



Next Generation Sequencing (NGS)

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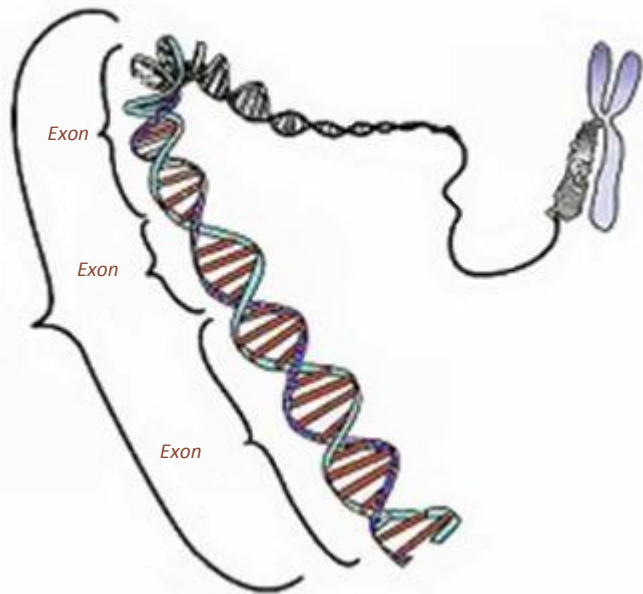
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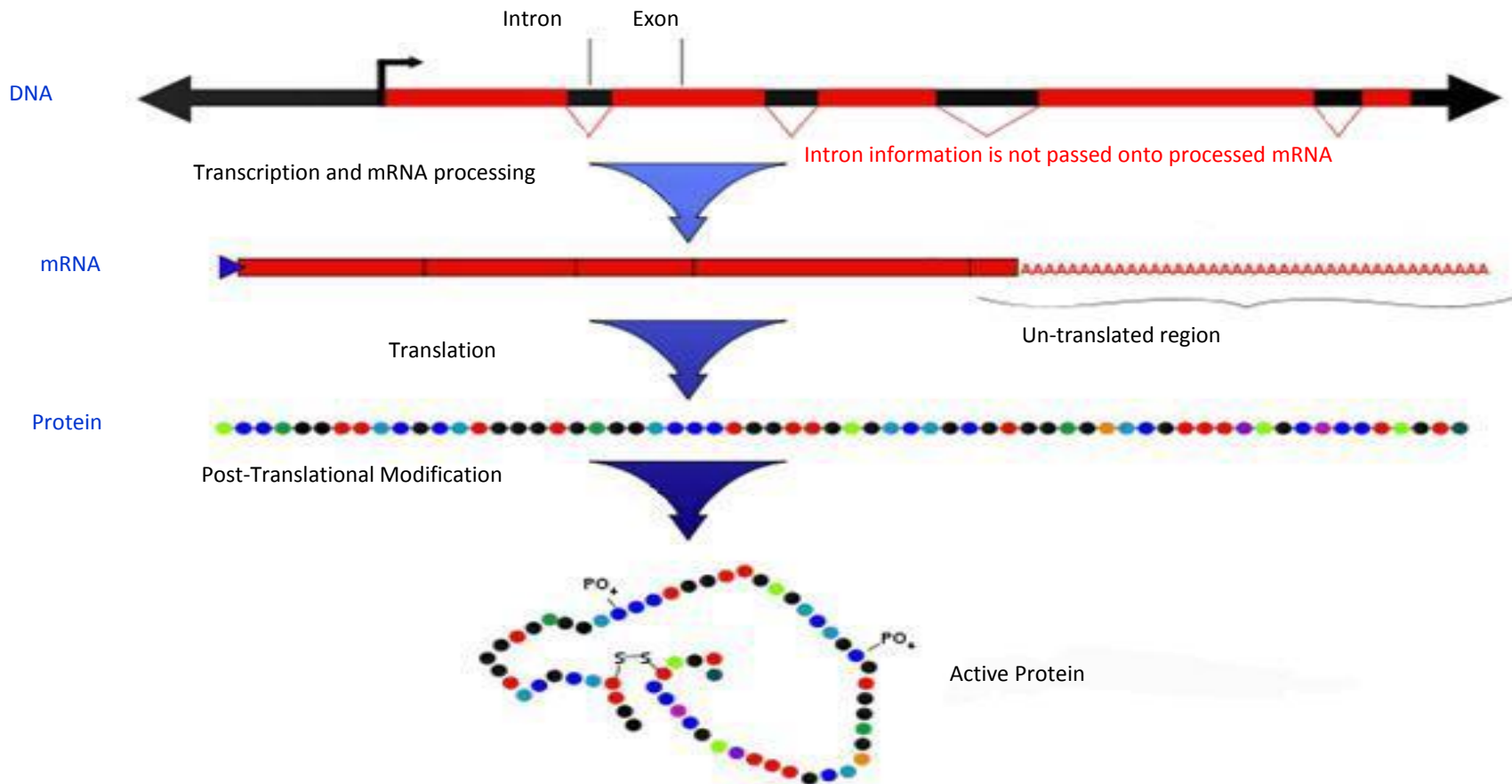
What is “DNA Sequencing”?



- various methods for determining the order of the nucleotide bases — **A**, **C**, **G**, **T** in molecule of DNA.



Basics of Transcription and Translation



What is NGS: next-generation sequencing

1953: Watson and Crick

1977: Sanger



1983: PCR



1994



1986: HGP



2001



2003
HGP



2008: NGS



1996

1986: sequencers



1984: DNA fingerprinting

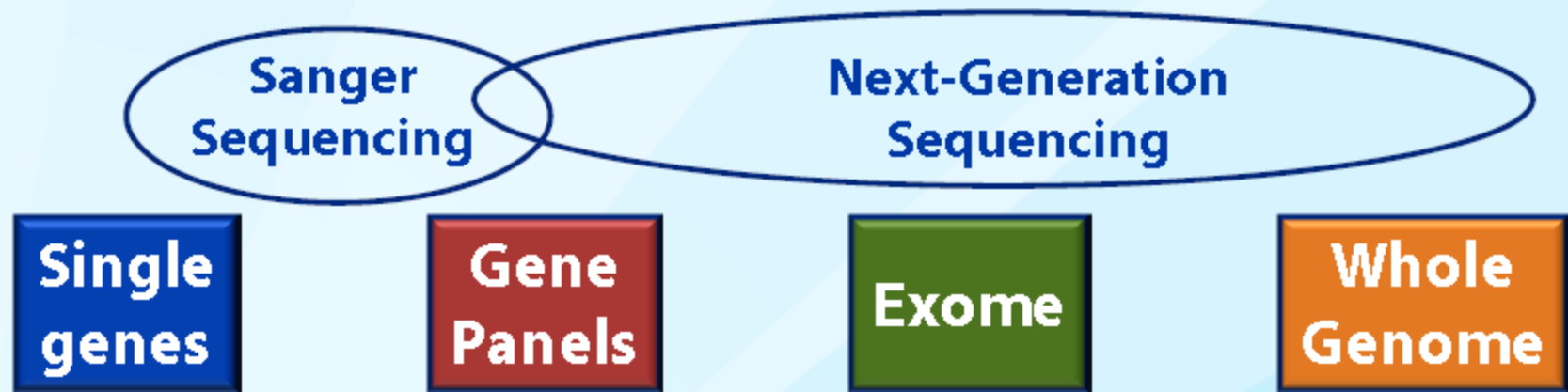


What is

Next Generation Sequencing?

- 1st Generation = Sanger Sequencing
 - high quality, long reads
 - slow, expensive
- •Human Genome Project: 1990 – 2004
- •Investments >3,000,000,000 \$ (~1\$/bp)
- 2nd Generation = Next Generation Sequencing
 - Millions of reads ,fast & cheap (low cost per bp)
 - needs sufficient knowledge of bioinformatics

From Sanger to Next-Generation Sequencing



Gene panels by Next-Generation Sequencing

- Congenital muscular dystrophy
- Congenital disorders of glycosylation
- X-linked intellectual disability
- Autism spectrum disorder
- Cardiomyopathy (Dilated / Hypertrophic)
- Mitochondrial encephalopathy
- Large scale carrier screening (in development)



Application Areas

- 1- Whole genome sequencing (WGS)
- Deeper understanding for genetic variations, drug response on a gene set, and many other complex/simple biological processes.
- 2- whole-exome sequencing (WES)
- The protein-coding portion of the genome is selectively captured and sequenced population genetics, genetic disease, and cancer studies.



Application Areas

- 3- Targeted Sequencing
- A subset of genes or regions of the genome are isolated and sequenced specific areas of interest such as cancer, cardiomyopathy, or autism.
- 4- Transcriptomics (RNA-Seq)
- mRNA, small RNA, noncoding RNA, or microRNAs



Application Areas

5- Epigenomics

- The study of heritable changes in gene activity caused by mechanisms other than DNA sequence changes. DNA methylation, small RNA-mediated regulation, DNA-protein interactions, histone modification.
- 6- NIPT