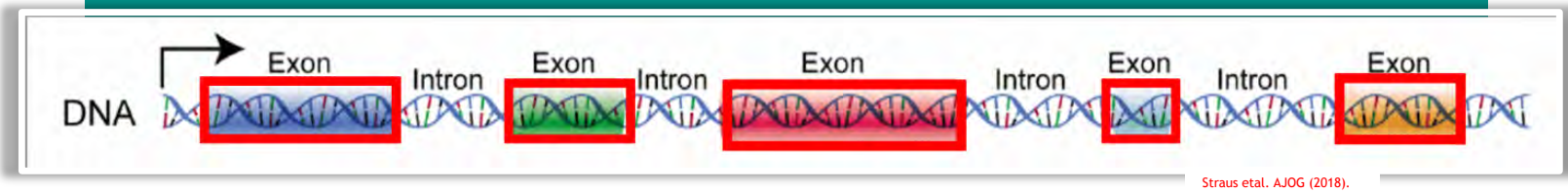
Whole Exome Sequencing

(WES)



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

Detects:

- Single Nucleotide Variants (SNV)
- Copy Number Variations (CNV)

Advantages:

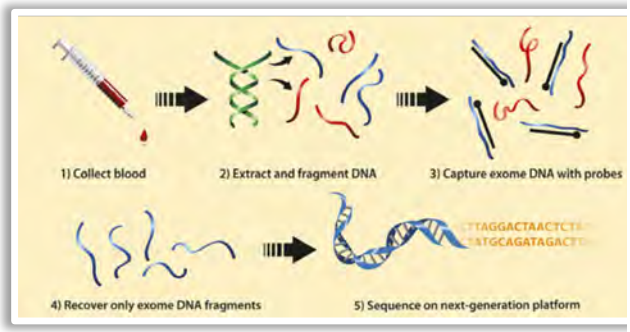
- >85% of disease-causing mutations are exonic

Limitations:

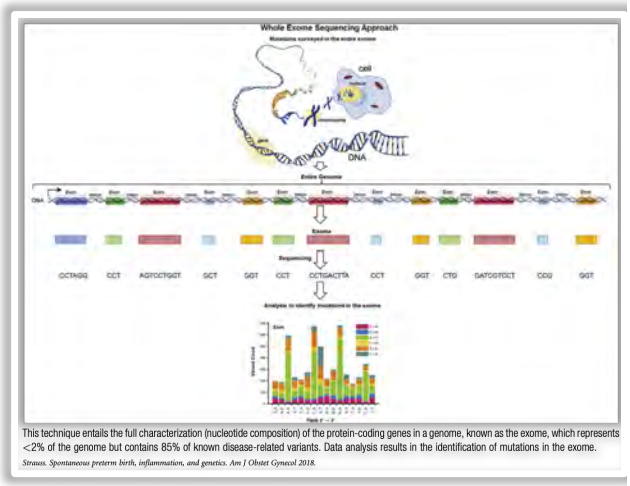
- Only coding variants evaluated (1.5-2% of the genome is sequenced)
- Variants of unknown significance
- Incidental findings
- Differences in the hybridization efficiency of capture probes
    - regions of the genome with no/poor coverage
- Turn-around time: weeks
- Cost

Clinical Applications

- High index of suspicion for genetic disease and patient characteristics not pathognomonic for one specific condition
- First tier genetic testing negative



From: The Journal of Bone & Joint Surgery 2013; 95(23): e185



This technique entails the full characterization (nucleotide composition) of the protein-coding genes in a genome, known as the exome, which represents <2% of the genome but contains 85% of known disease-related variants. Data analysis results in the identification of mutations in the exome.

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