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Modern Genomic Technologies (LF:DSMGT01)

Lecture 3b : Structural variants

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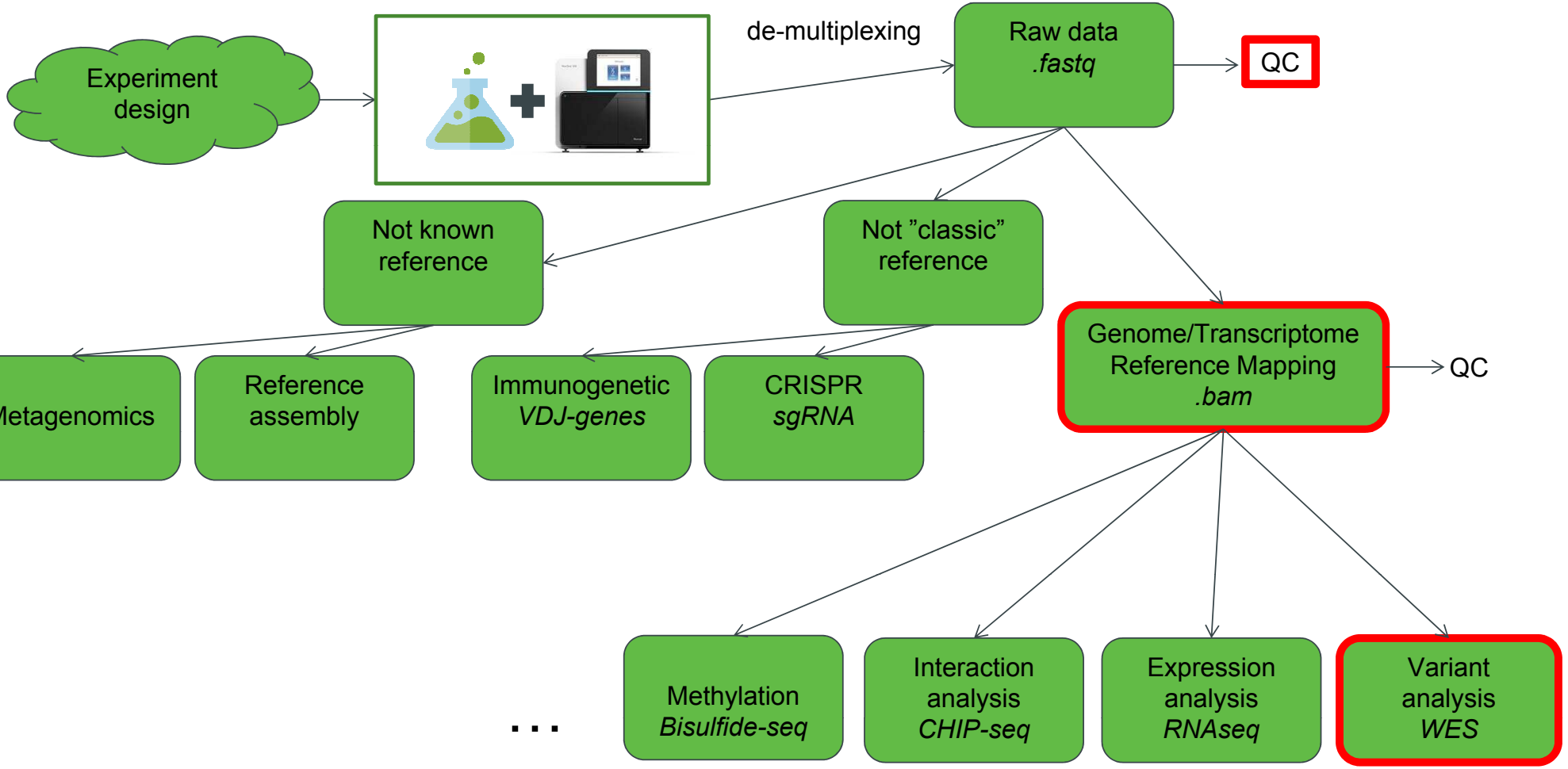
NGS data analysis



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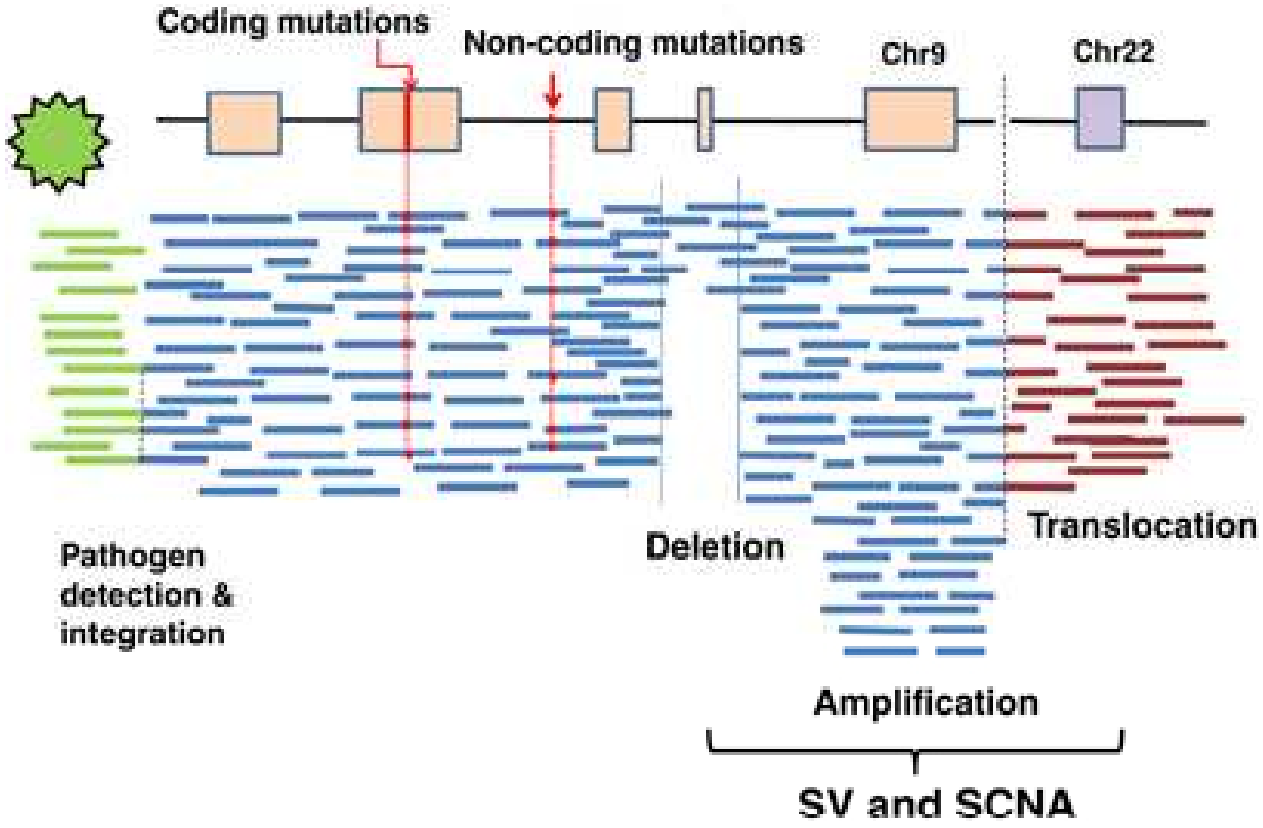
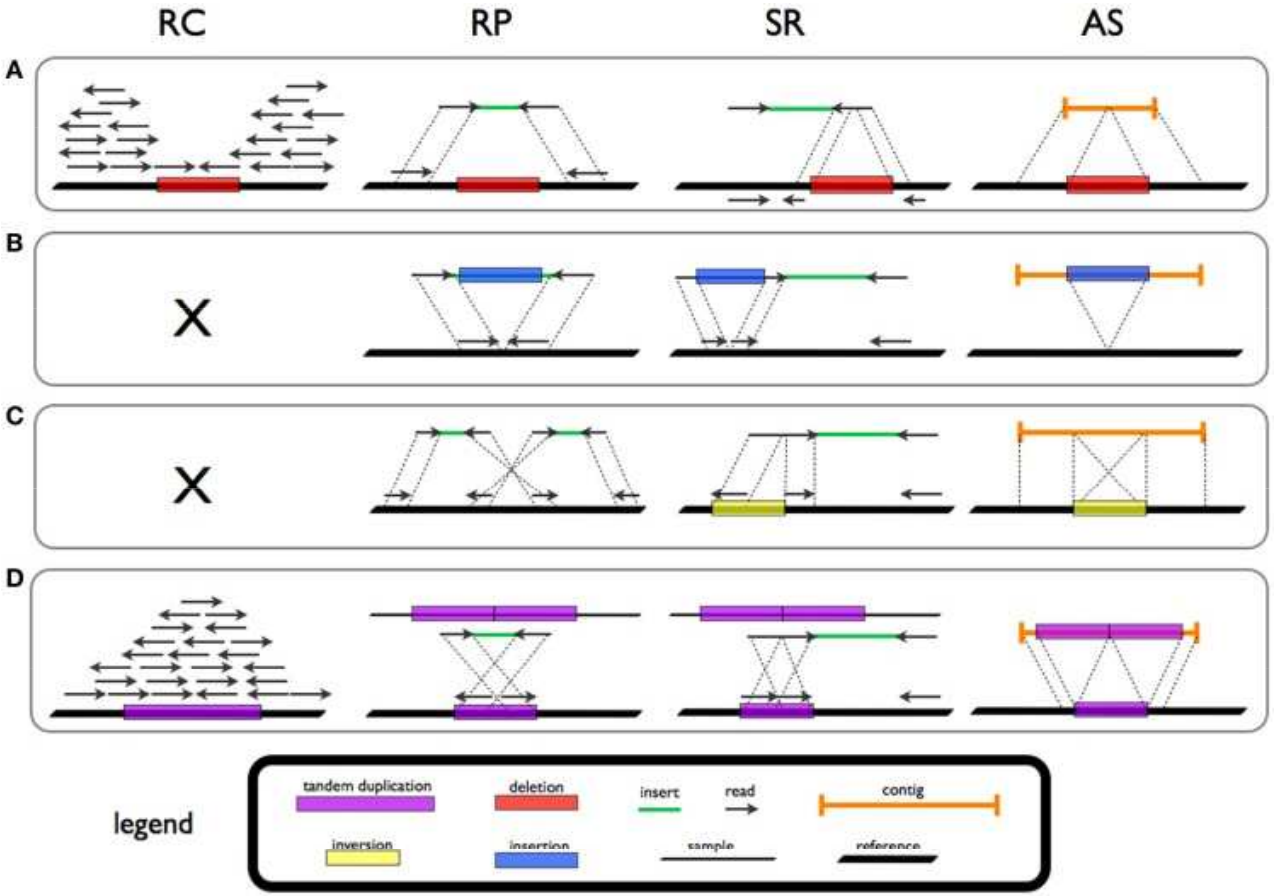
Structural variants calling



Structural variants calling

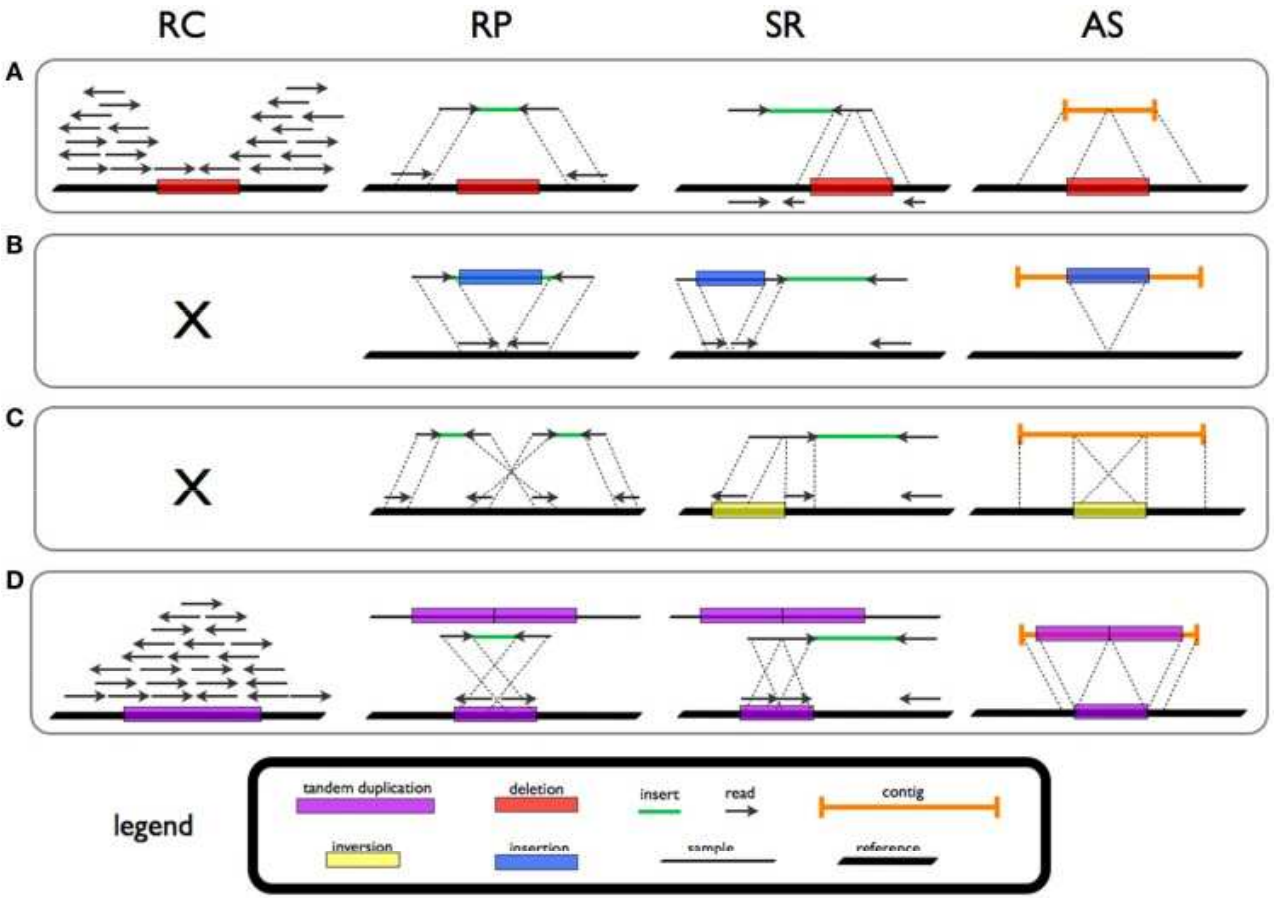
- RC = Read count
  - Copy number variants (CNV)
- RP = Read pair
  - Pair-end sequencing
- SR = Split reads
- AS = Assembled read



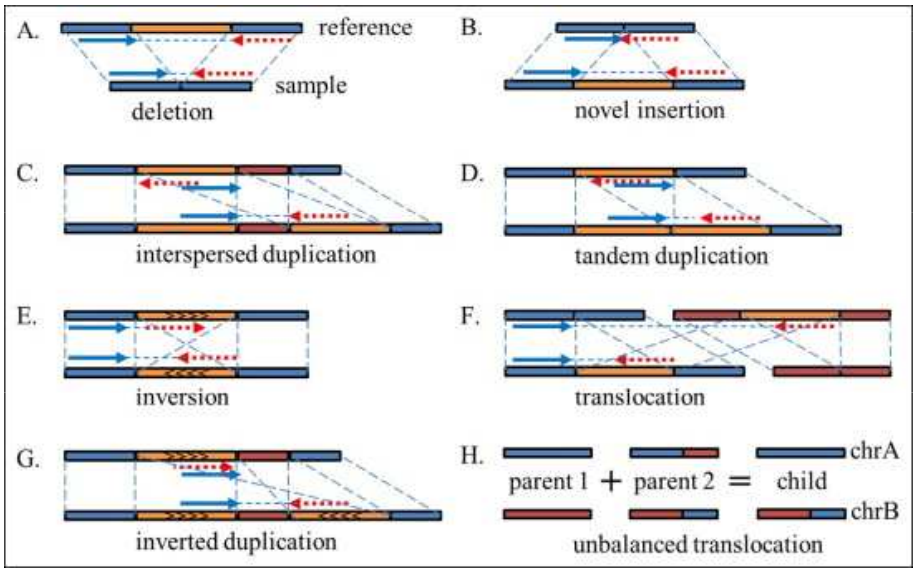
Structural variants calling

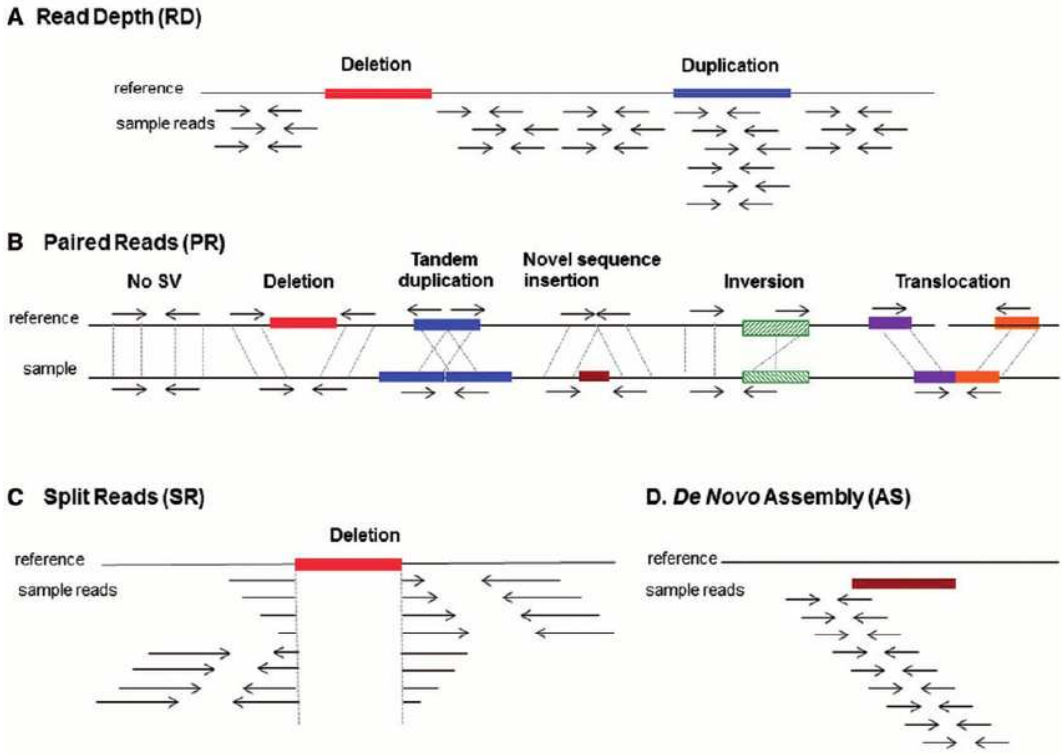
- RC = Read count
  - Copy number variants (CNV)
- RP = Read pair
  - Pair-end sequencing
- SR = Split reads
- AS = Assembled read

- A = DELETION
- B = INSERTION
- C = INVERSION
- D = DUPLICATION



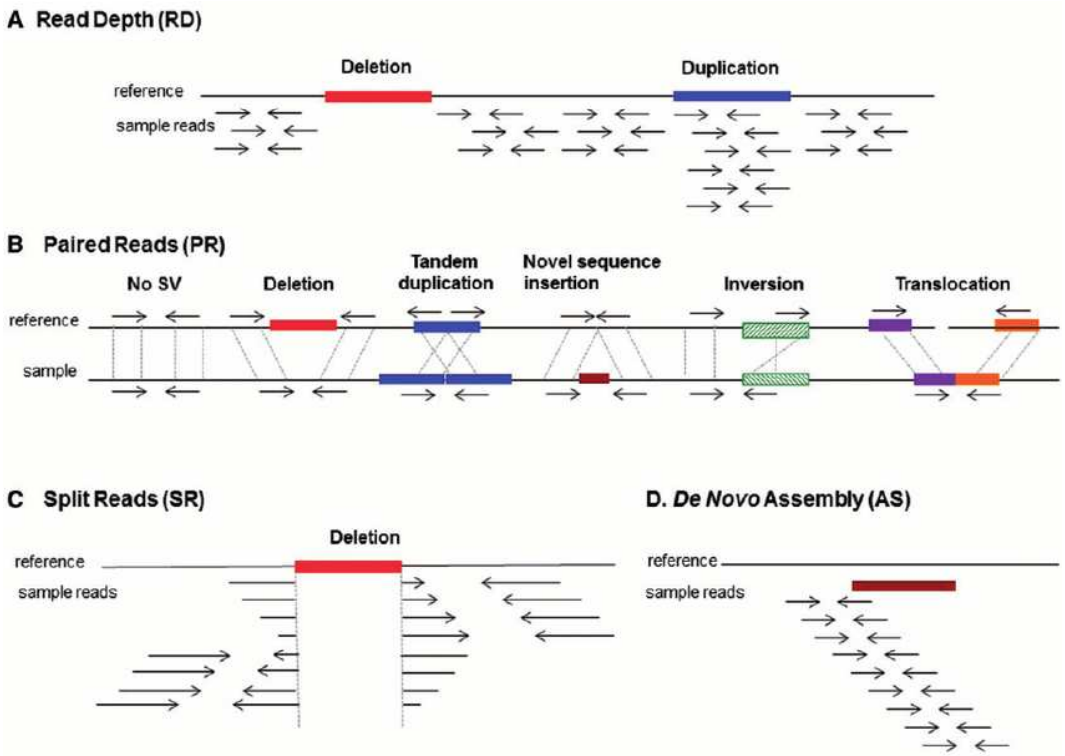
Structural variants calling





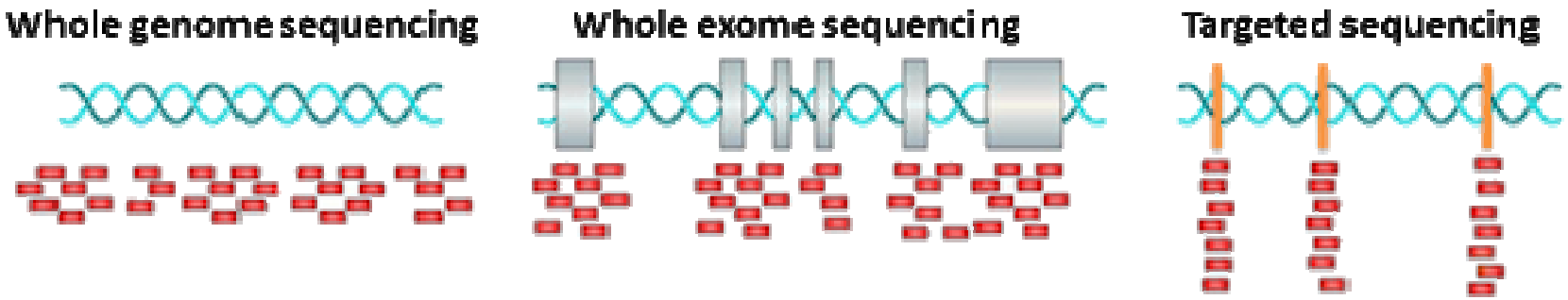
Structural variants calling

- Copy number variants (CNV)
  - Copy number analysis (CNA)
- Structural variants (SV)
  - Discordant reads analysis
- Fusion genes analysis
  - SV in coding sequence
  - From RNA-seq
  - Medical application



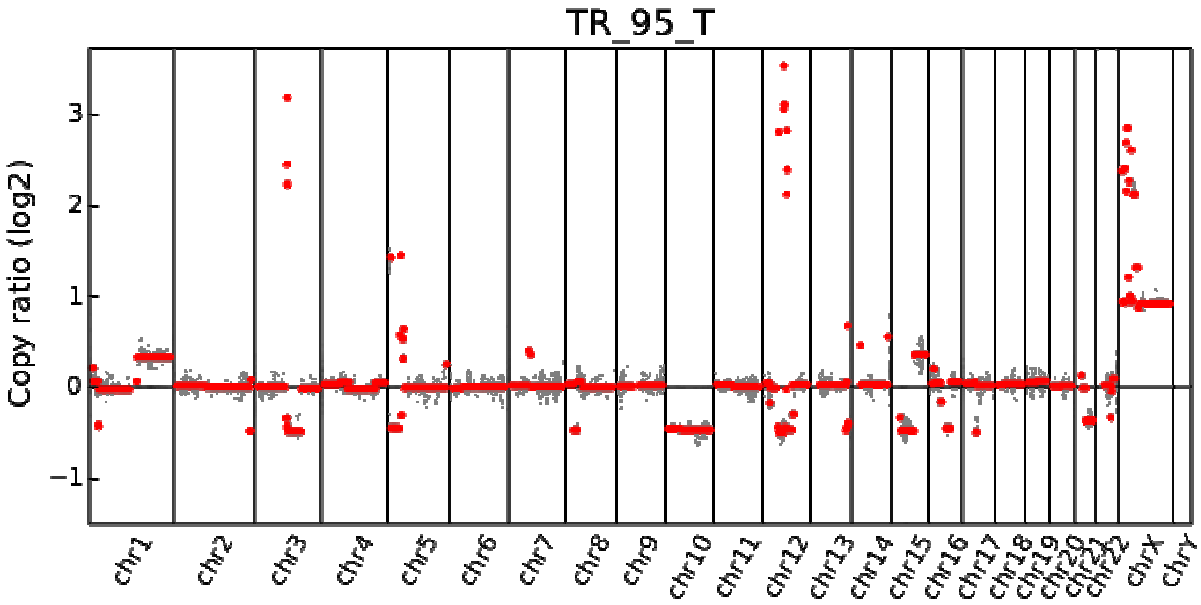
Copy number variants

- Not-PCR amplified (WGS)
- PCR amplified (WES,targeted)

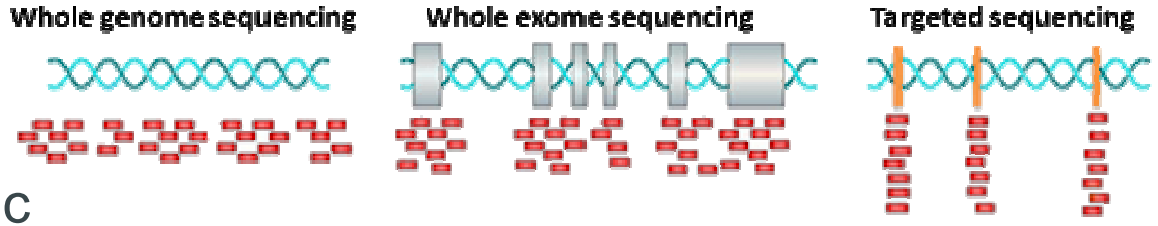


WGS copy number variants

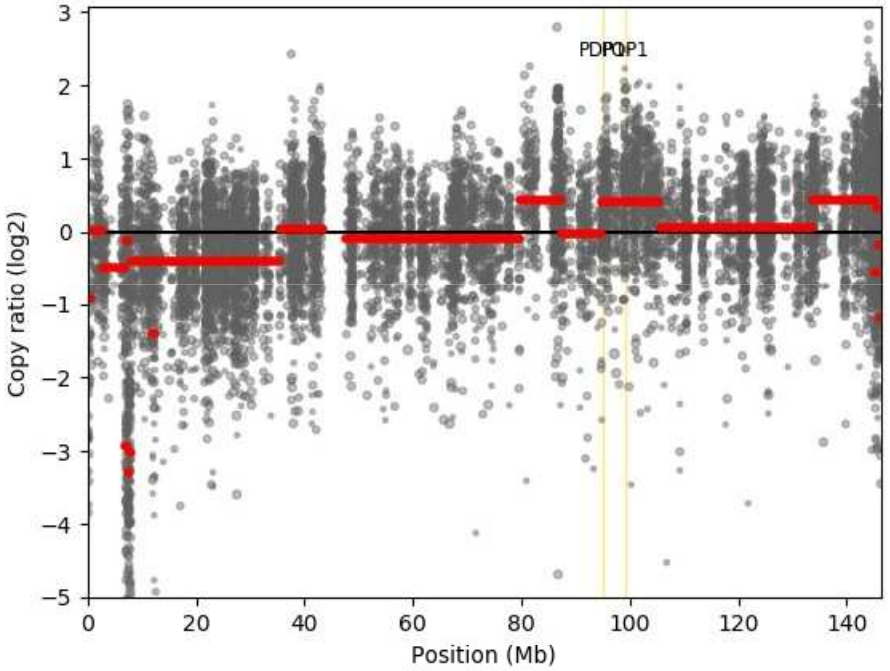
- Running window
- Normalization to the absolute coverage



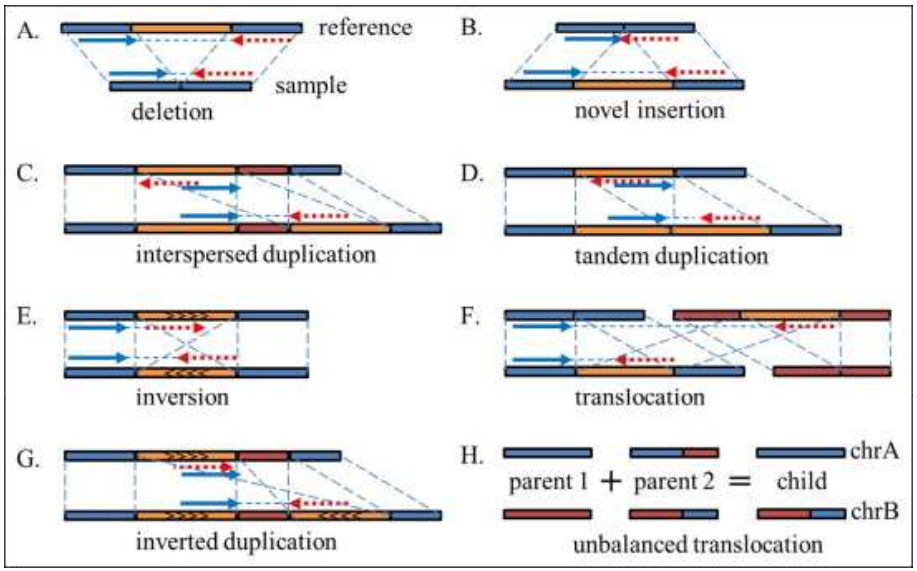
PCR amplified CNV

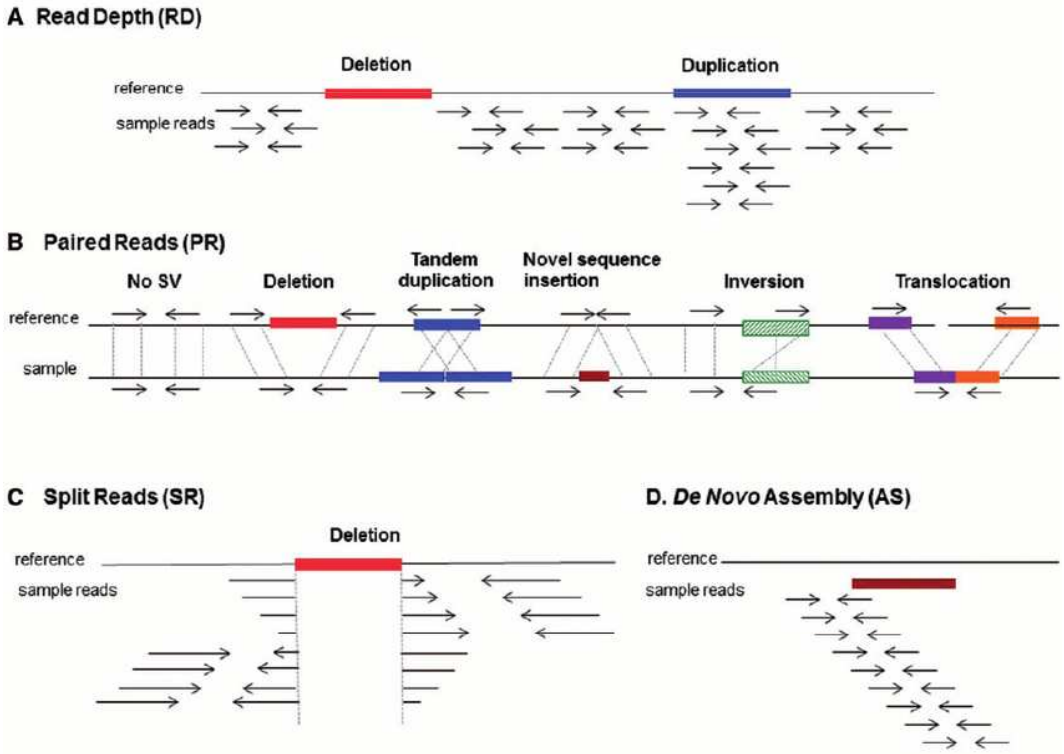


- Individual analysis problematic
- Somatic tumor-normal pairs
- Panel of (normal) samples

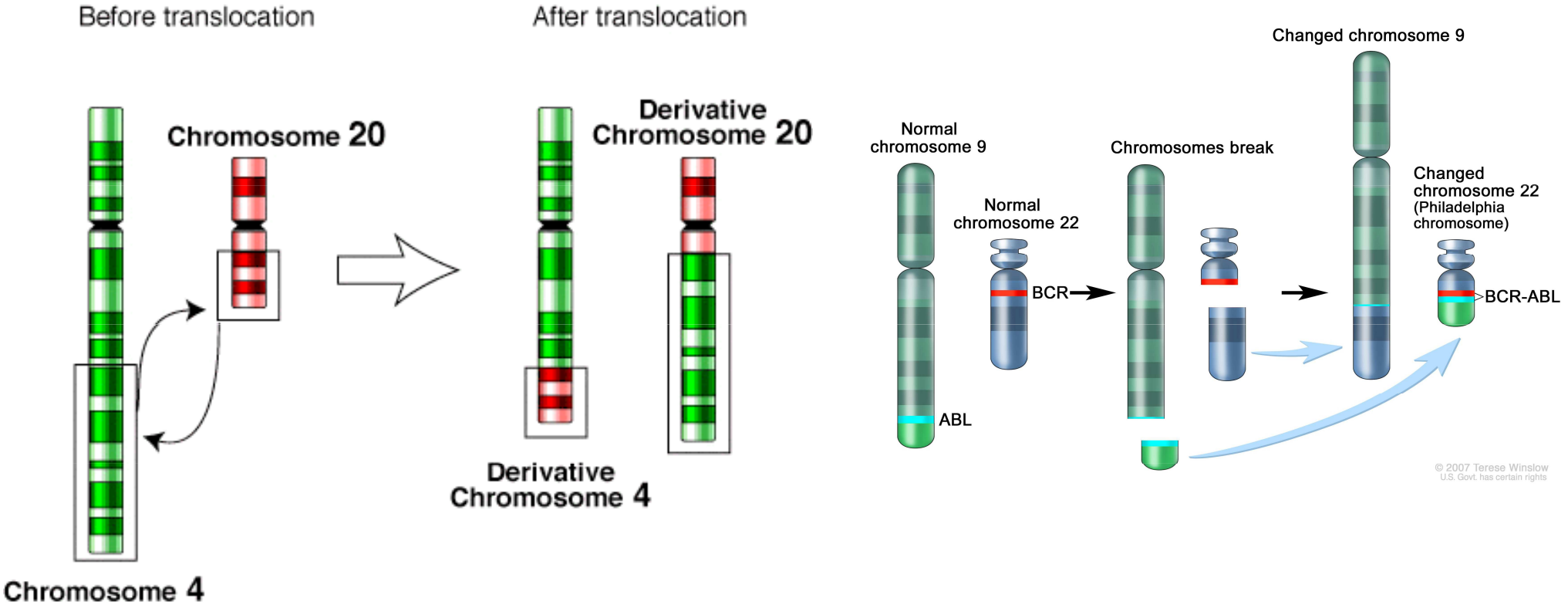


Structural variants calling





Fusion genes



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Thank you for your attention!

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