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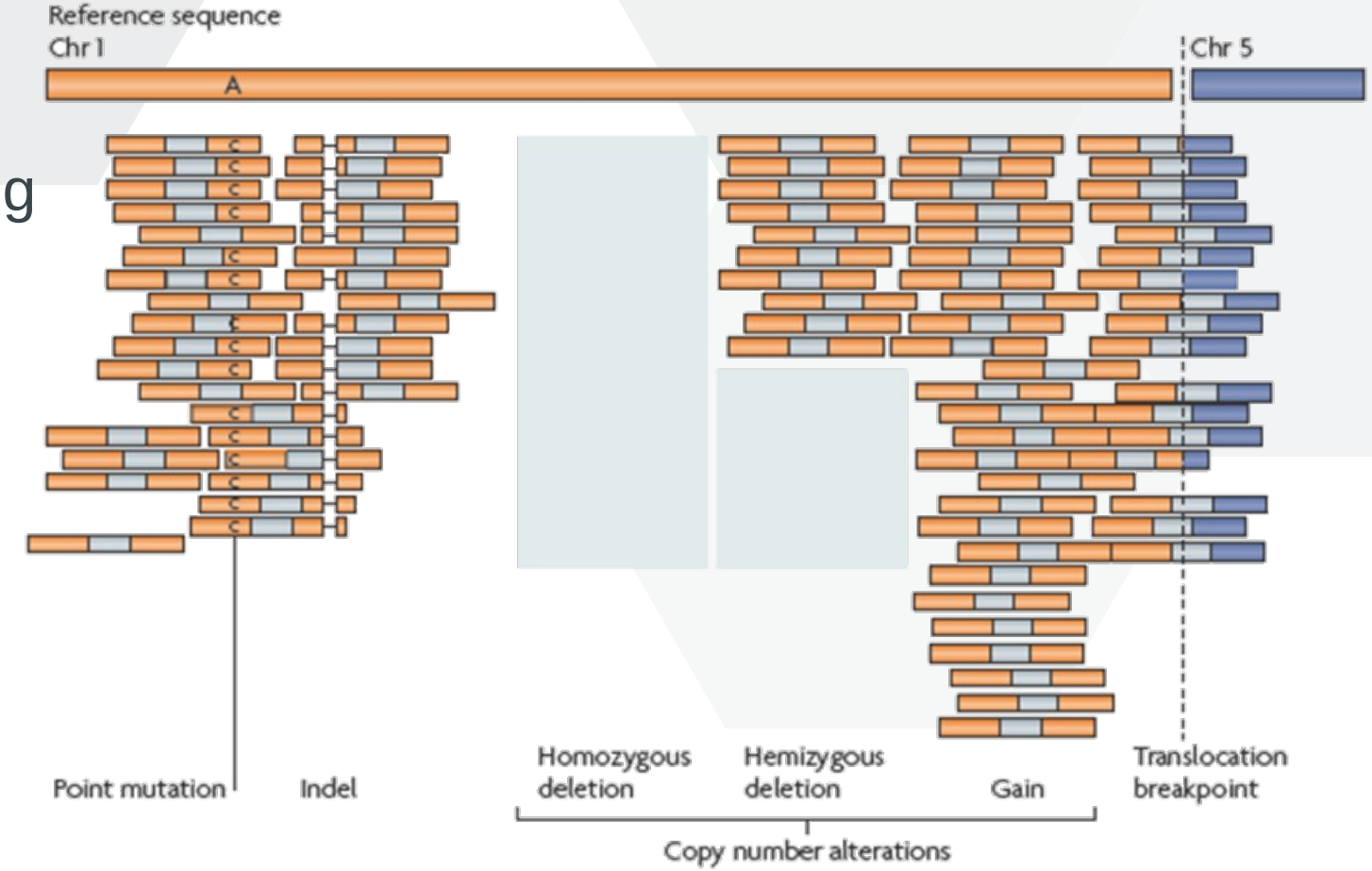
DNA re-sequencing

DNA re-sequencing

Comparisson with reference sequence (genome)

- WGS – whole genome sequencing
- WES – whole exome sequencing
- Genes, gene panels
- SNV – single nucleotide variant
- CNV – copy number variant

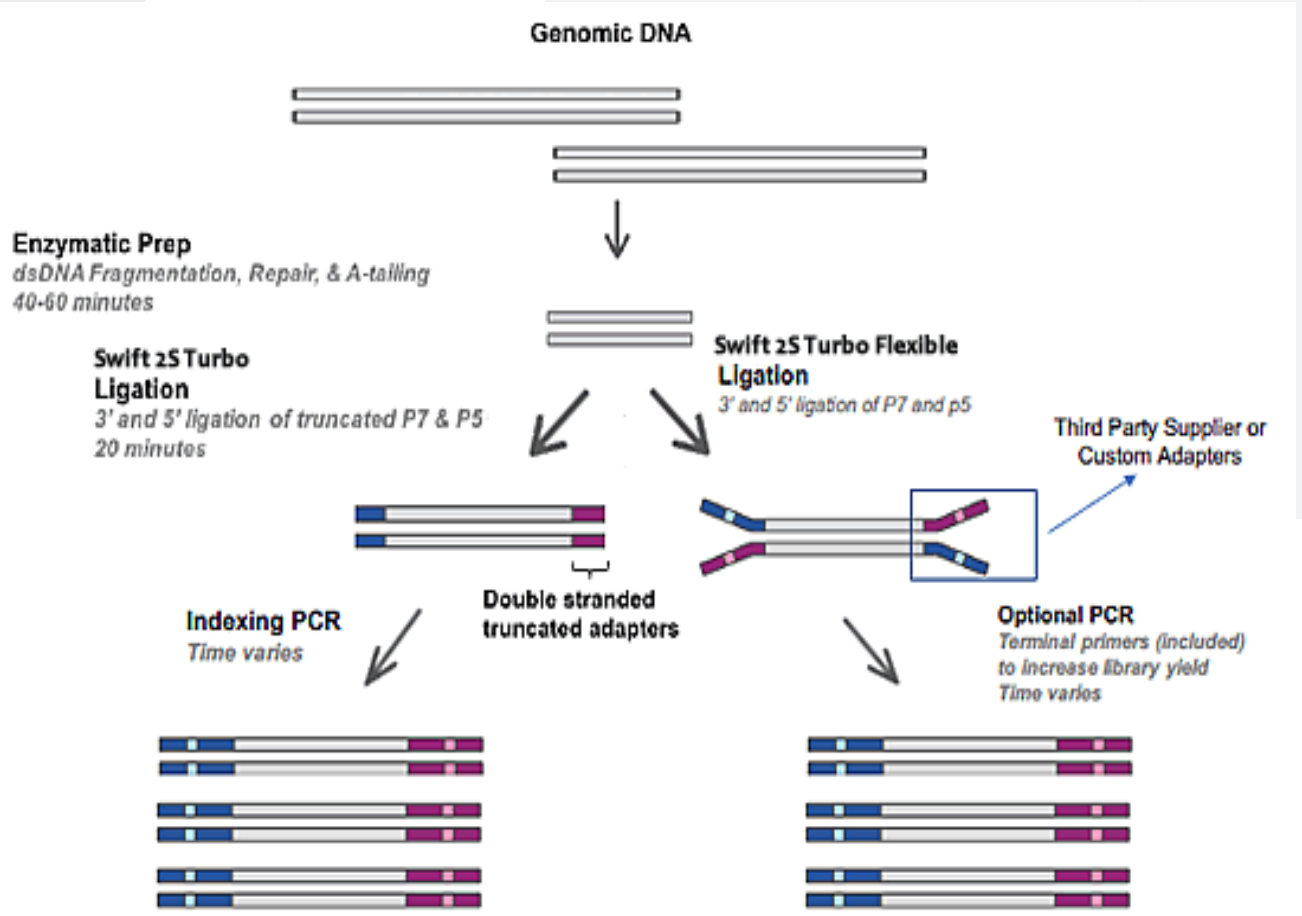


Nature Reviews Genet. 2010 Oct ;11(10):685-96.

DNA re-sequencing

WGS – whole genome sequencing

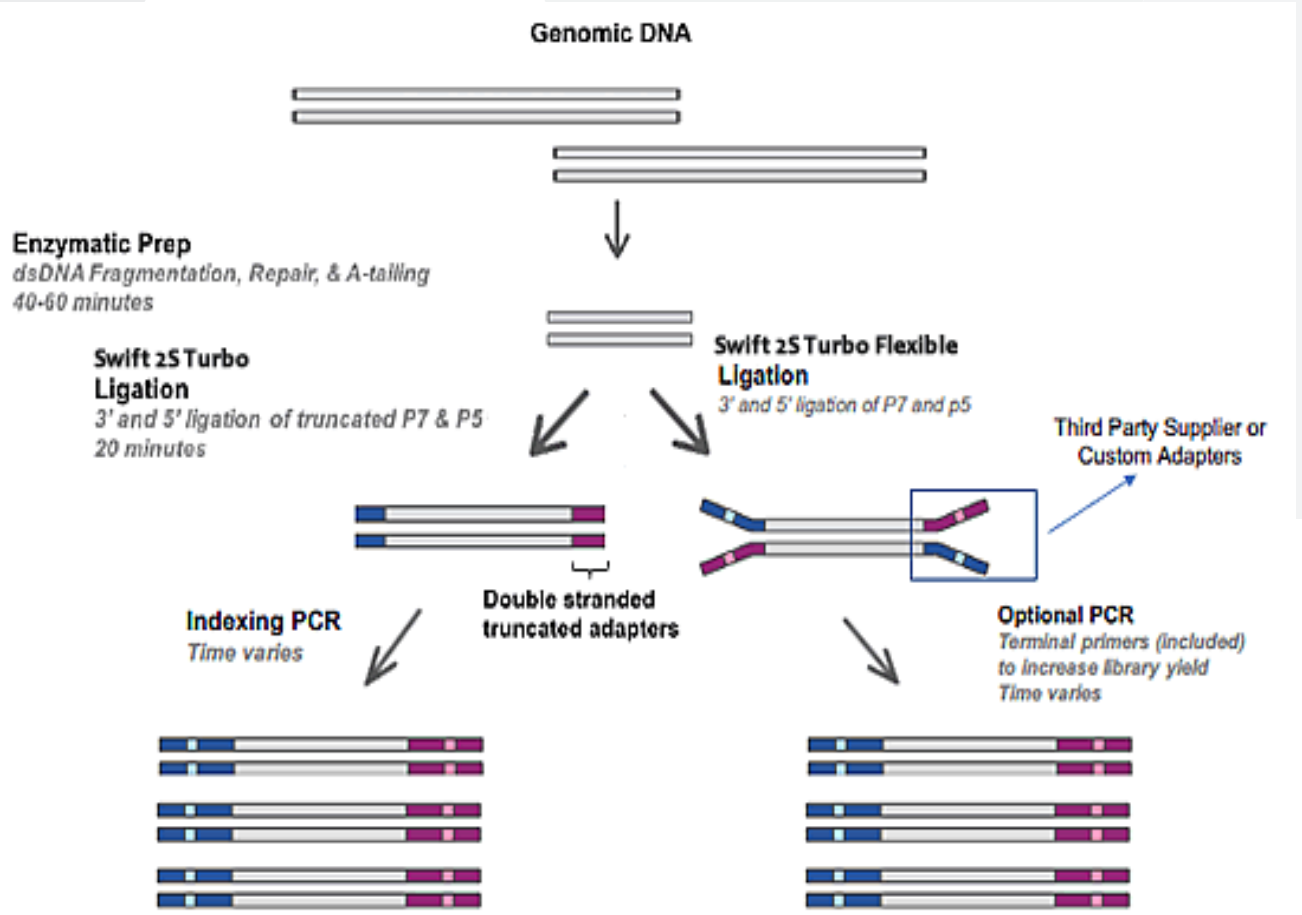
- DNA sequencing without selection
- Fragmentation
  - Ultrasound
  - Enzymes
  - Longer fragments (> 300bp)
- PCR-free – GC-bias
- Paired-end sekvencing
- Germline - 30x coverage ~ 100Gb



DNA re-sequencing

WES – whole exome sequencing

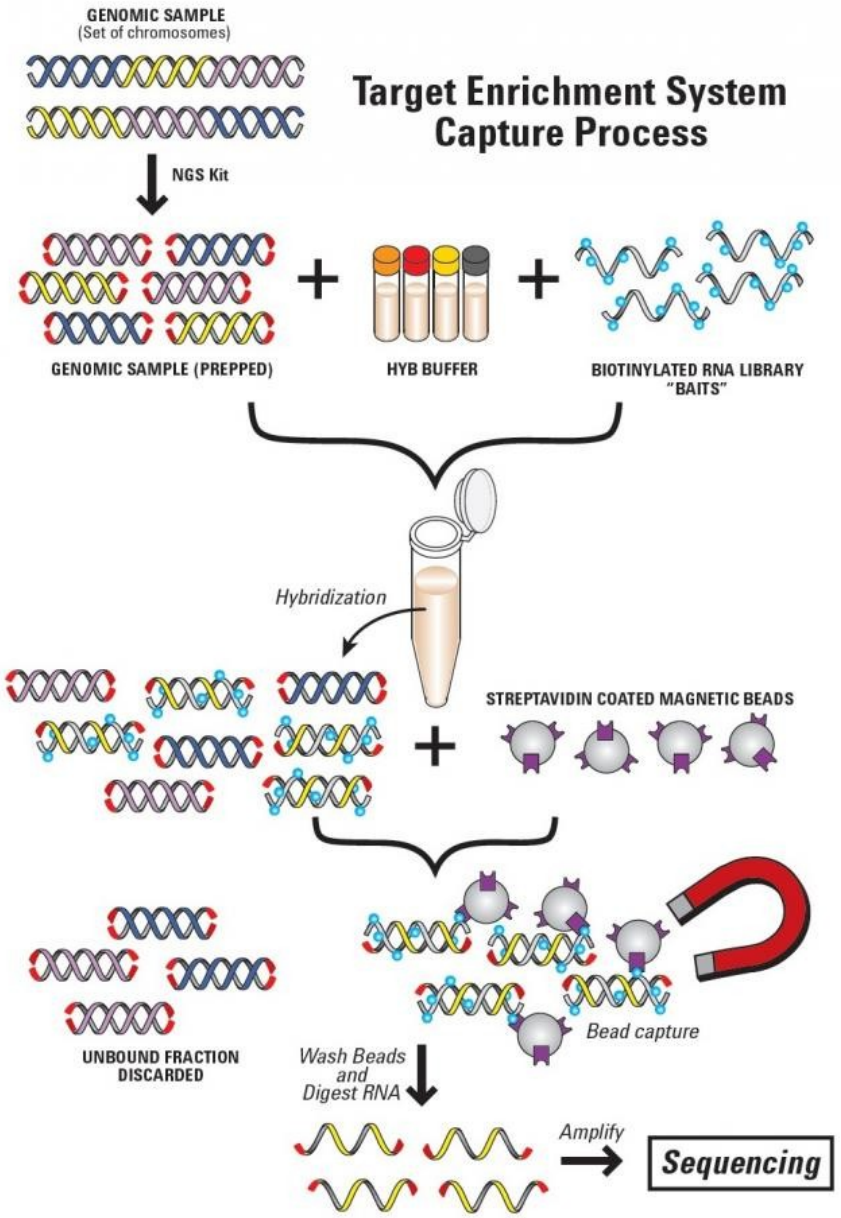
- Coding sequence selection
- Fragmentation
  - Ultrasound
  - Enzymes
  - Fragments 150-250bp
- PCR
- Paired-end sequencing



DNA re-sequencing

WES – whole exome sequencing

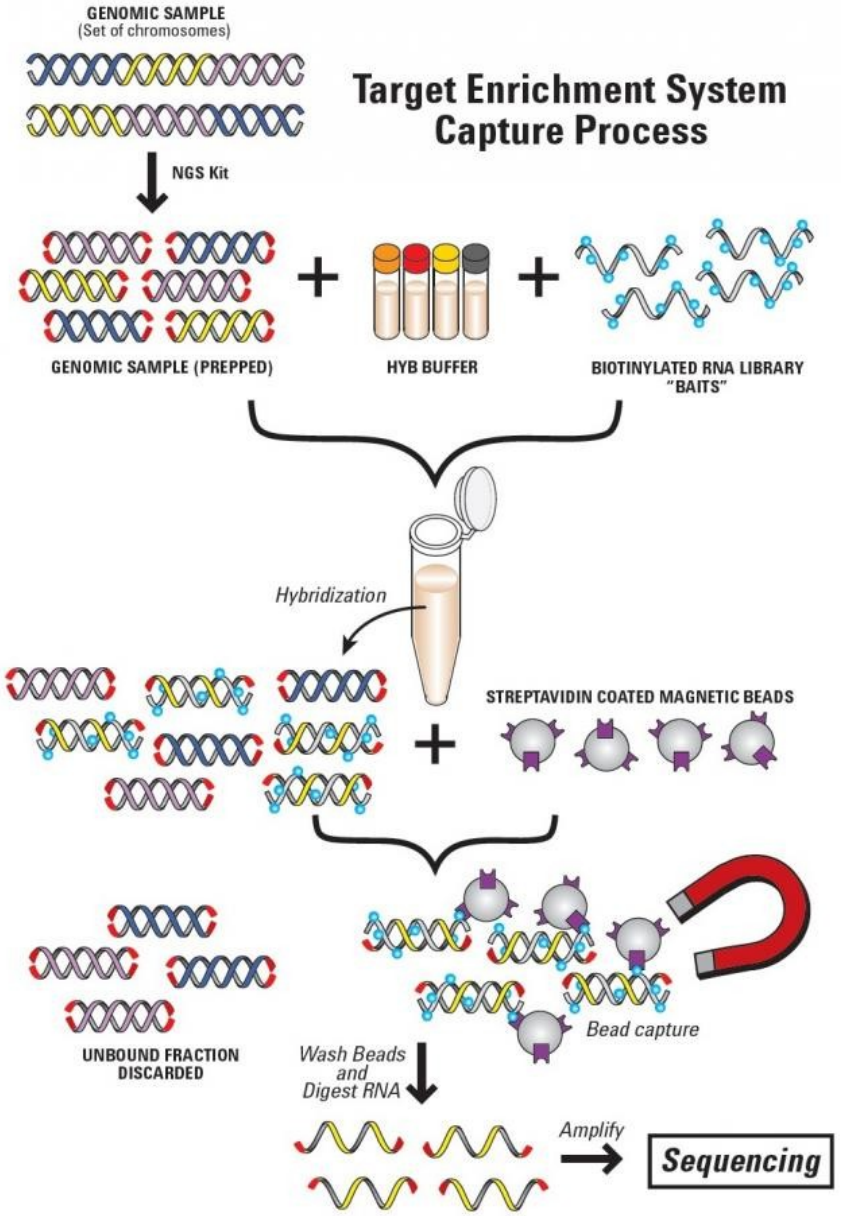
- Coding sequence selection
- Fragmentace
  - Ultrasound
  - Enzymes
  - Fragments 150-250bp
- PCR
- Paired-end sekvenace
- Germline - >50x coverage, >3Gb



DNA re-sequencing

Gene panels

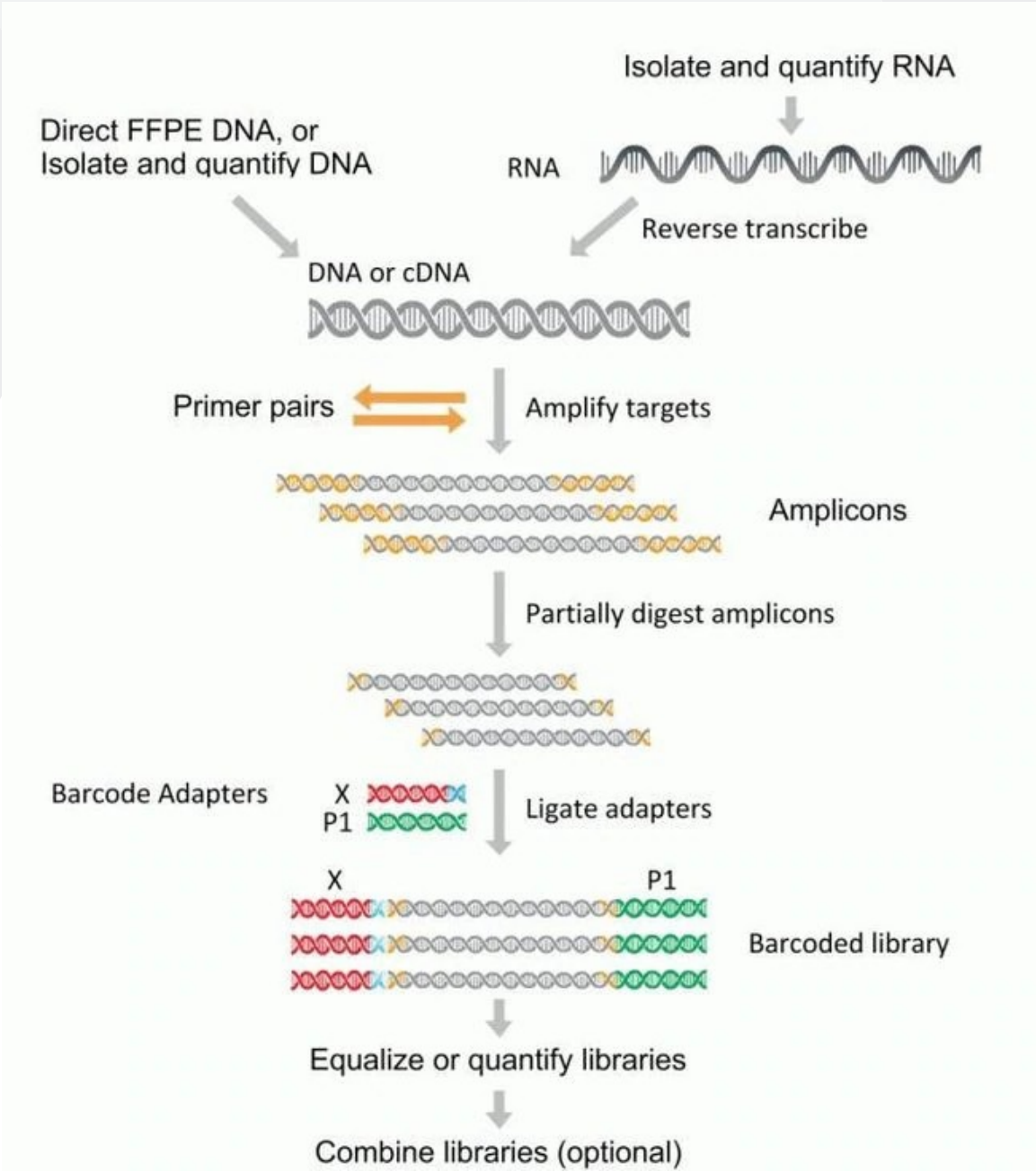
- Coding sequence selection
- Fragmentation
  - Ultrasound
  - Enzymes
  - Fragments 150-250bp
- PCR
- Paired-end sekvenace
- Germline - >50x coverage



DNA re-sequencing

Gene panels

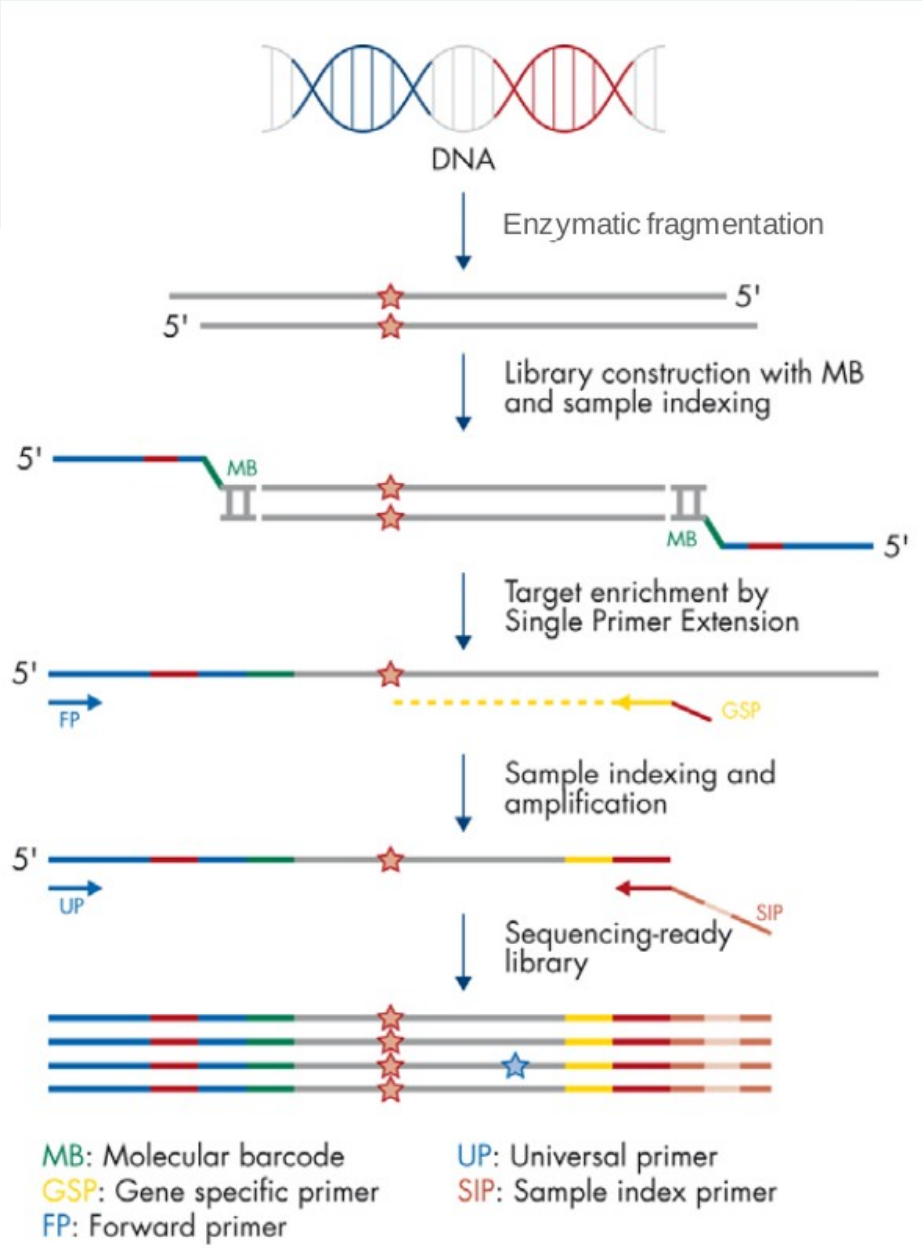
- Coding sequence selection
- **PCR enrichment**
- Single primer PCR
- Inverse PCR + capture
- Capture + probe extension
- Paired-end sequencing
- Germinální - >50x coverage



DNA re-sequencing

Gene panels

- Coding sequence selection
- PCR enrichment
- **Single primer extension**
- Inverzní PCR + capture
- Capture + probe extension
- Paired-end sekvenace
- Germinální - >50x coverage



DNA re-sequencing

Gene panels

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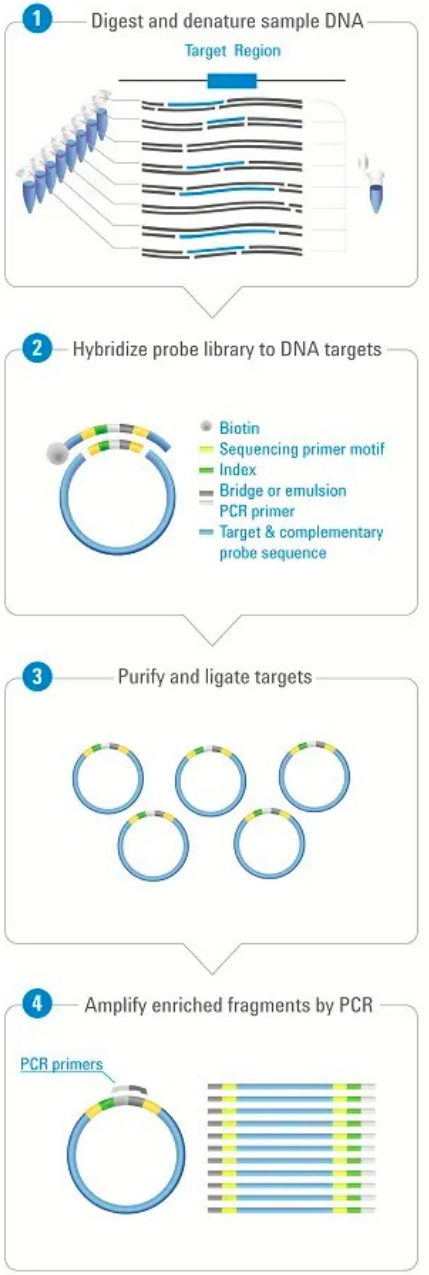

DNA re-sequencing

Gene panels

- Coding sequence selection
- PCR enrichment
- Single primer PCR
- Inverse PCR
- **Capture + probe extension**
- Paired-end sekvenace
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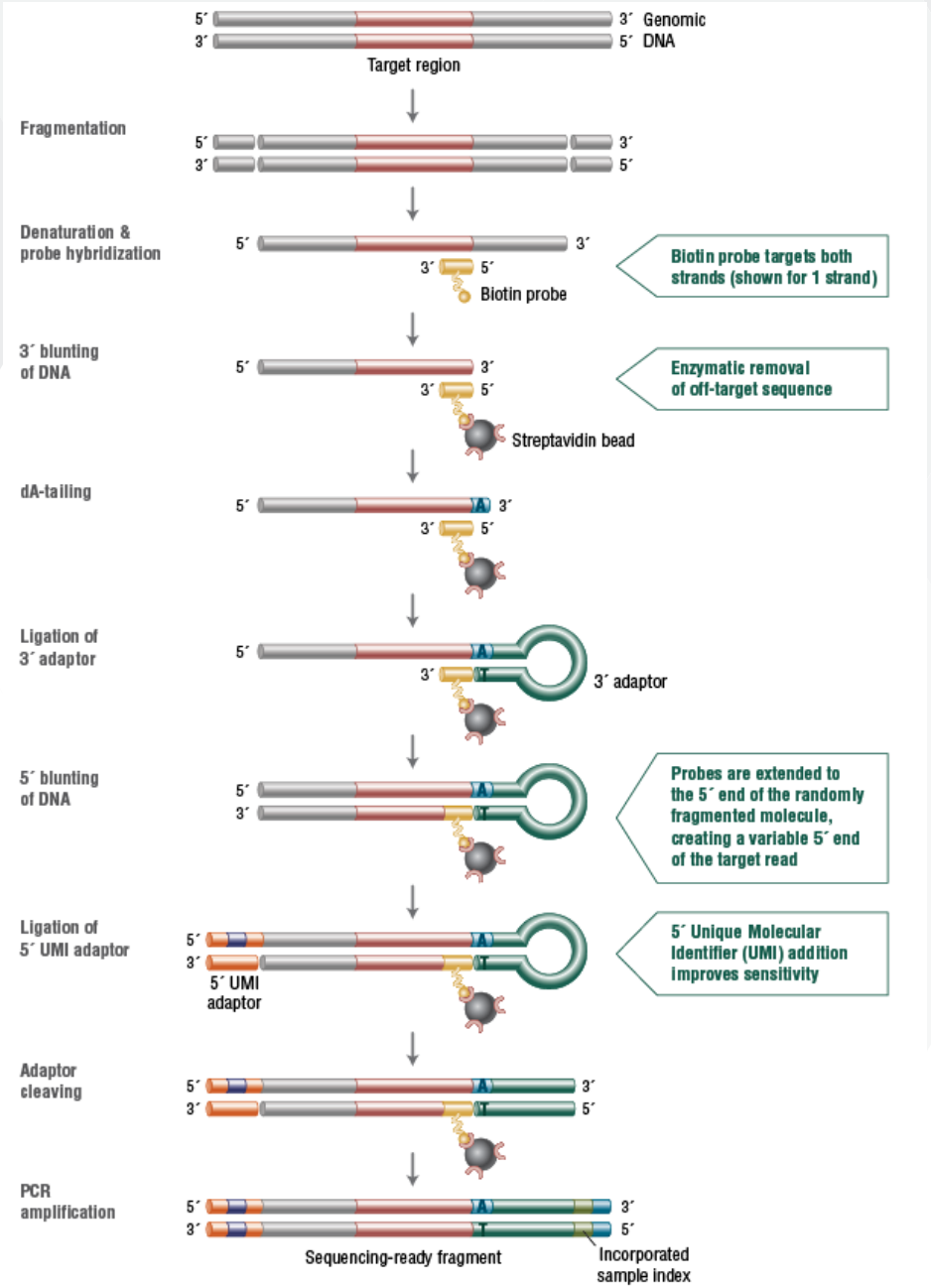
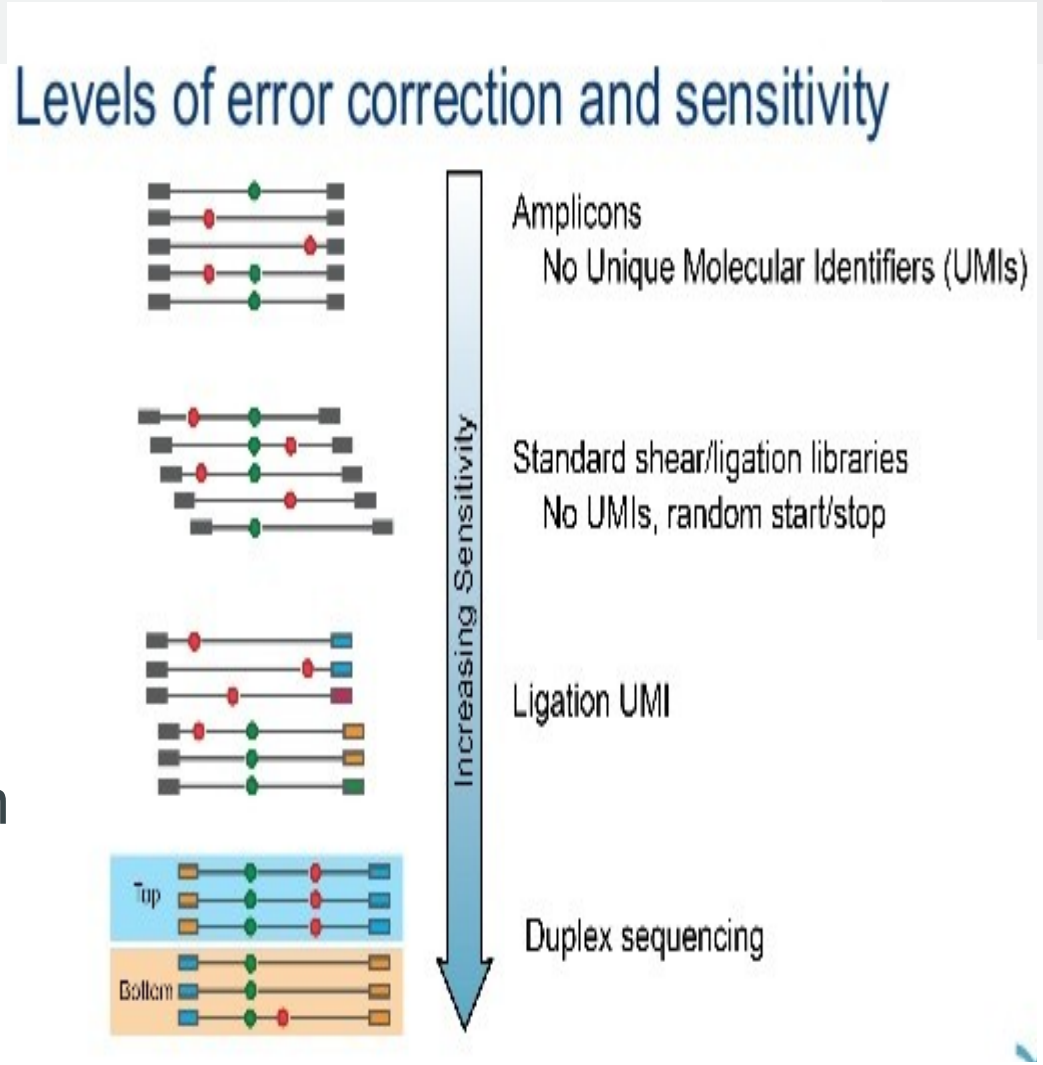


DNA re-sequencing

Somatic variants

- Tumor x Normal
  - WGS, WES, panels
  - Tumor only variants
- Tumor only
  - Panels, genes
  - Variants germline i somatic
  - Interpretation according to VAF, anotation
- UMI, Duplex-seq adaptors
  - Identification of PCR duplicates
  - Error correction



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Thank you for your attention

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