Whole Exome Sequencing (WES)

Overview

- Sequences only protein-coding regions (exons)
- Covers ~1-2% of genome (~30-50 Mb)
- Captures ~85% of known disease-causing variants

WES Advantages

- Lower cost than WGS
- Higher coverage per dollar
- Easier data analysis
- Smaller file sizes

WES Limitations

- Misses regulatory variants
- Limited structural variant detection
- Capture bias
- Non-coding regions excluded

Coverage	Cost	Diagnostic Yield
100-150X	**$300-500**	**25-40%**

Preferred for Mendelian disorders and cancer driver mutations