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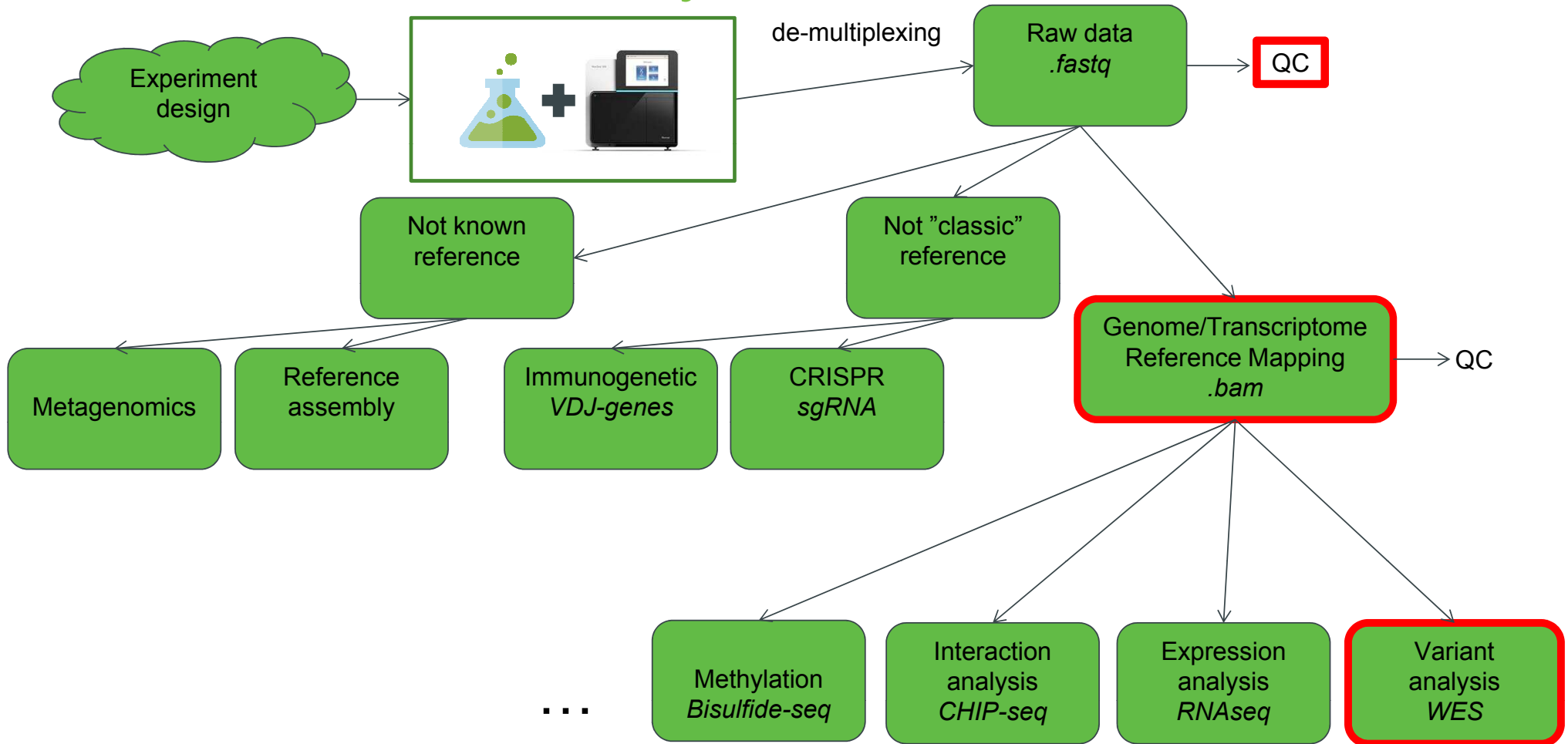


## Modern Genomic Technologies (LF:DSMGT01 )

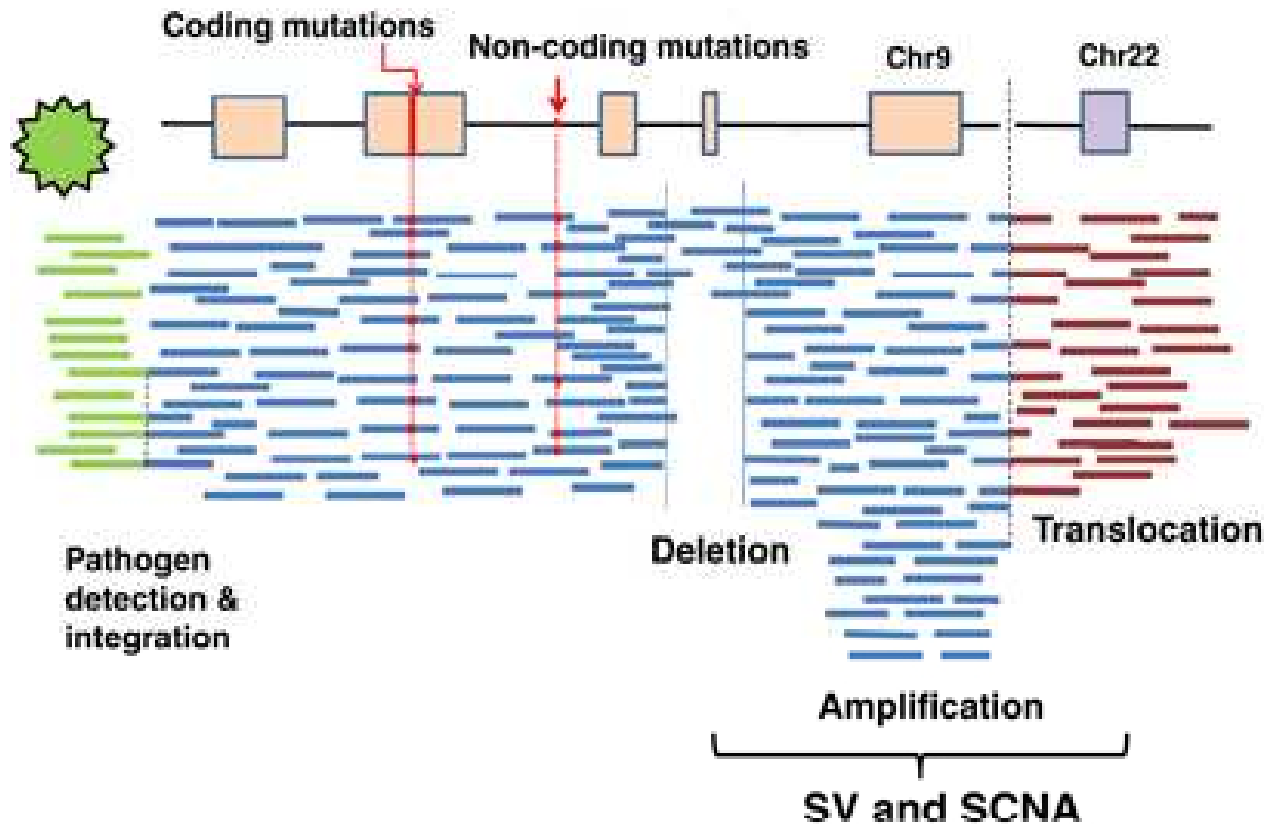
# Lecture 3b : Structural variants

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[vojtech.bystry@ceitec.muni.cz](mailto:vojtech.bystry@ceitec.muni.cz)

# NGS data analysis

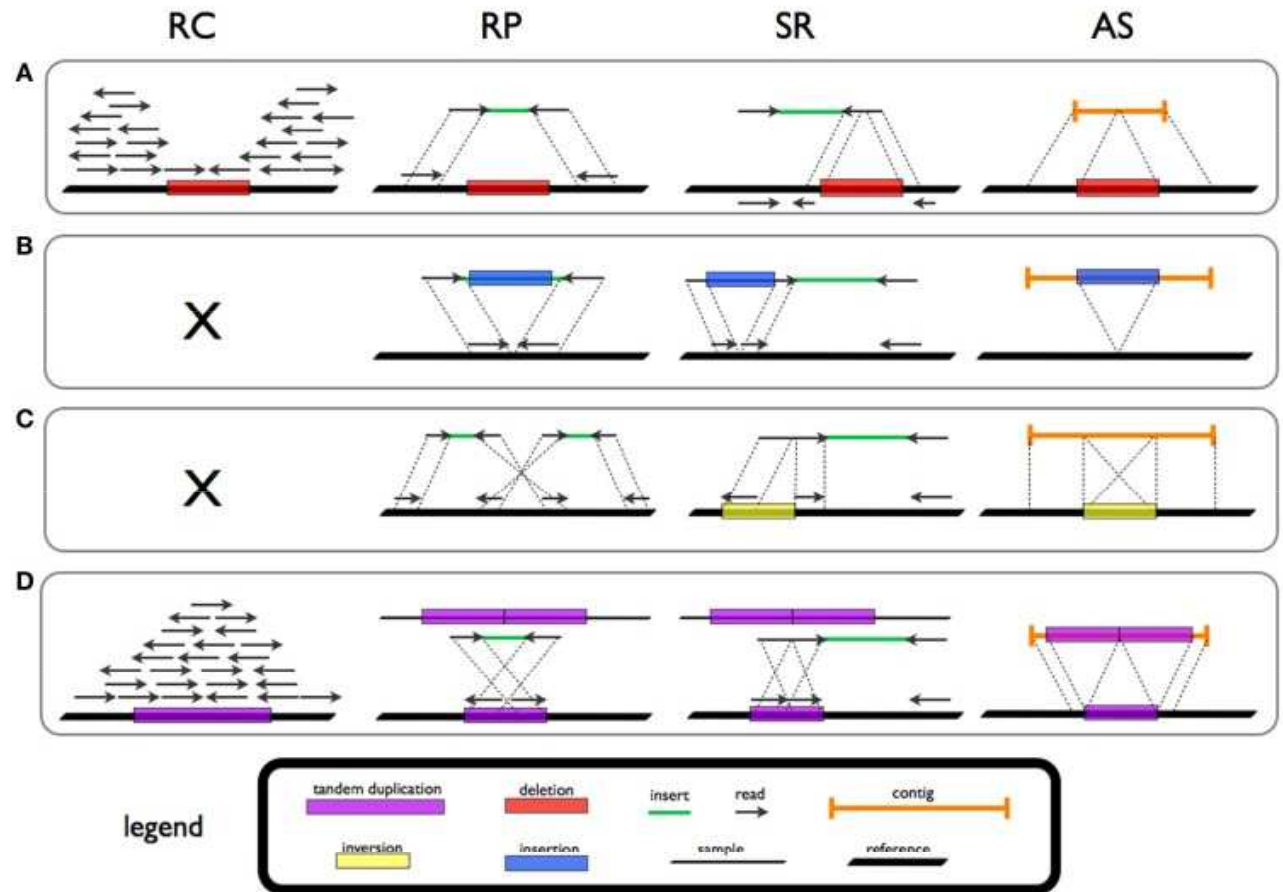


# Structural variants calling



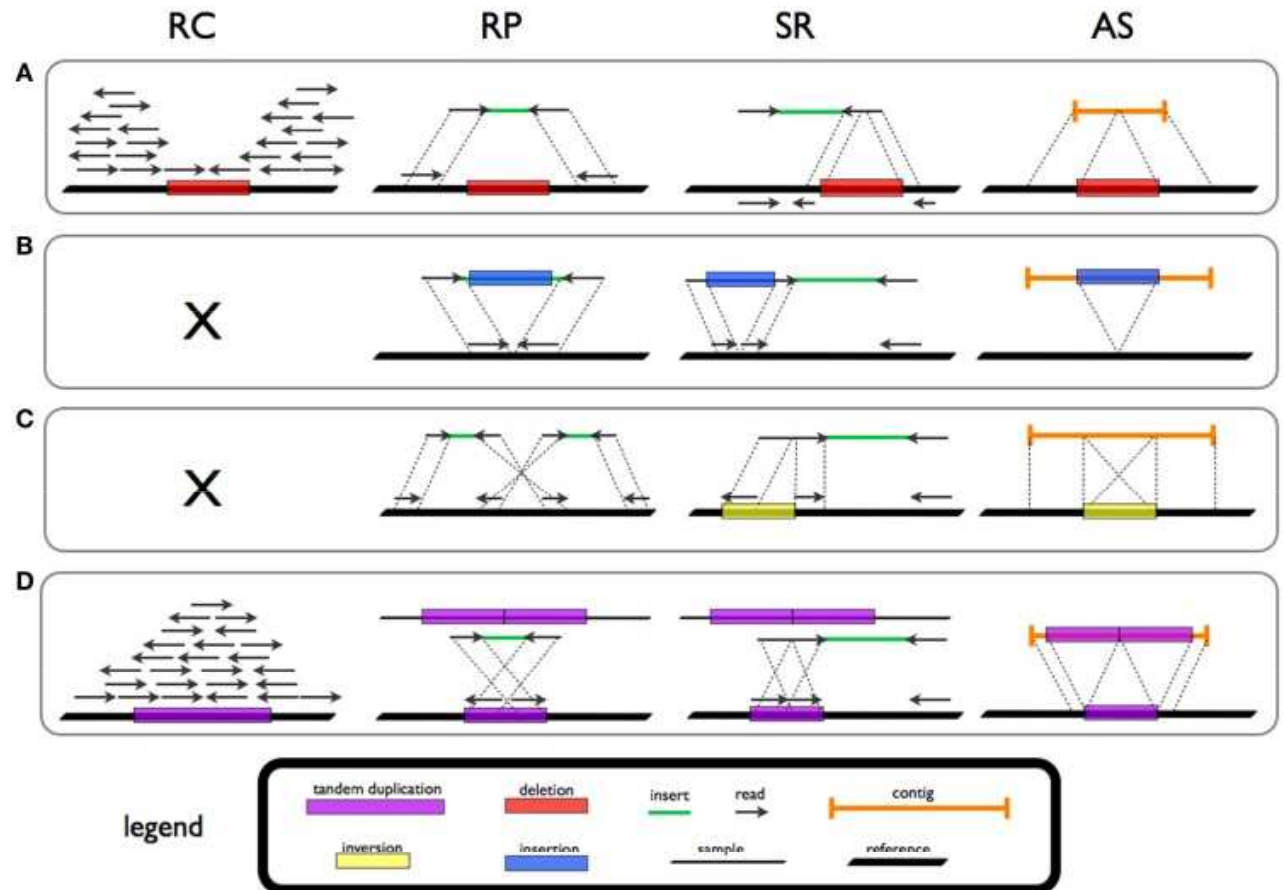
# Structural variants calling

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

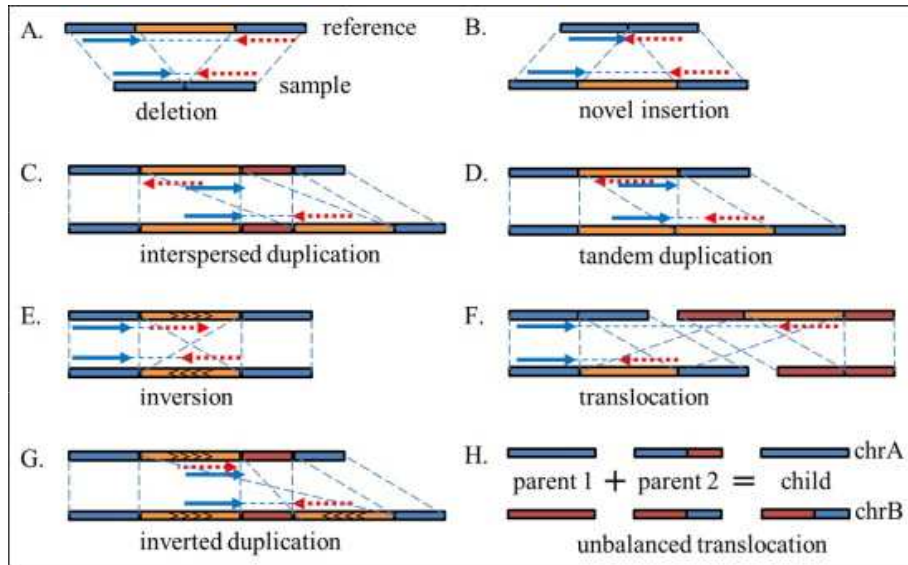


# Structural variants calling

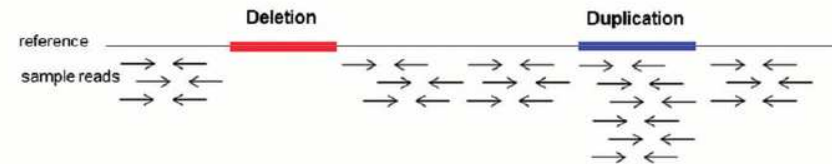
- RC = Read count
  - Copy number variants (CNV)
- RP = Read pair
  - Pair-end sequencing
- SR = Split reads
- AS = Assembled read
  
- A = DELETION
- B = INSERTION
- C = INVERSION
- D = DUPLICATION



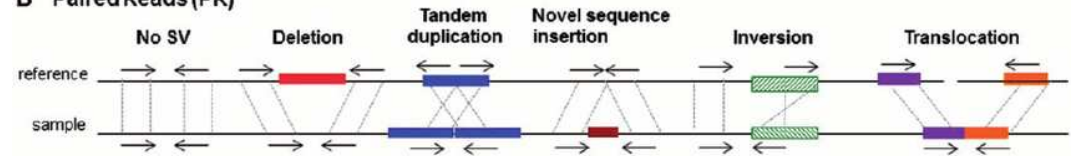
# Structural variants calling



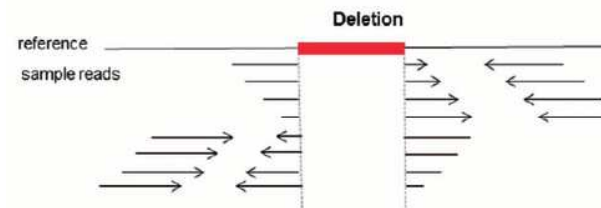
## A Read Depth (RD)



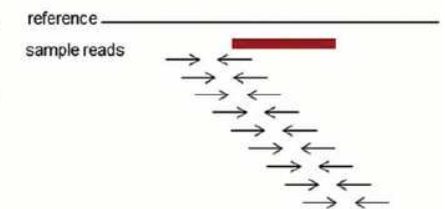
## B Paired Reads (PR)



## C Split Reads (SR)

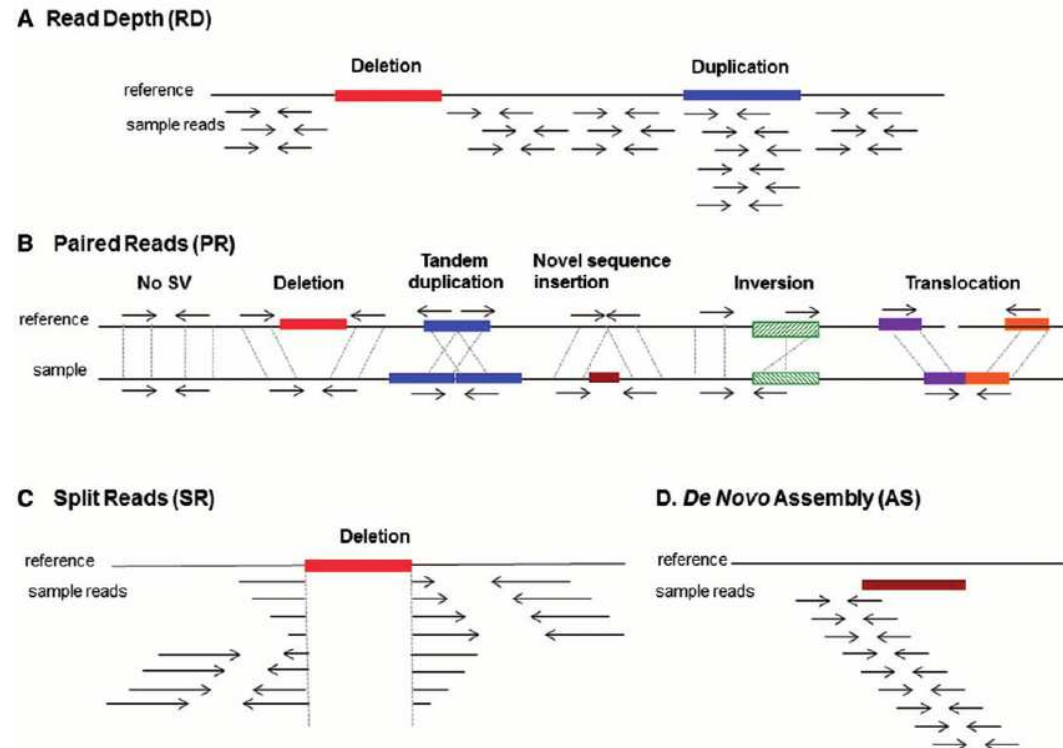


## D De Novo Assembly (AS)



# Structural variants calling

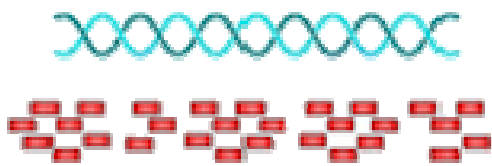
- Copy number variants (CNV)
  - Copy number analysis (CNA)
- Structural variants (SV)
  - Discordant reads analysis
- Fusion genes analysis
  - SV in coding sequence
  - From RNA-seq
  - Medical application



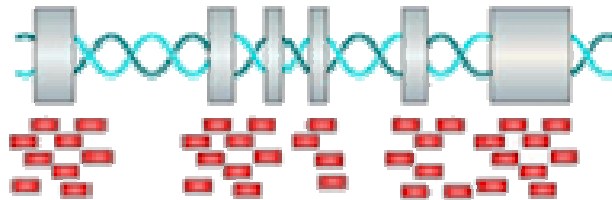
# Copy number variants

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

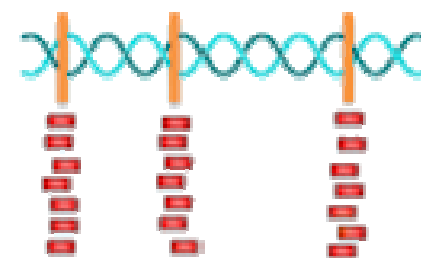
Whole genome sequencing



Whole exome sequencing



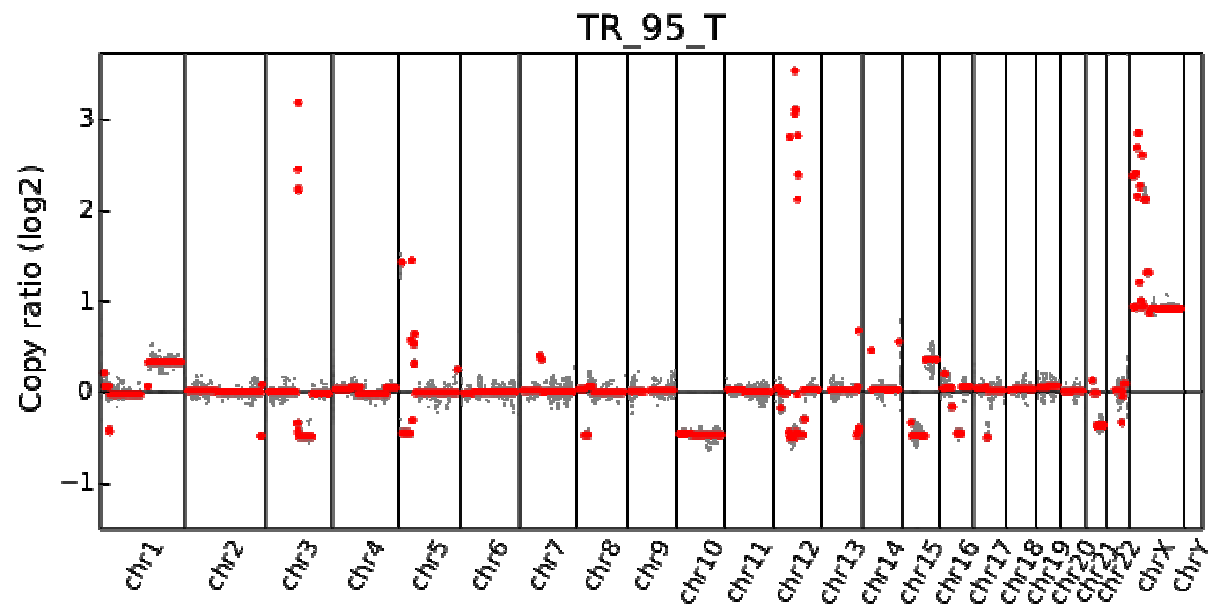
Targeted sequencing



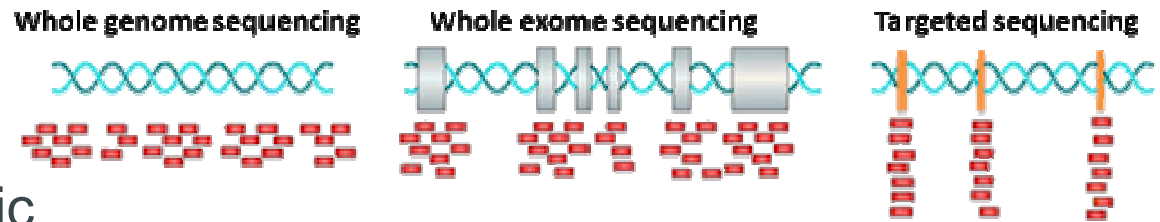


# WGS copy number variants

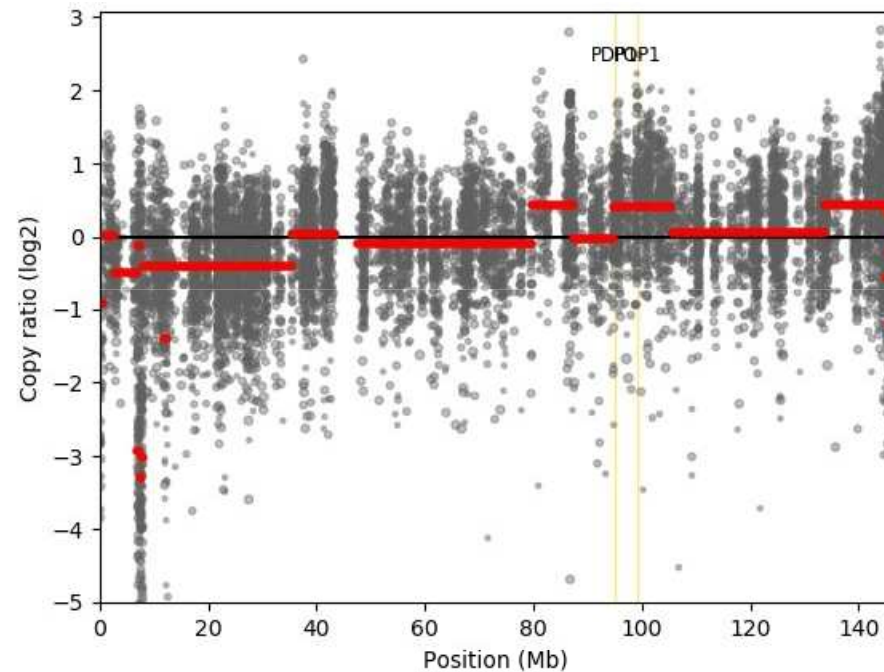
- Running window
- Normalization to the absolute coverage



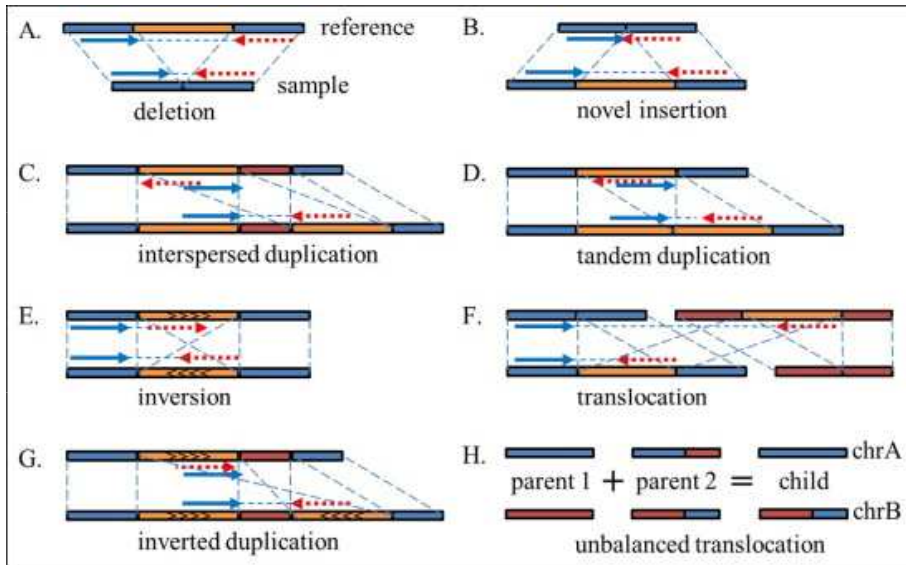
# PCR amplified CNV



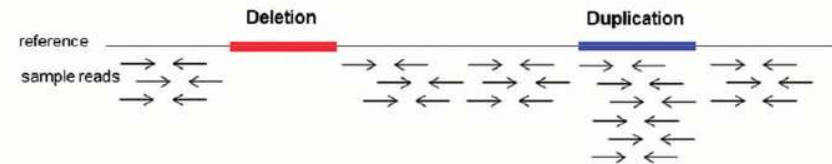
- Individual analysis problematic
- Somatic tumor-normal pairs
- Panel of (normal) samples



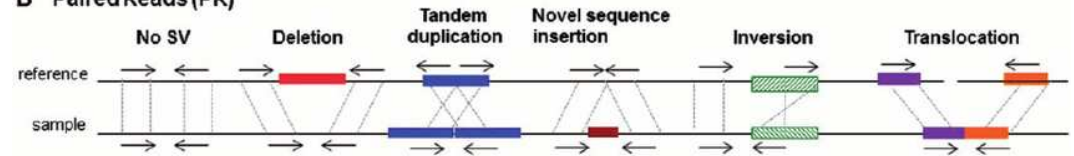
# Structural variants calling



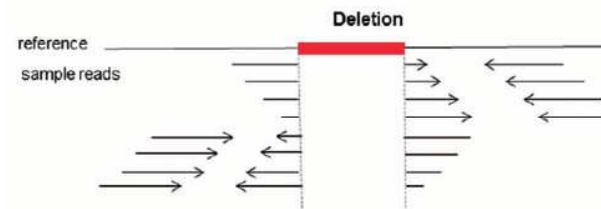
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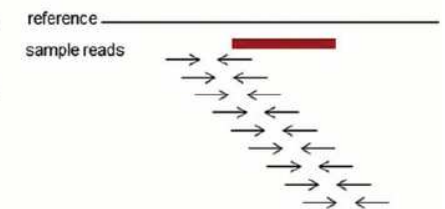
## B Paired Reads (PR)



## C Split Reads (SR)



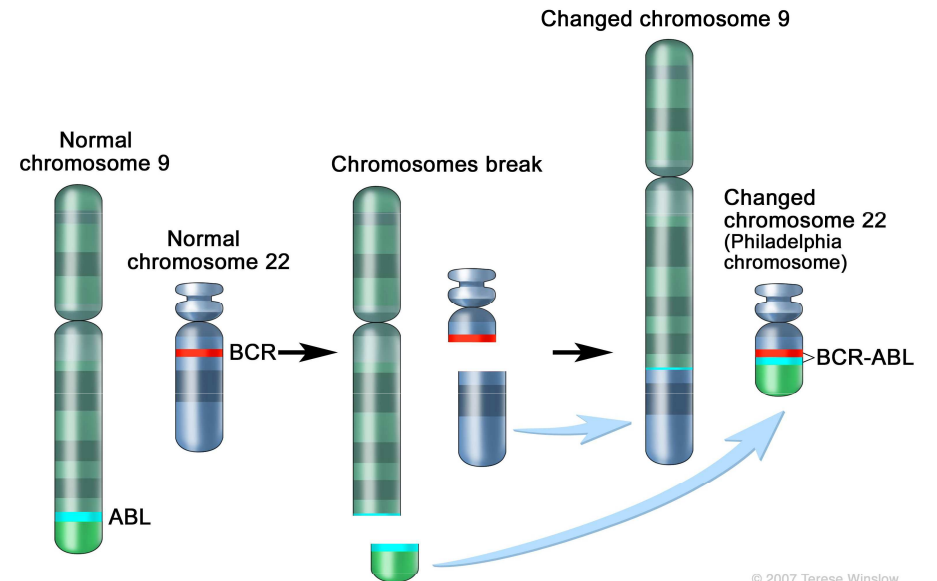
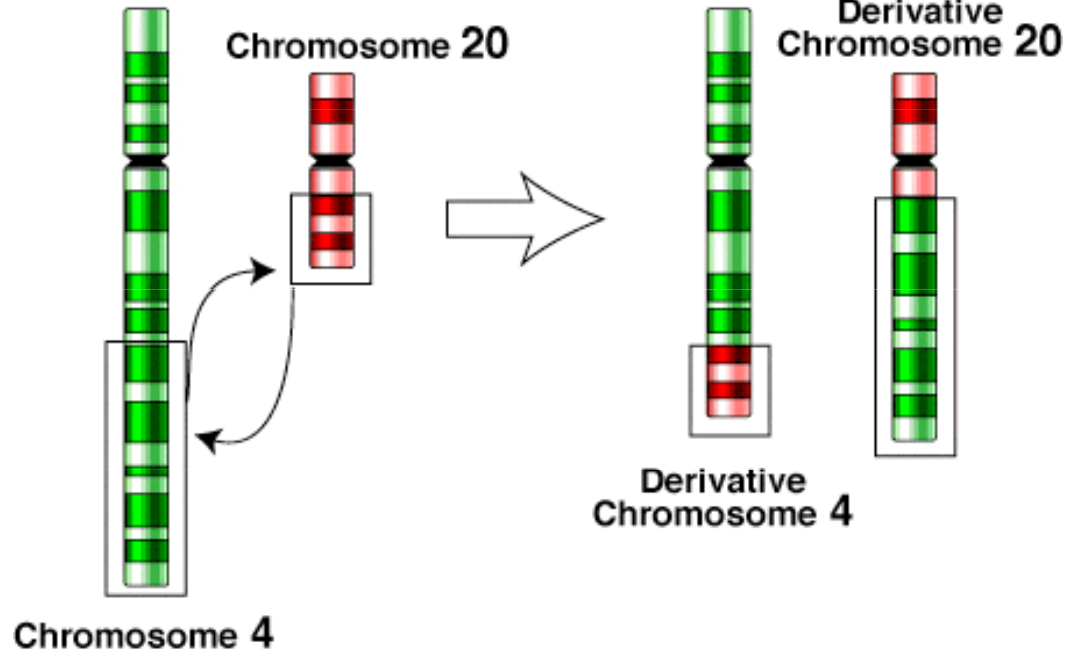
## D De Novo Assembly (AS)



# Fusion genes

Before translocation

After translocation



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Thank you for your attention!