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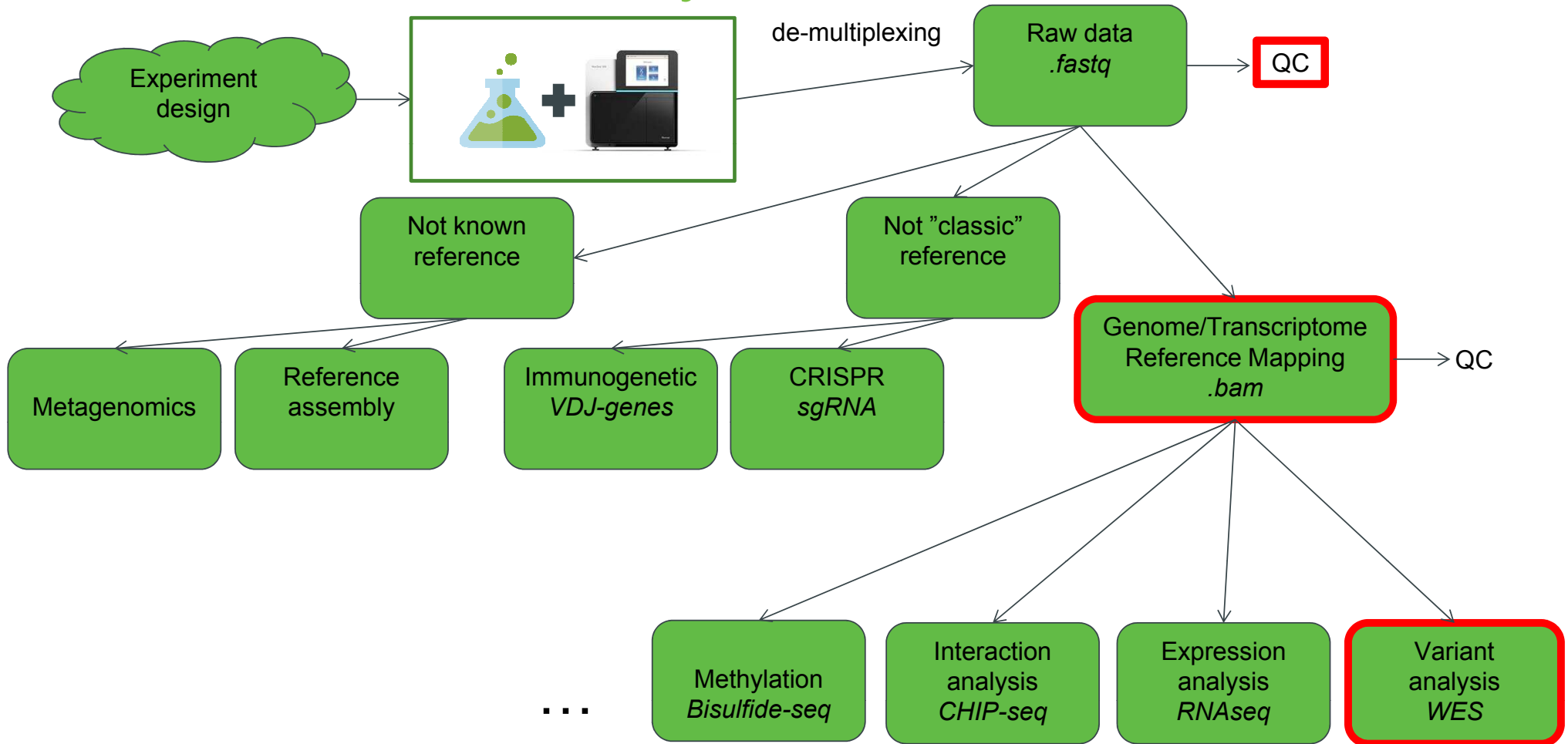


Modern Genomic Technologies (LF:DSMGT01)

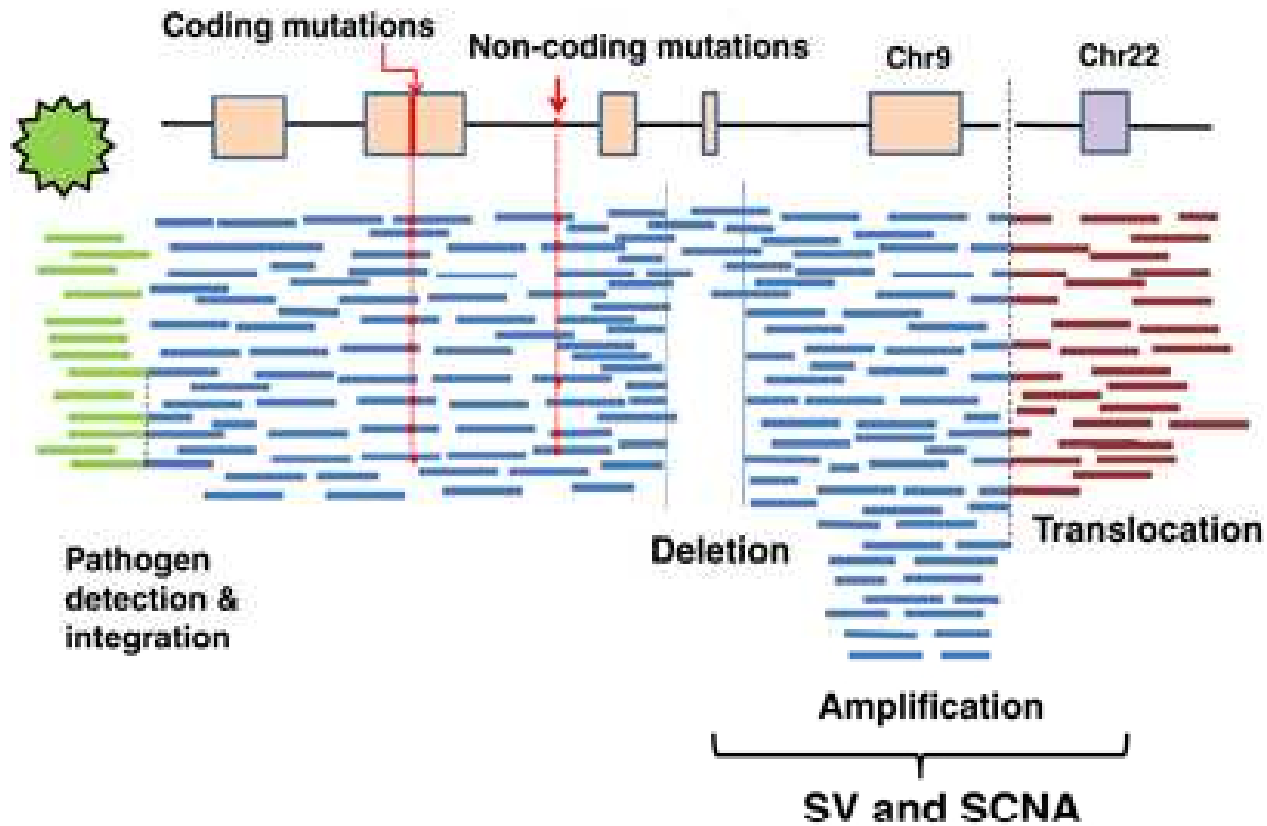
Lecture 3b : Structural variants

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vojtech.bystry@ceitec.muni.cz

NGS data analysis

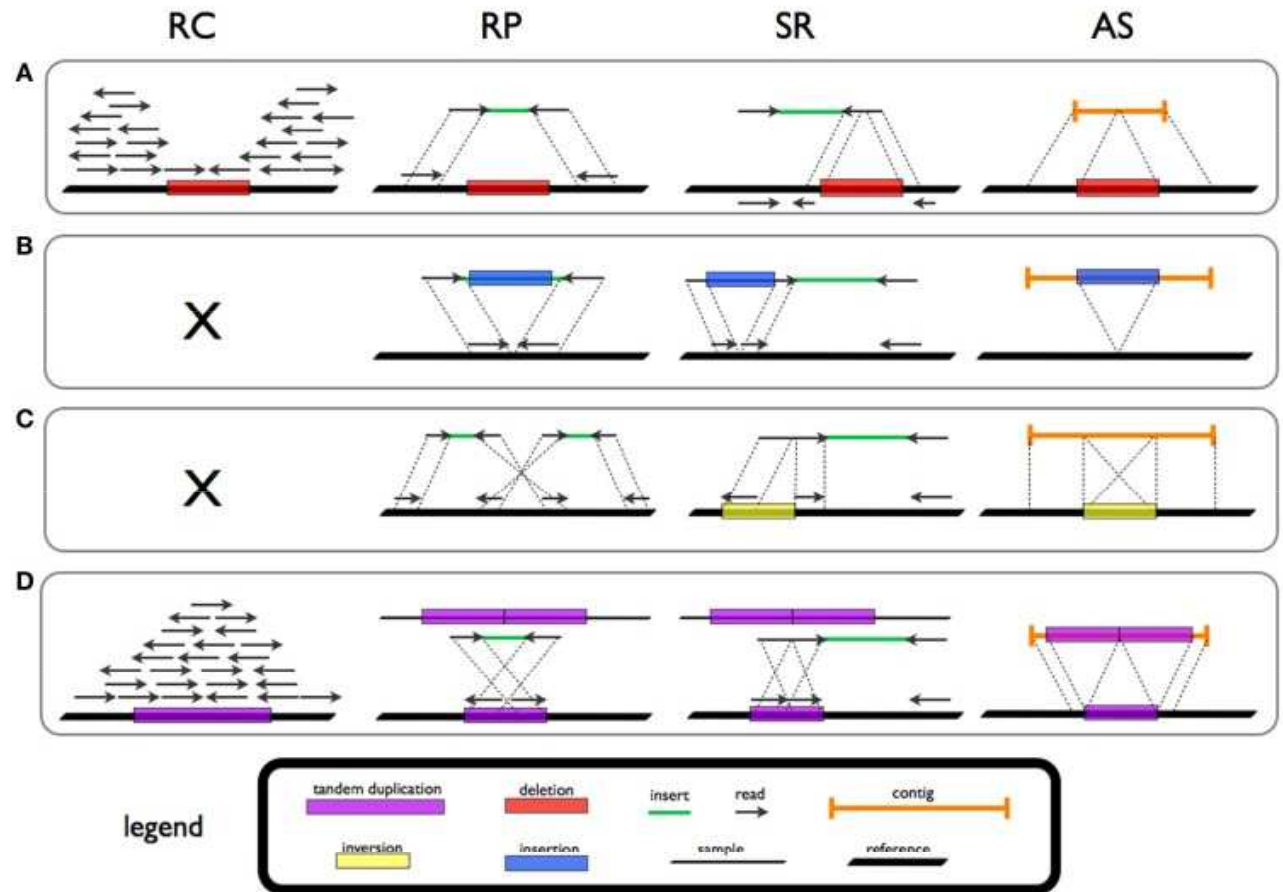


Structural variants calling



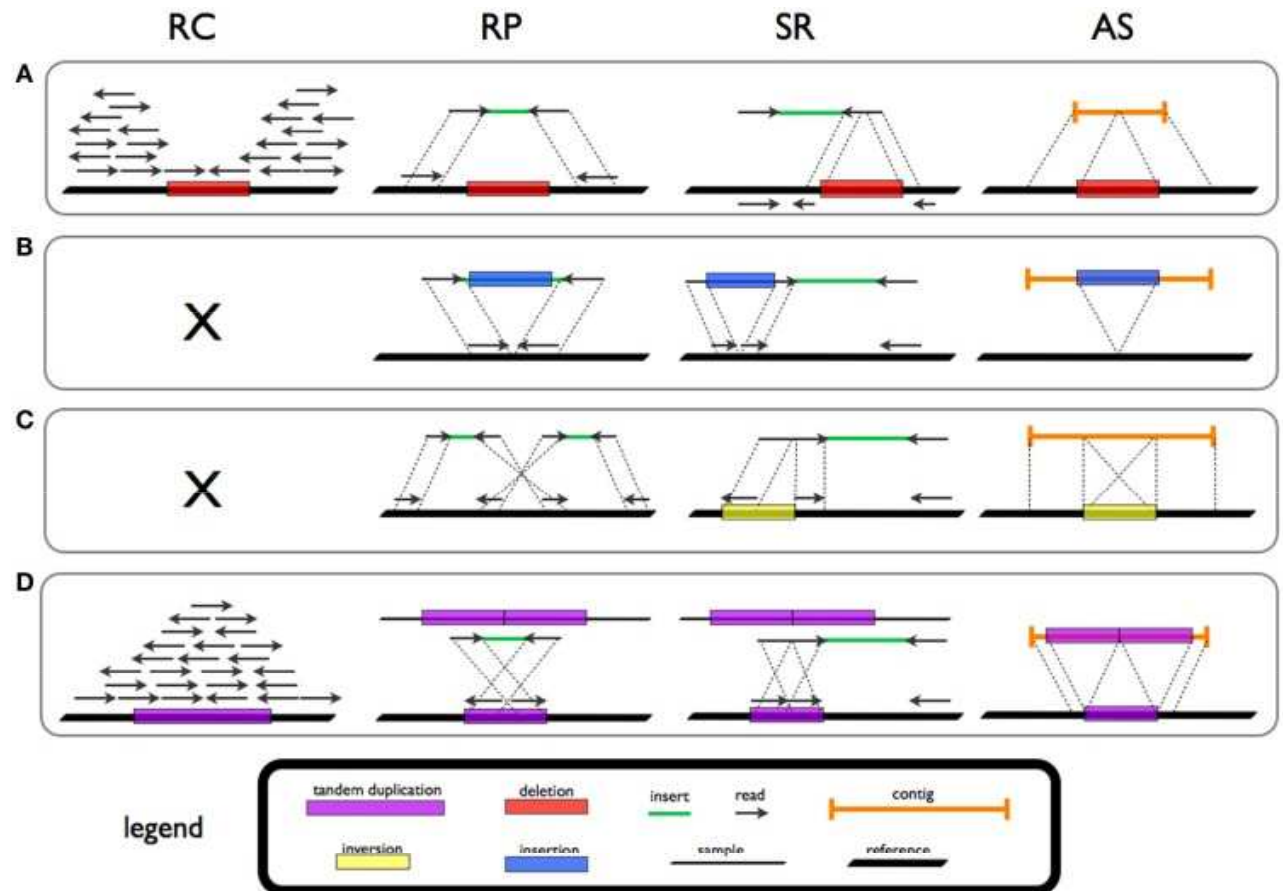
Structural variants calling

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

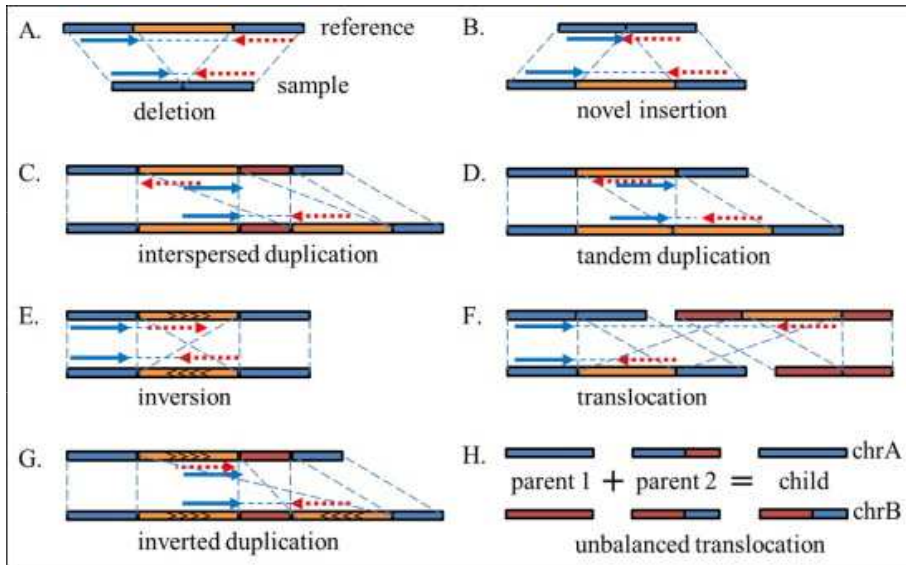


Structural variants calling

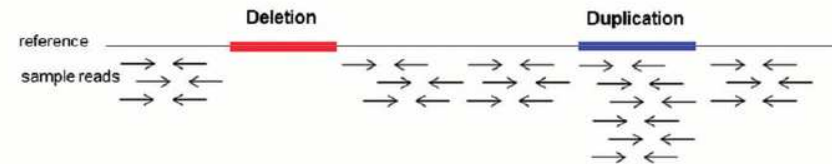
- RC = Read count
 - Copy number variants (CNV)
 - RP = Read pair
 - Pair-end sequencing
 - SR = Split reads
 - AS = Assembled read
-
- A = DELETION
 - B = INSERTION
 - C = INVERSION
 - D = DUPLICATION



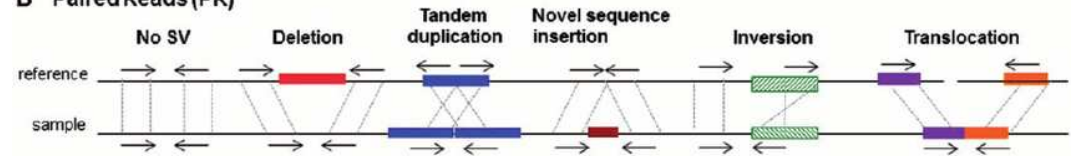
Structural variants calling



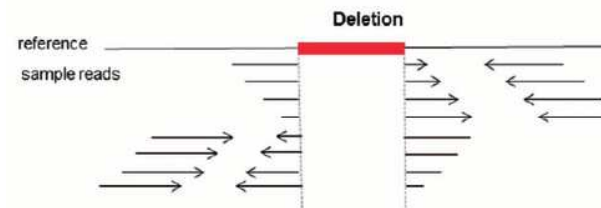
A Read Depth (RD)



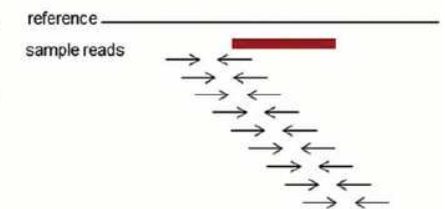
B Paired Reads (PR)



C Split Reads (SR)

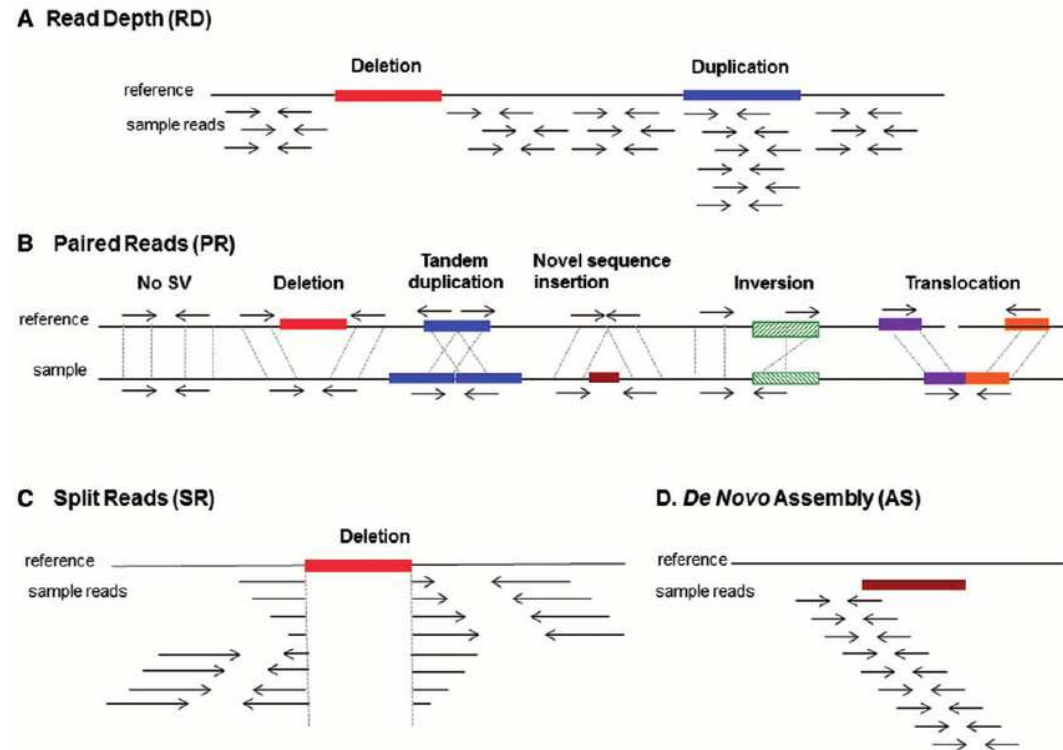


D De Novo Assembly (AS)



Structural variants calling

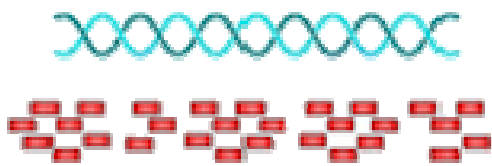
- Copy number variants (CNV)
 - Copy number analysis (CNA)
- Structural variants (SV)
 - Discordant reads analysis
- Fusion genes analysis
 - SV in coding sequence
 - From RNA-seq
 - Medical application



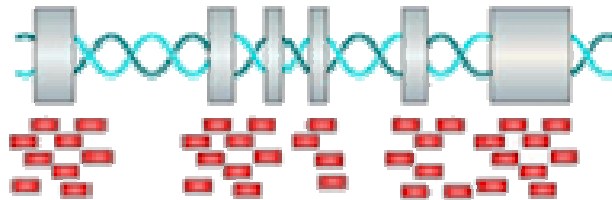
Copy number variants

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

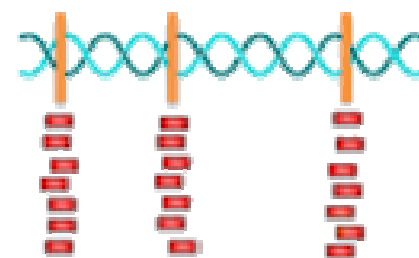
Whole genome sequencing



Whole exome sequencing

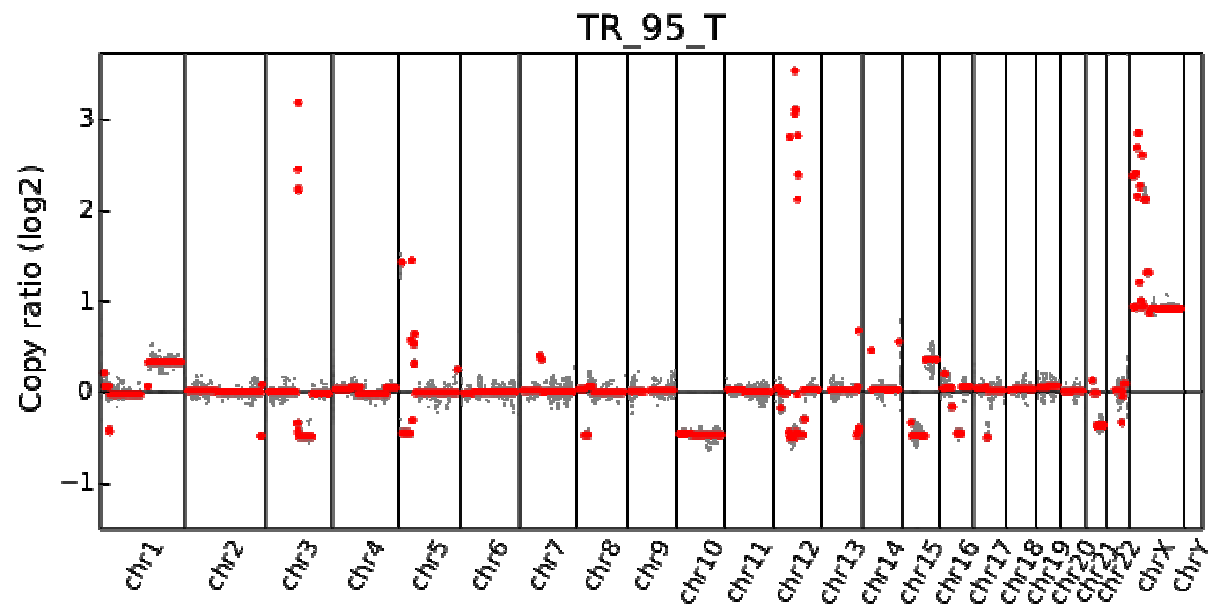


Targeted sequencing

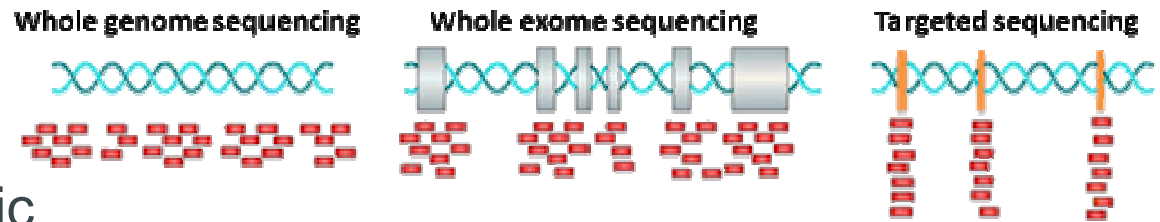


WGS copy number variants

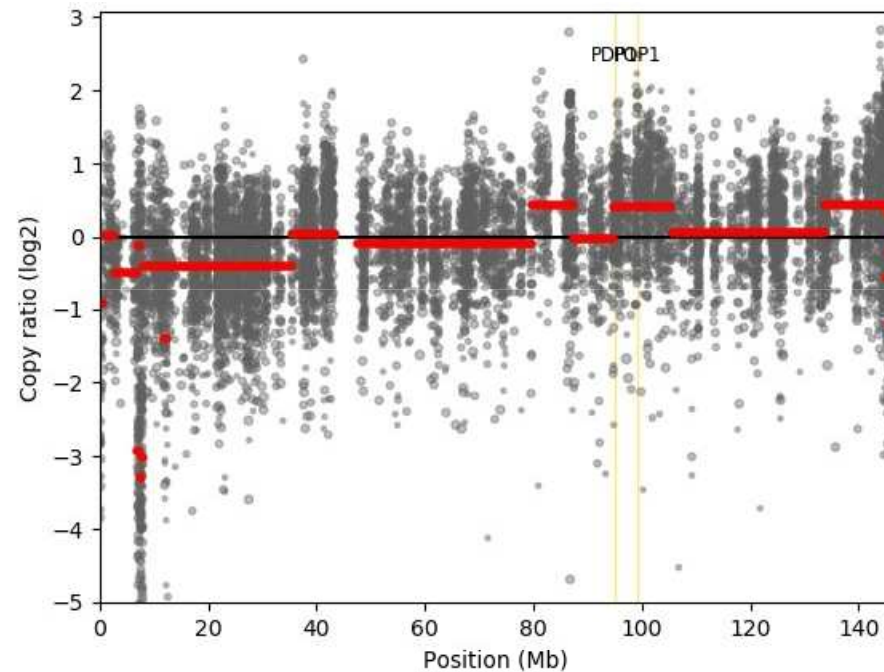
- Running window
- Normalization to the absolute coverage



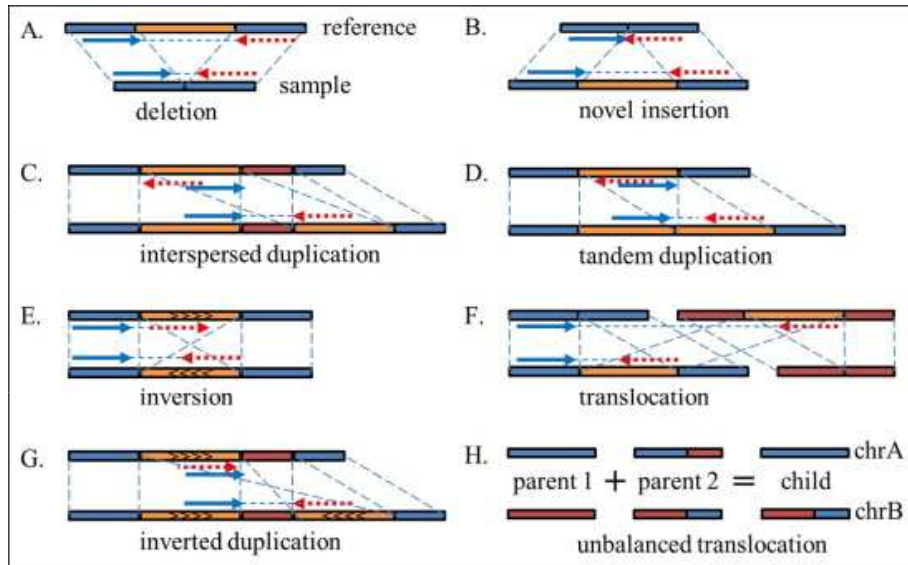
PCR amplified CNV



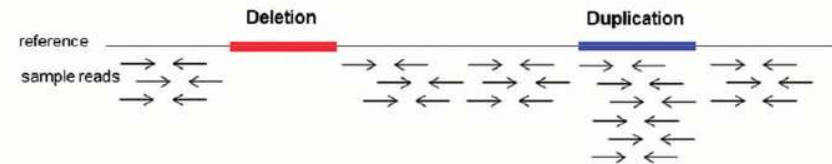
- Individual analysis problematic
- Somatic tumor-normal pairs
- Panel of (normal) samples



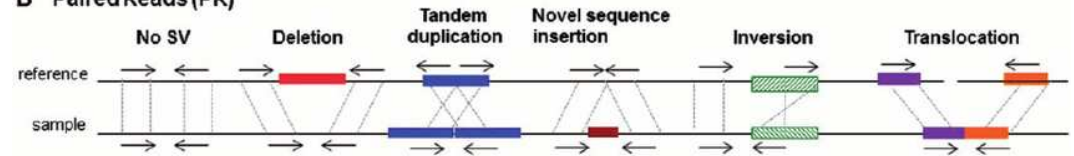
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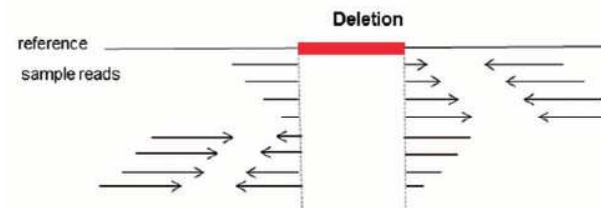
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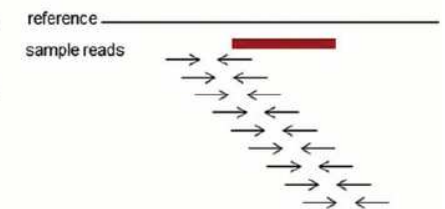
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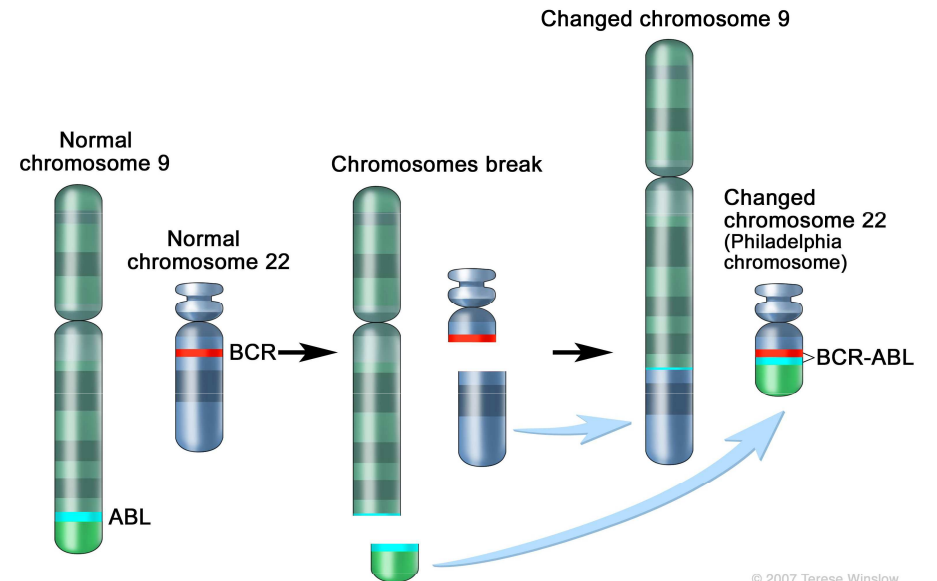
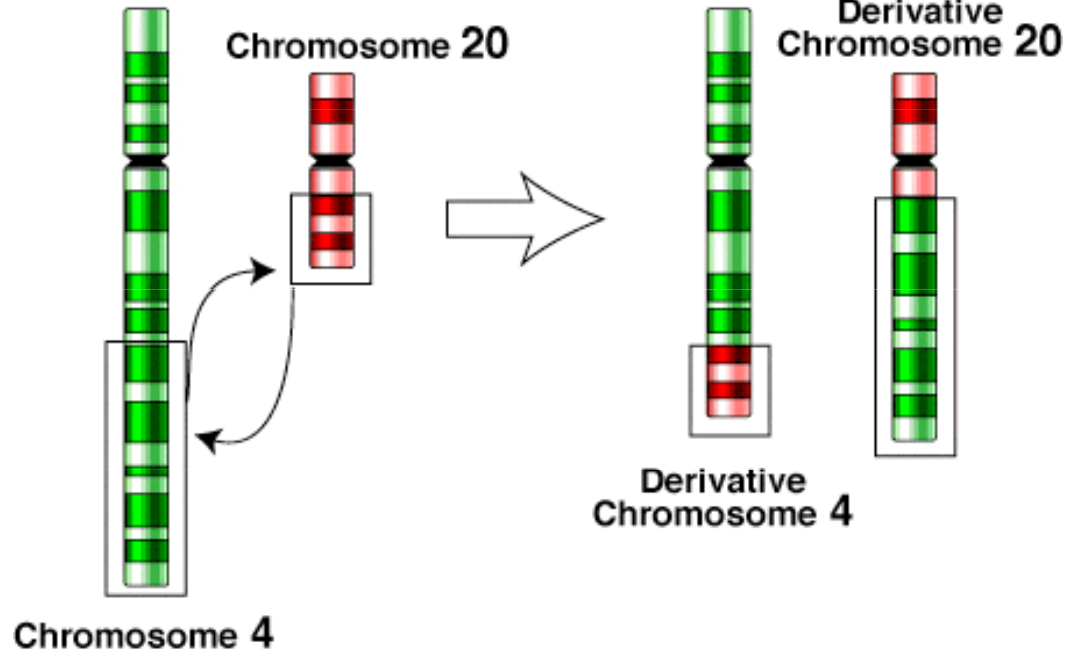
D De Novo Assembly (AS)



Fusion genes

Before translocation

After translocation



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Thank you for your attention!