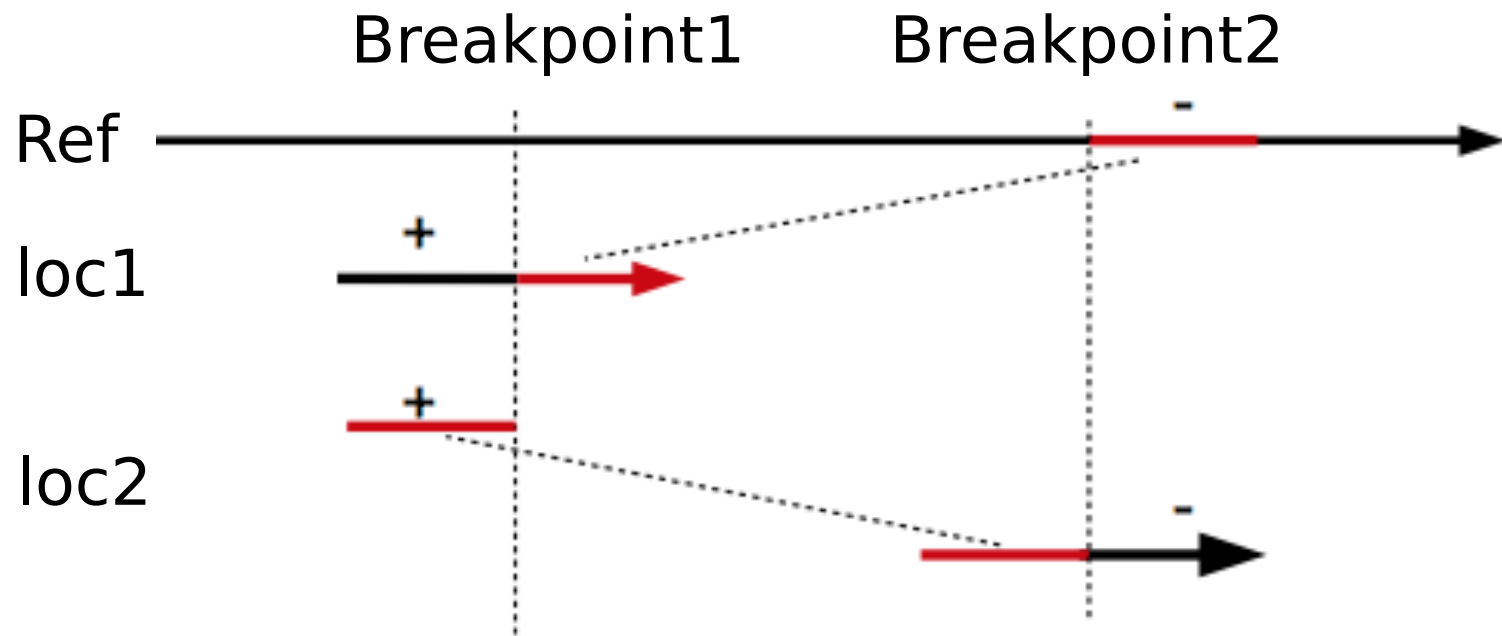


Jan Schröder et al. *Bioinformatics* 2014;30:1064-1072

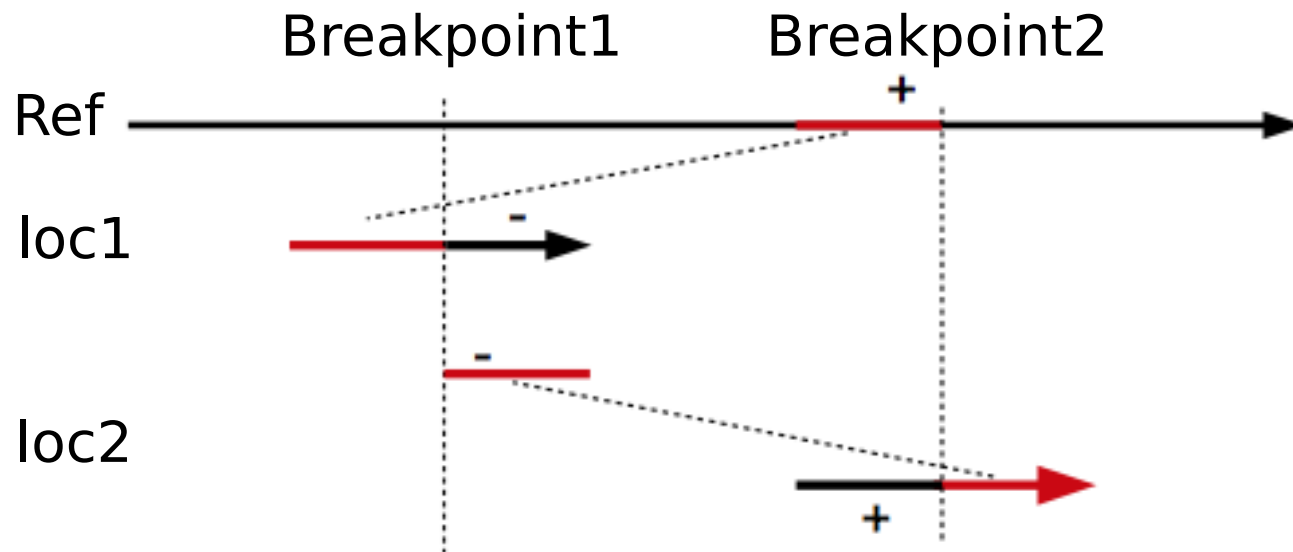
Reads around the breakpoint



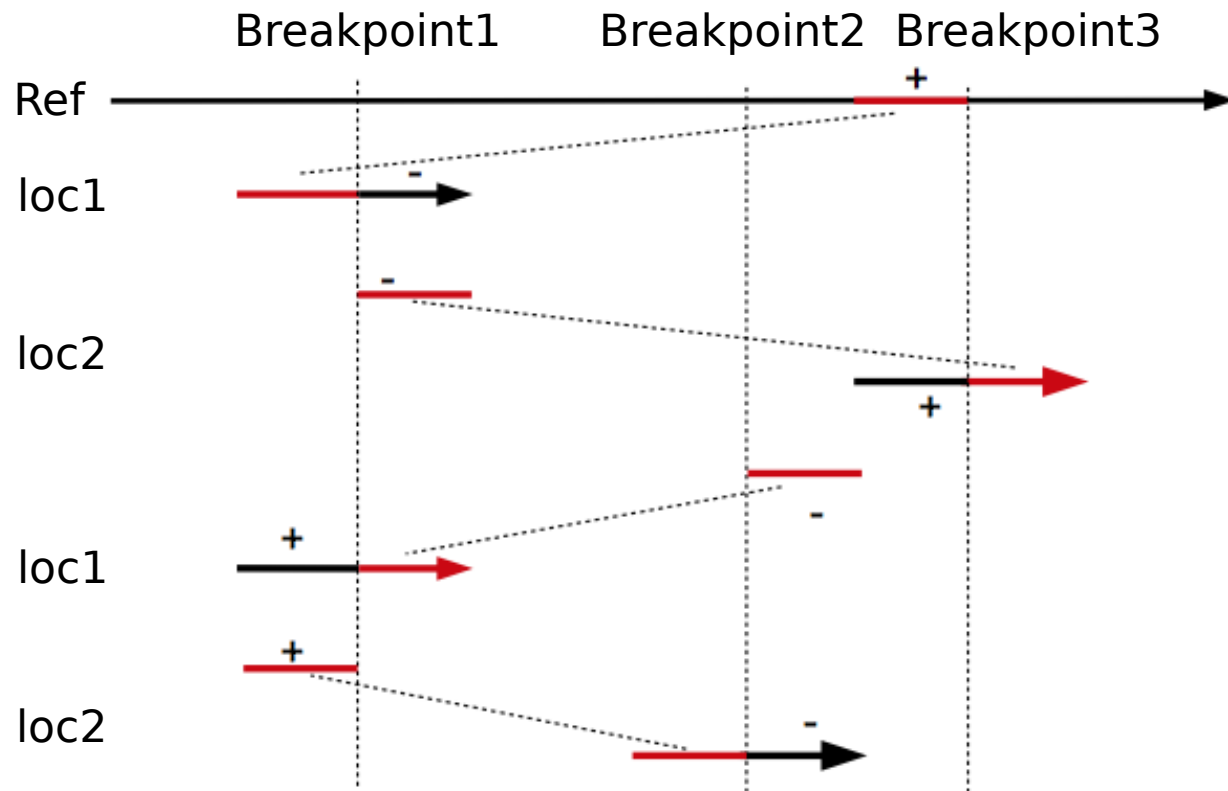
Deletion



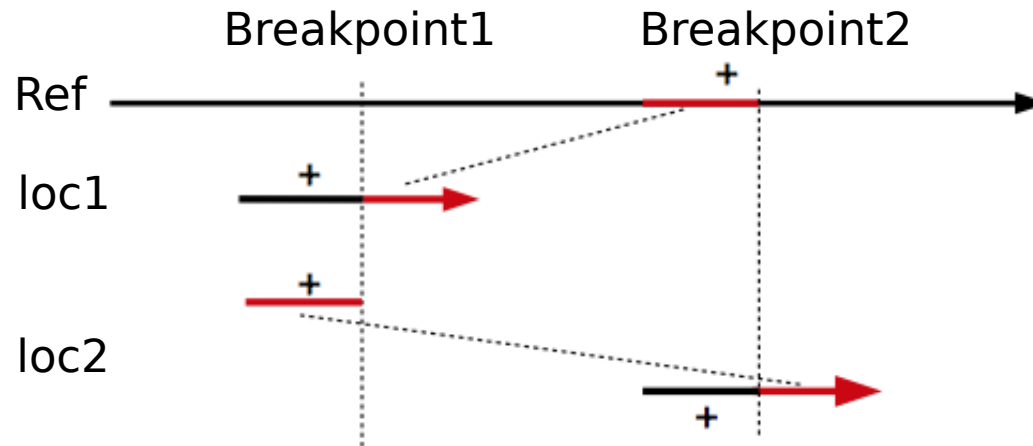
Duplication



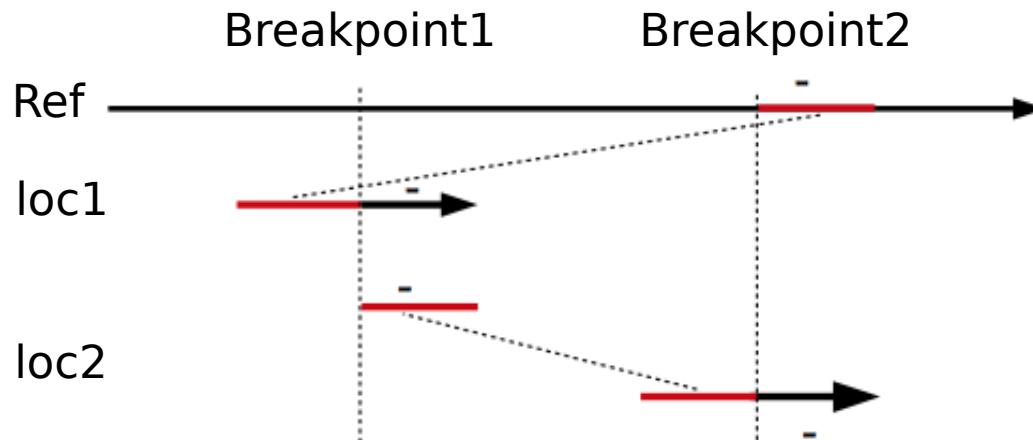
Interspersed duplication



Inversion



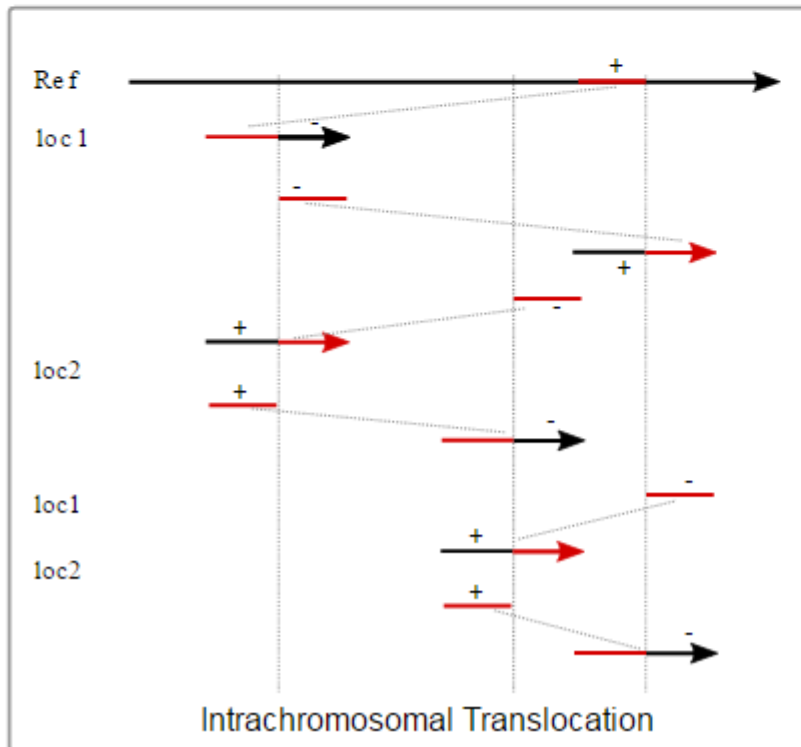
(a)



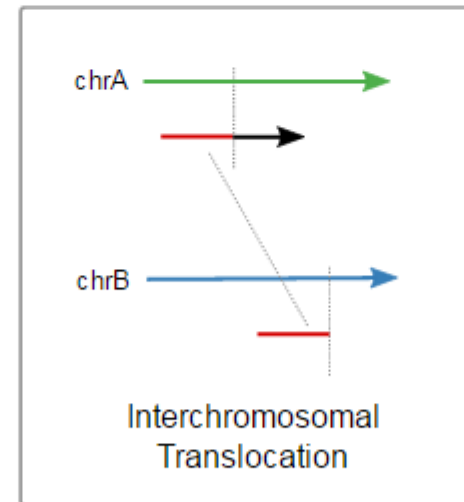
(b)

Translocation

E



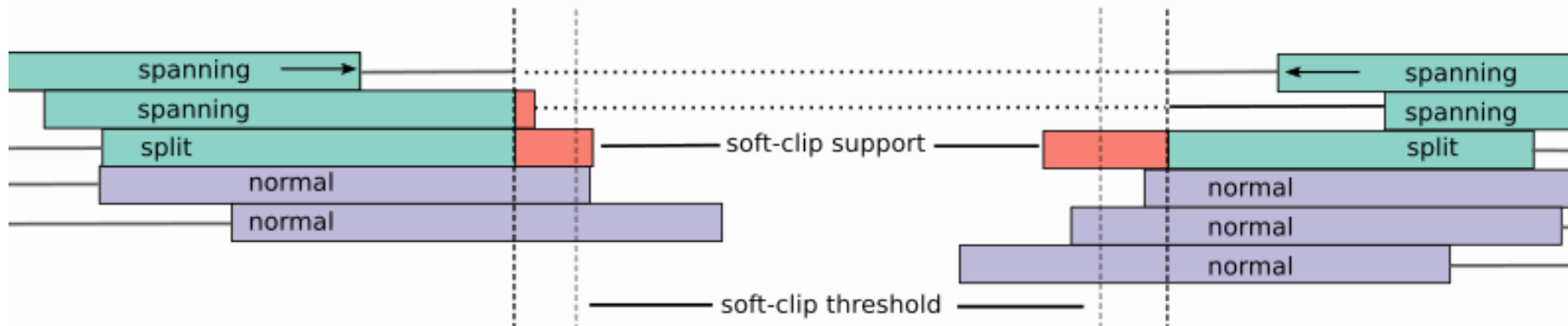
F



Counting supporting reads

Breakpoint 1	
split reads	1
spanning reads	2
normal reads	2
Raw VAF	0.6

Breakpoint 2	
split reads	1
spanning reads	2
normal reads	3
Raw VAF	0.5



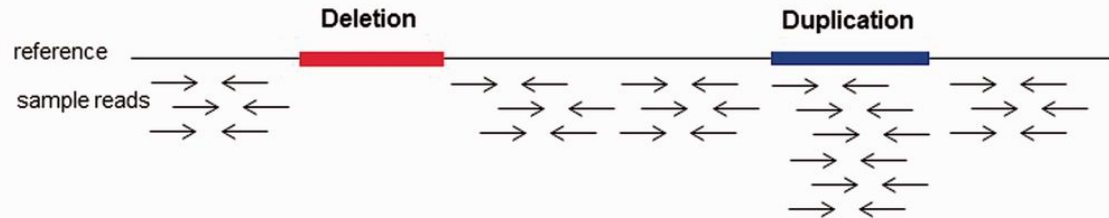
Read counting criteria

Read	Crosses break	SC > threshold	Facing break	Valid insert
Spanning	Y	N	Y	Y
Split	Y	Y	-	Y
Normal	Y	N	-	Y

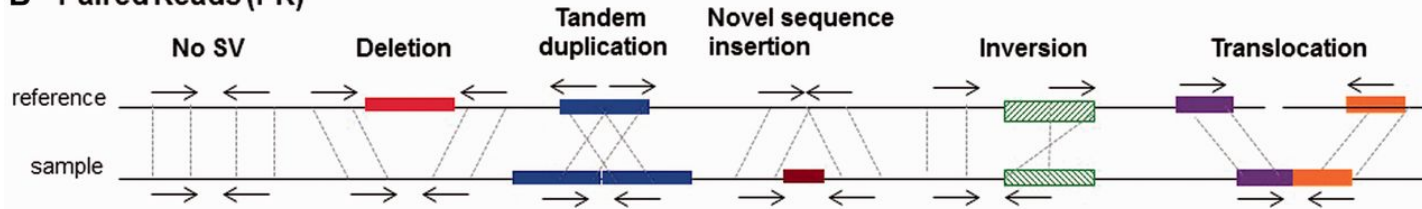
tumour reads
 normal reads

Strategies for structural variant (SV) detection.

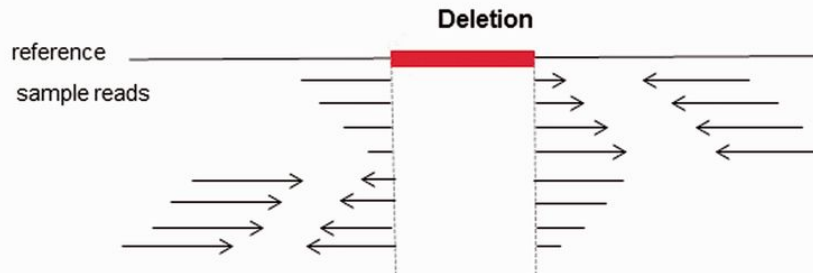
A Read Depth (RD)



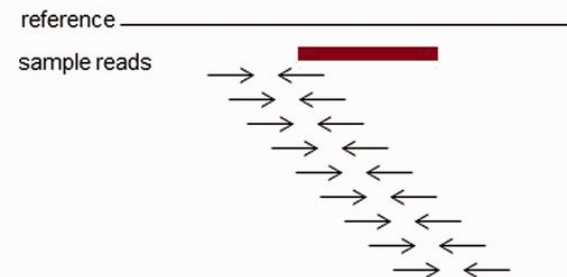
B Paired Reads (PR)



C Split Reads (SR)



D. De Novo Assembly (AS)



Geòrgia Escaramís et al. Briefings in Functional Genomics
2015;14:305-314

Methods

For a comprehensive list see the table in

<http://www.sciencedirect.com/science/article/pii/S1046202316300184>

BRASS – Breakpoint by assembly

- <https://github.com/cancerit/BRASS>
- Method type: read-pair plus assembly

Workflow:

1. Search for all discordant read pairs
2. Calculate copy-number changes
3. Filter candidate reads
4. Assemble reads around each breakpoint
5. Determine the class of each SV (includes merging)

EXERCISE 2