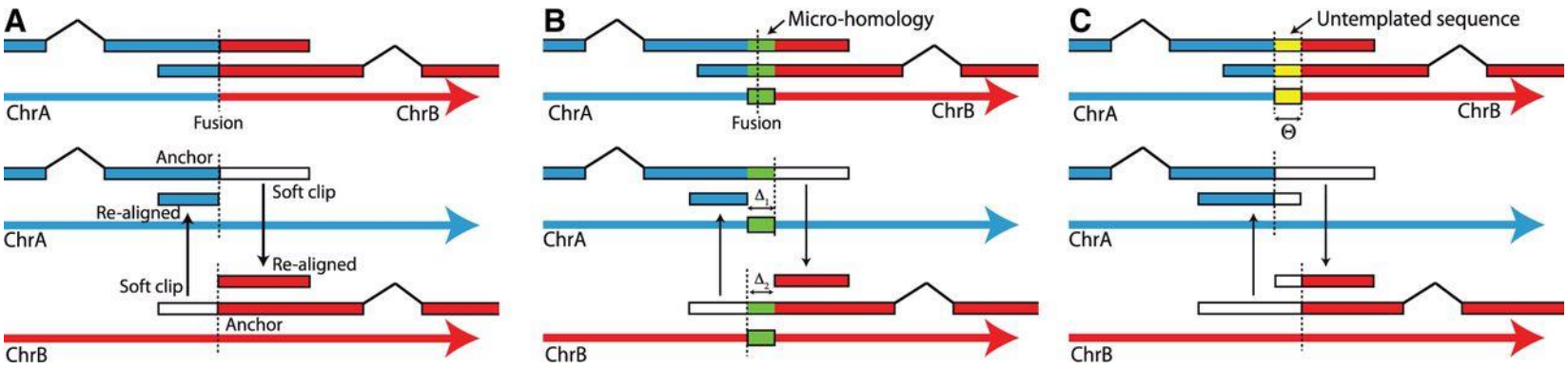
METHODS FOR CALLING SVS

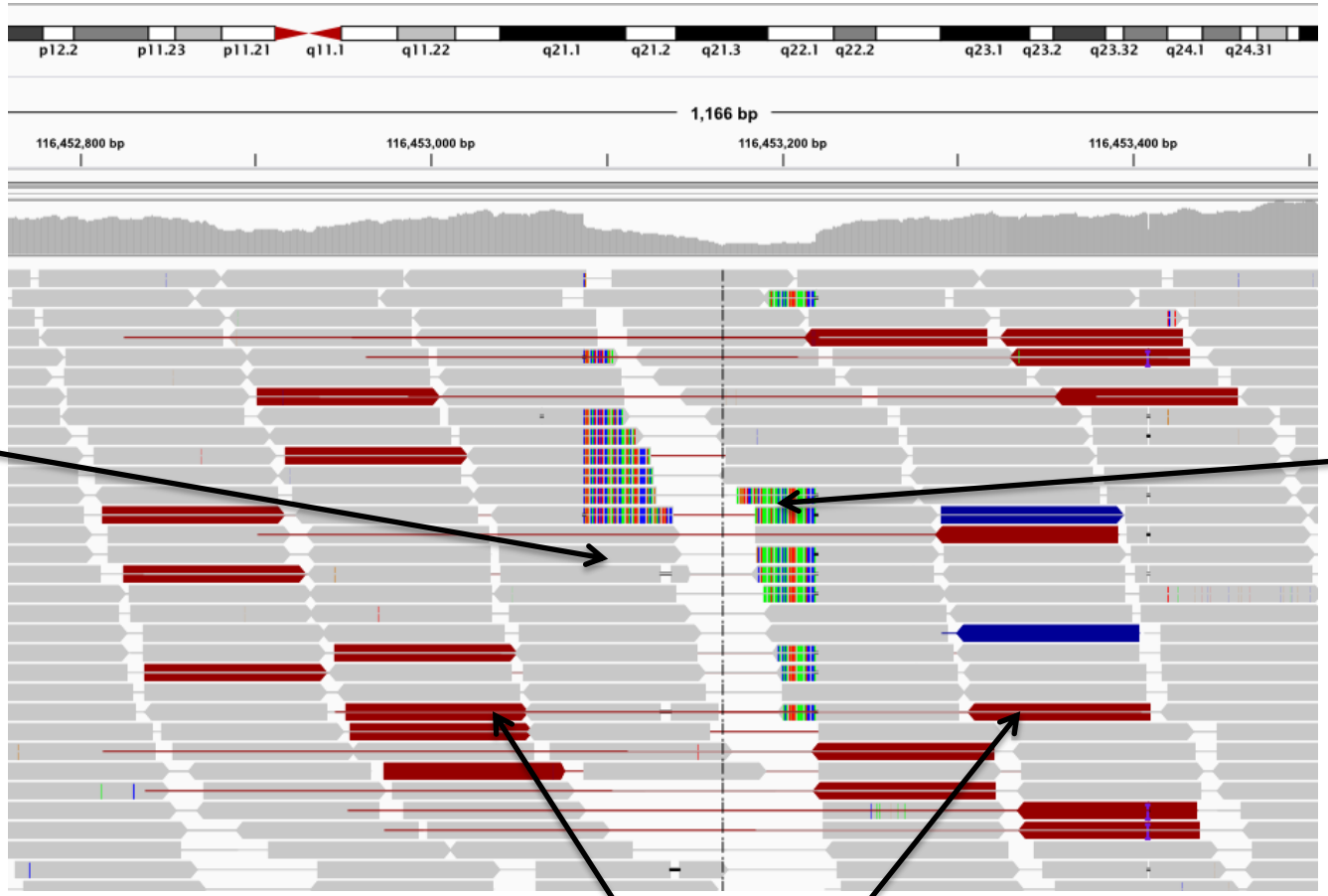
Reads overlapping the breakpoint



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

Bioinformatics

Reads around the breakpoint

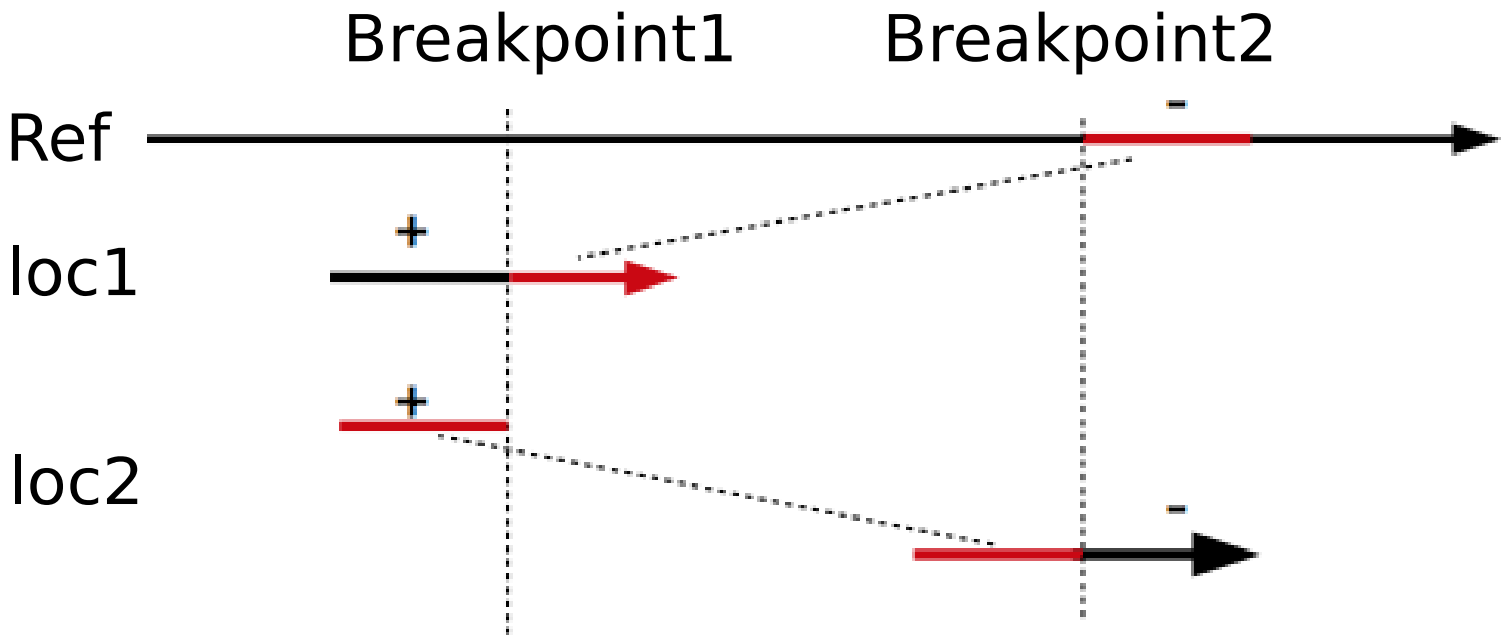


Normal reads

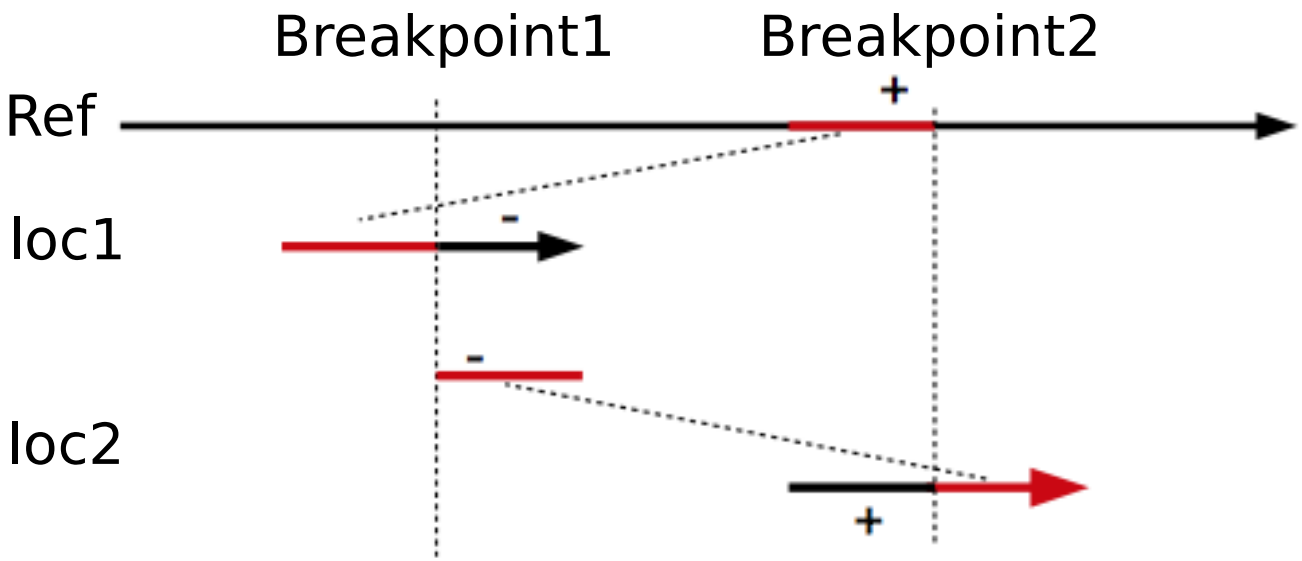
Split reads

Discordant spanning reads

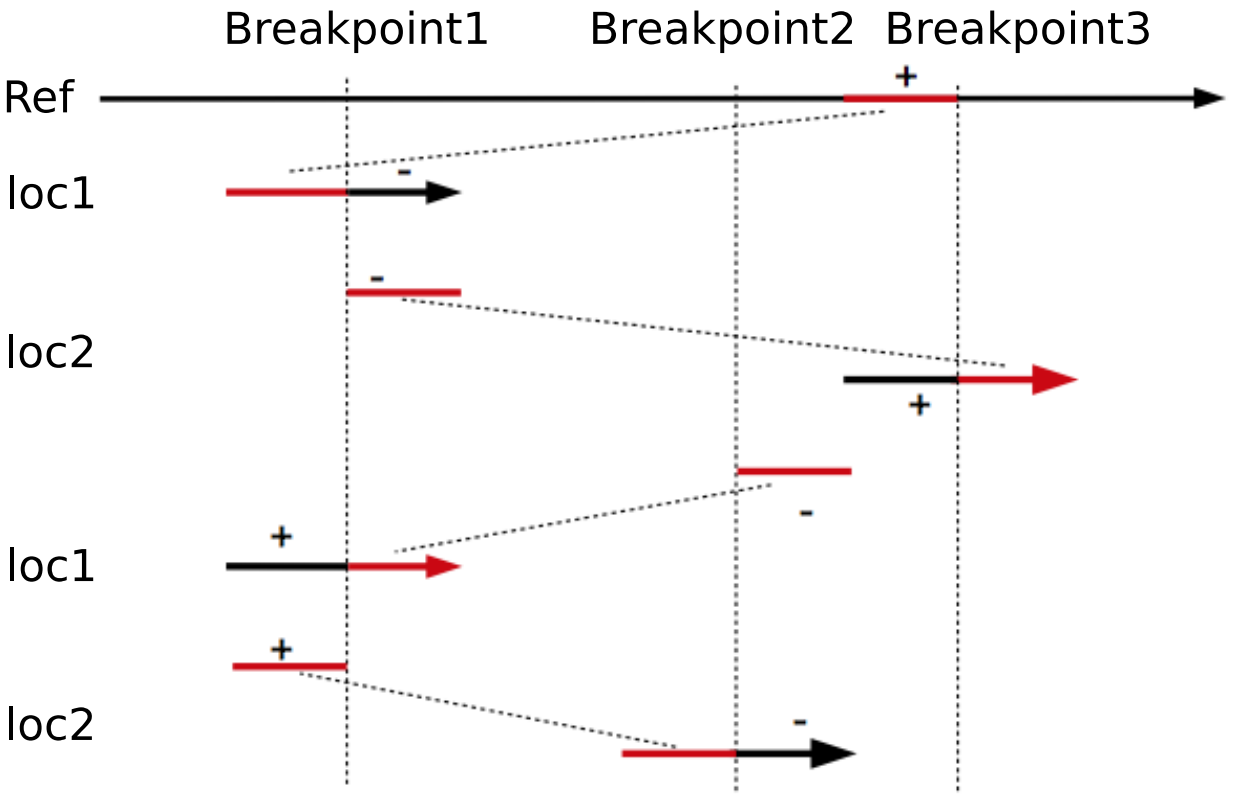
Deletion



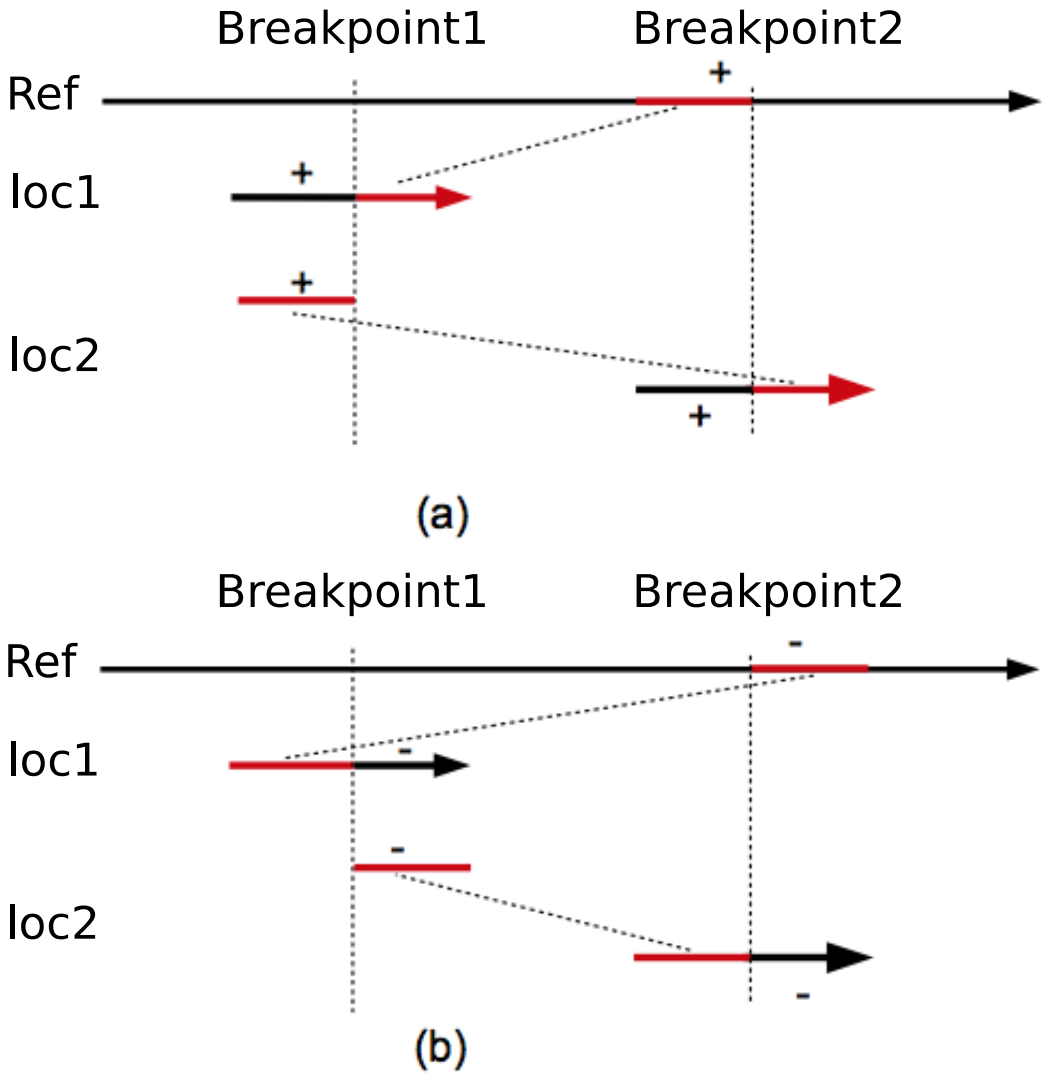
Duplication



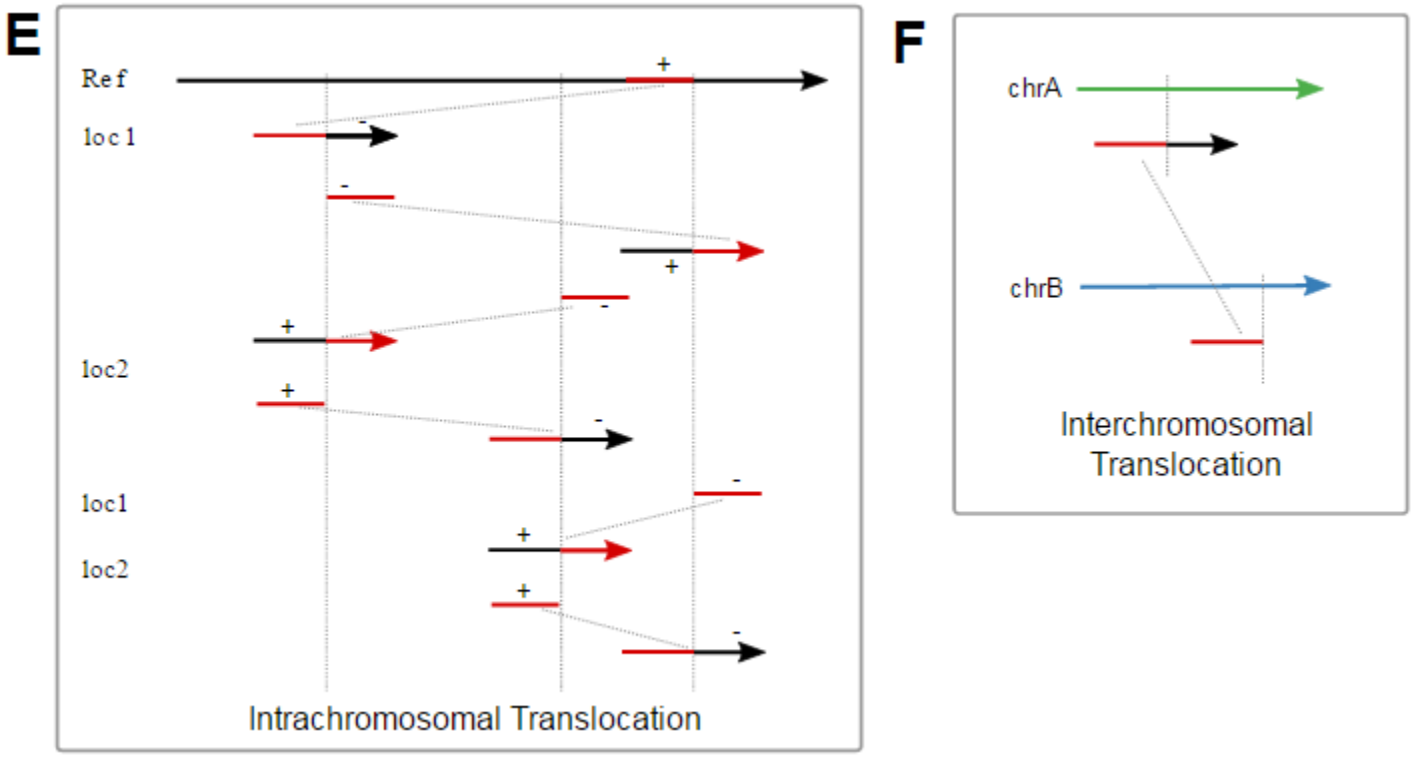
Interspersed duplication



Inversion


Breakpoint1
Breakpoint2
Ref
loc1
loc2
+
+
+
+
(a)
Breakpoint1
Breakpoint2
Ref
loc1
loc2
-
-
-
-
(b)

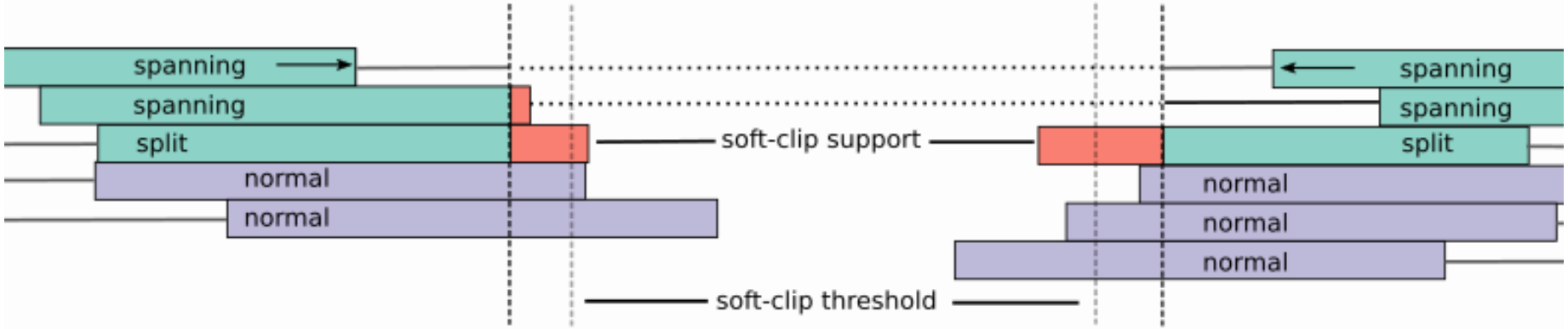
Translocation



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

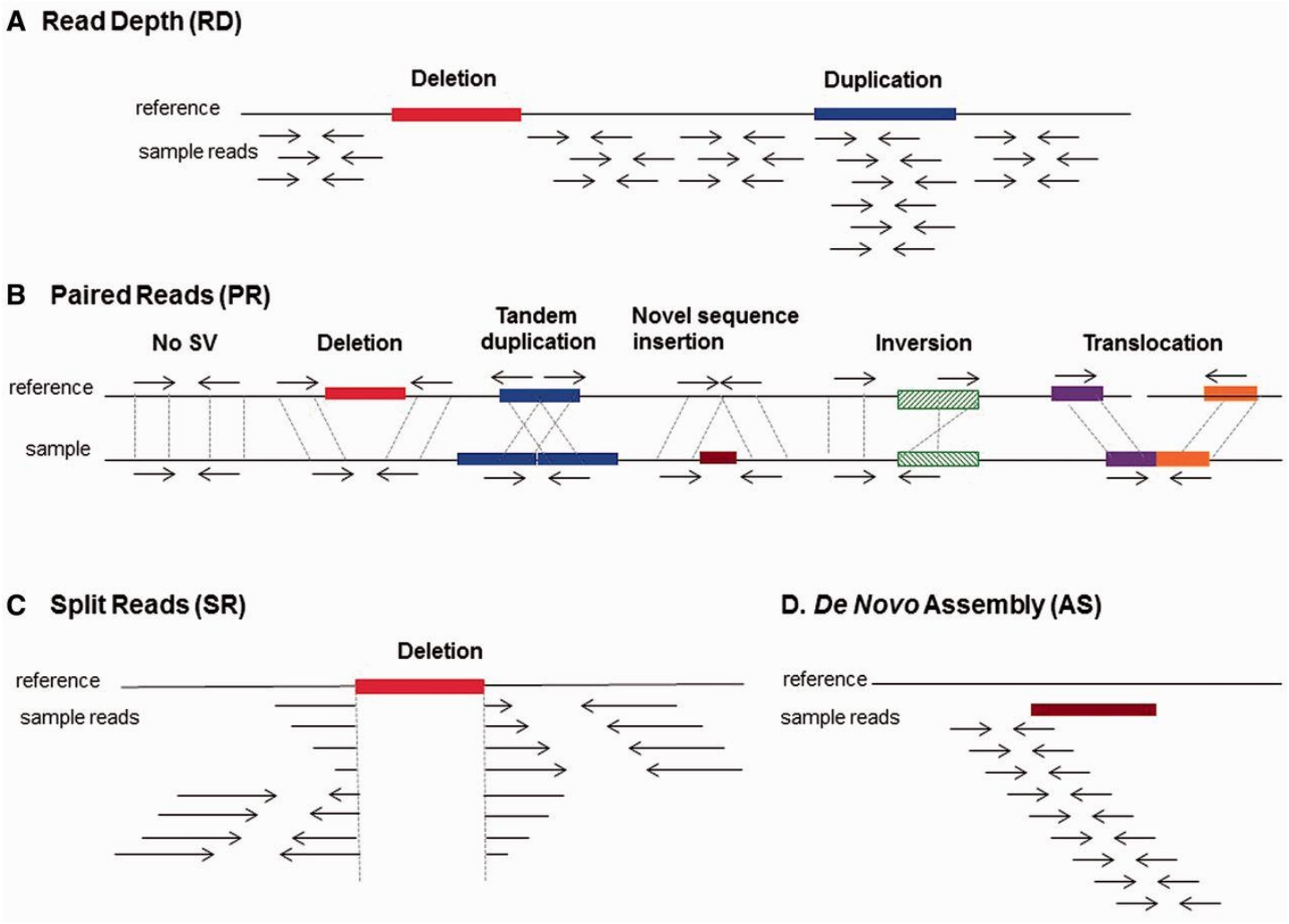


tumour reads
normal reads

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

Strategies for structural variant (SV) detection.



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

Briefings in Functional Genomics

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