Basics of NGS Technologies

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Next Generation Sequencing

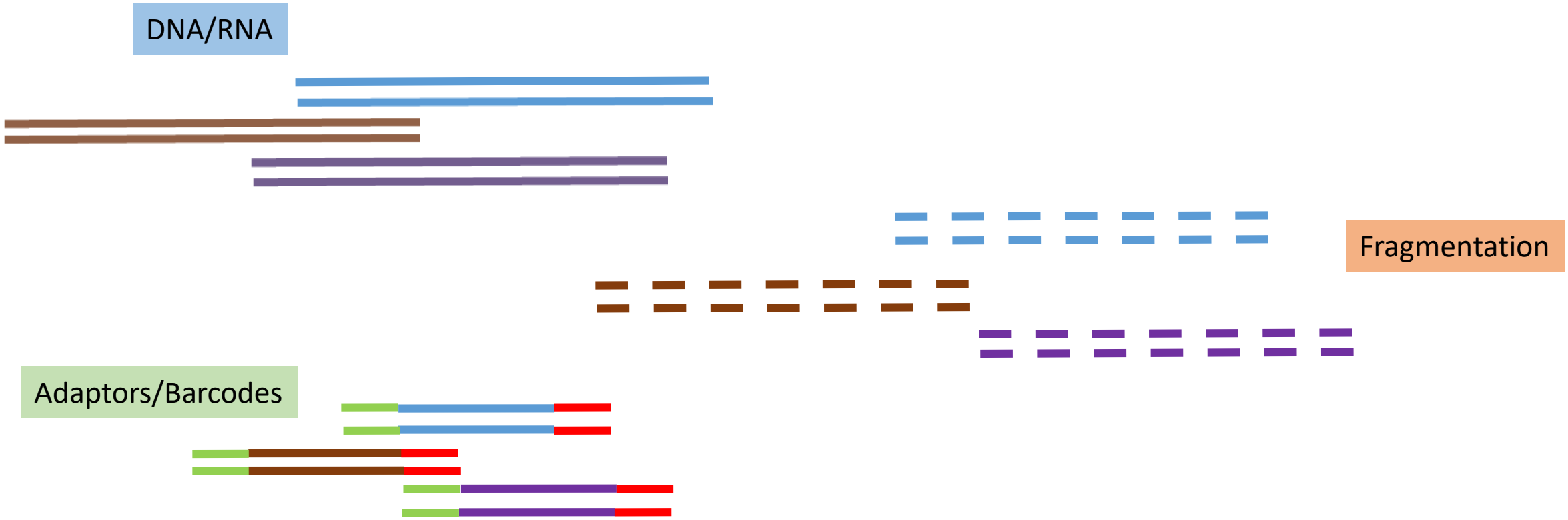
- Revolutionised medical science
- Enabled genomic studies/tests that were not possible
- Able to screen many genomic regions simultaneously
- Massively parallel sequencing
- Rapidly replacing traditional ways of mutation screening

NGS vs Sanger sequencing

	Sanger sequencing	**NGS**
The Good	Few genomic targets >20 per sample.	Multiple samples and targets many genomic regions
	Fast, Reliable and low error rate Validate NGS findings	Higher discovery and variant resolution
	Targeted quick analysis	More data with less DNA/RNA input
The Bad	Can only seq one gene region at a time or hot-spot regions	If sequencing few sequencing targets-Less cost effective
	Less cost-effective for high number of regions	More analysis time and complex
		Amplification bias, Sequencing errors

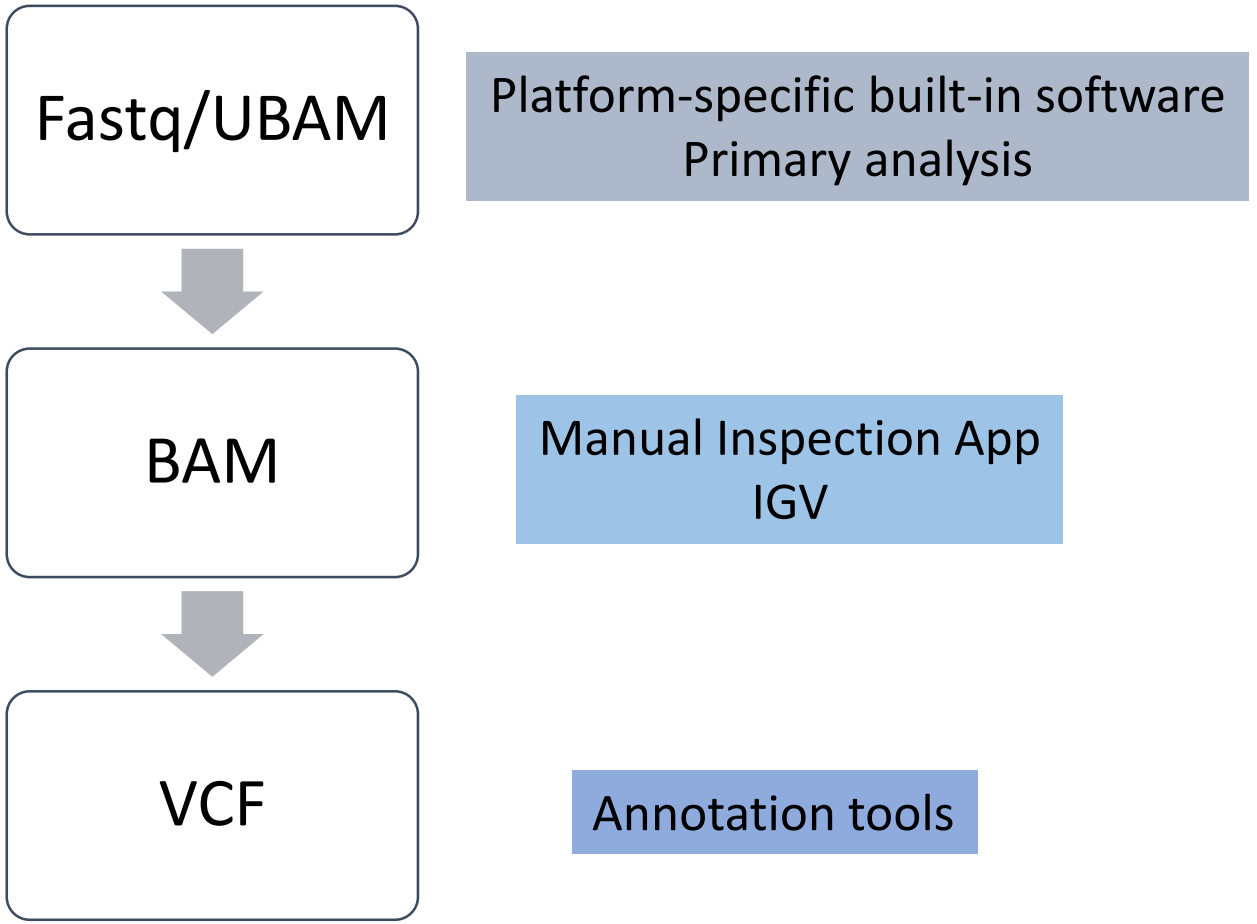
Basic processes

Library preparation



Sequence

Analyse


Fastq/UBAM
Platform-specific built-in software
Primary analysis
BAM
Manual Inspection App
IGV
VCF
Annotation tools

NGS Platforms

Short reads

And others……

Long reads

PacBio

Oxford Nanopore

Flongle	MinION	GridION (5 flow cells)	PromethION (48 flow cells)
			

Recap

- NGS - The way to go
- Involves three basic processes
- Various platforms to choose from

Next up

Differences in NGS Technologies…..