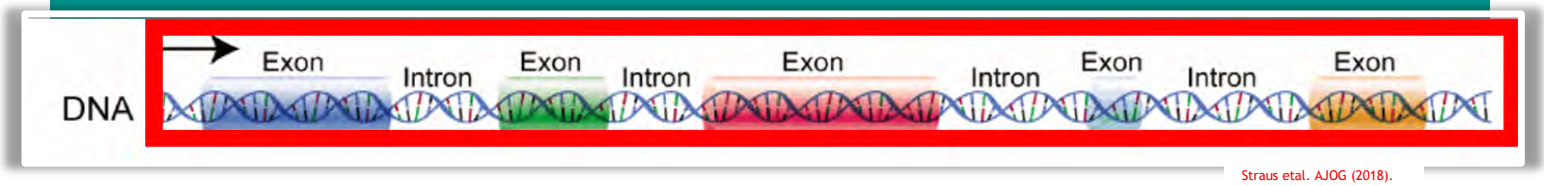
Whole Genome Sequencing

(WGS)



Straus etal. AJOG (2018). Volume 218, Issue 3, Pages 294-314.e2

Detects:

- Coding and Non-Coding Region
  - Single Nucleotide Variants (SNV)
  - Copy Number Variations (CNV)

Advantages:

- More reliable sequence coverage than WES
- More uniform coverage than WES
- Faster turn-around time than WES

Limitations:

- Large number of Variants of Unknown Significance (VUS)
- Incidental findings
- Regions of the genome with no/poor coverage
- Cost

Clinical Applications:

- High index of suspicion for genetic disease and patient characteristics not pathognomonic for one specific condition
- First tier genetic testing negative
- Caveats:
  - Patient critically ill
  - Results will inform management

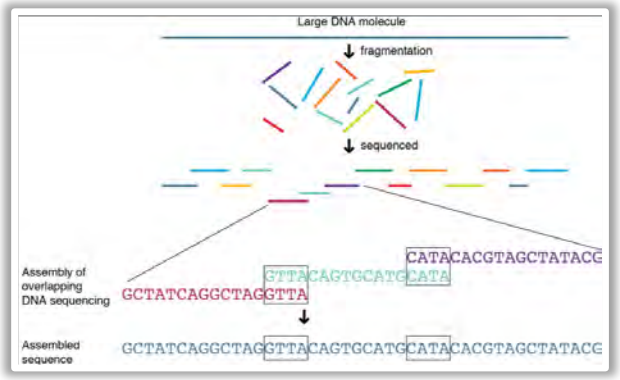
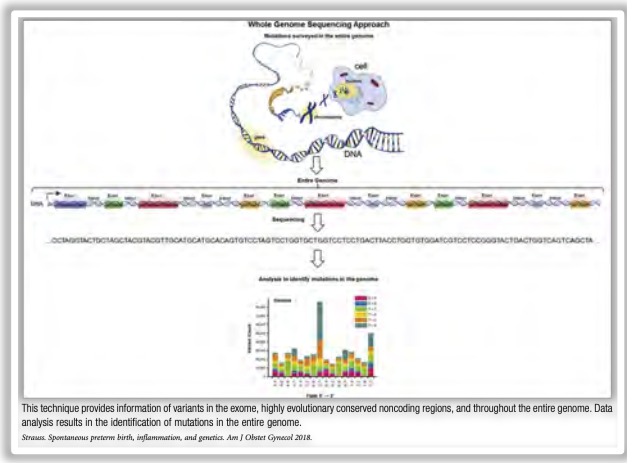


Image Courtesy of National Human Genome Research Institute.



This technique provides information of variants in the exome, highly evolutionary conserved noncoding regions, and throughout the entire genome. Data analysis results in the identification of mutations in the entire genome.

Straus. Spontaneous preterm birth, inflammation, and genetics. Am J Obstet Gynecol 2018.