Transforming genomic medicine with Next Generation Sequencing

How NGS contributes to diagnosing heritable disease

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~10 000 Rare diseases - affecting fewer than 1 in 2000 people

80% Of rare diseases are genetic

~6% Of people will be affected by a rare disease at some point in their life

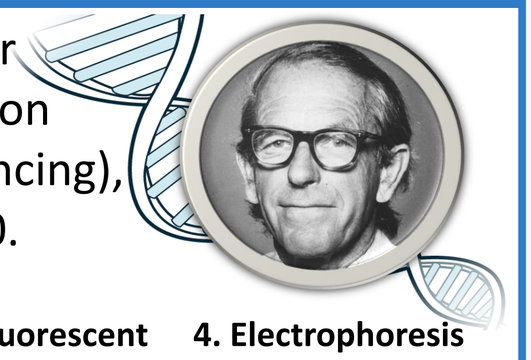
What is DNA sequencing?

Sequencing decodes the genetic blueprint of life, providing the foundation for diagnosing and treating diseases at their root cause.

We determine how the patient's DNA sequence has changed, allowing to diagnose genetic disease

How Sanger Sequencing works

The British biochemist Frederick Sanger developed the dideoxy chain termination sequencing method (i.e. Sanger sequencing), which won him the Nobel Prize in 1980.

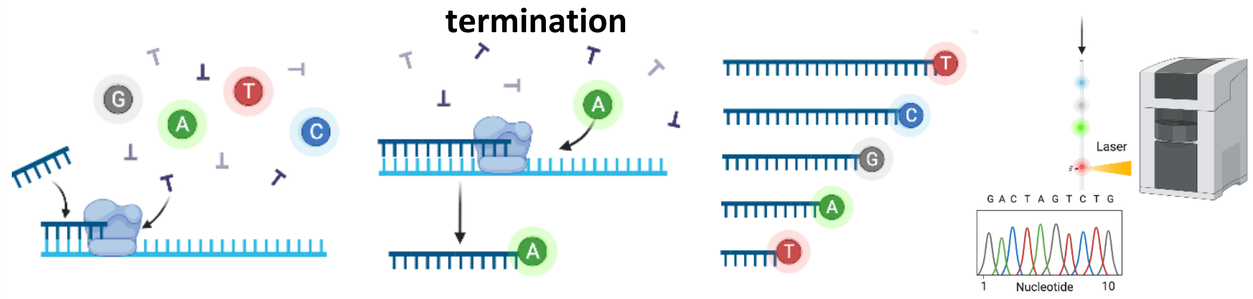


1. Primer annealing and chain extension

2. ddNTP binding and chain termination

3. Fluorescent DNA oligos

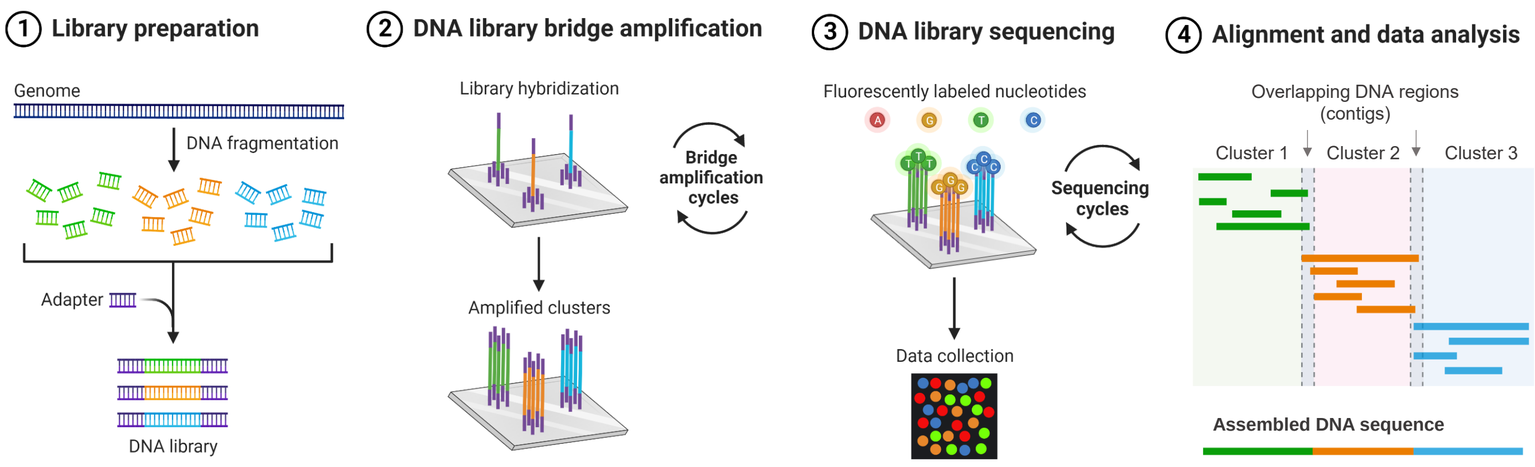
4. Electrophoresis and detection of



Low throughput ***Costly*** ***Time consuming***

NGS: Sequencing by synthesis

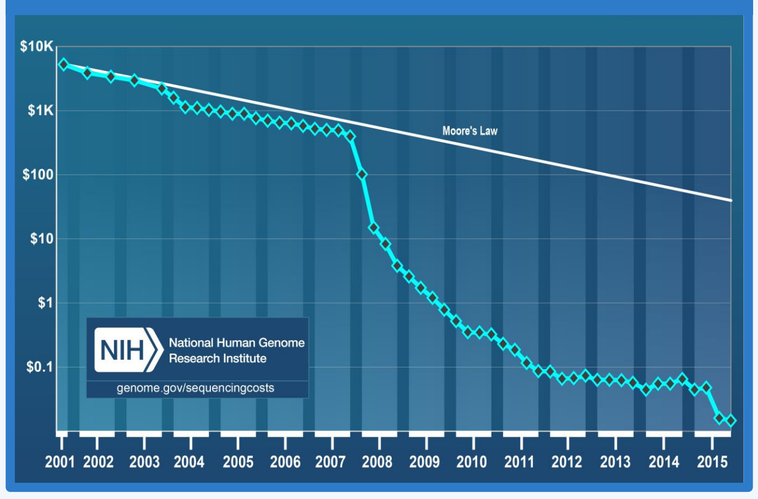
Sequencing by synthesis (NGS) was developed in the mid-2000s. DNA strands are copied, and each nucleotide is identified as it is added to the growing strand. Fluorescently labelled nucleotides emit a signal when added, which is detected by the sequencing machine. This process happens simultaneously for millions of DNA fragments.



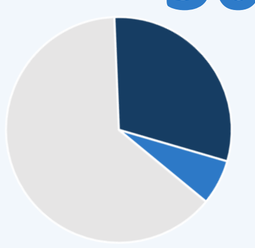
Benefits of NGS sequencing:

- **Fast:** results in hours instead of weeks
- **Cost effective**, sensitive and versatile
- **Large scale**: massively parallel, suitable for small panels to whole genome sequencing - ***generates a lot of data!***

Cost per raw Mb of DNA



Our Whole Exome Sequencing service



30% Have pathogenic or likely pathogenic variants

This can inform patient management, potential treatments and family planning

6% Have variants of uncertain significance (VUS)

Underscoring the need for more research to strengthen evidence and improve interpretation

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