

RTMS.e11 Next-Generation Sequencing: Methodology and Application

QUESTIONS

1. The basic methodological steps of NGS include the following:

- A. Template preparation, emulsion PCR, sequencing, data analysis.
- B. Template preparation, sequencing and imaging, data analysis.**
- C. Template amplification, sequencing and imaging, data analysis.
- D. Template preparation, sequencing and imaging, alignment to a reference genome.
- E. DNA fragmentation, sequencing, data analysis.

2. Advantages of targeted sequencing as opposed to full-genome, exome, or transcriptome sequencing include the following:

- A. Affordable and efficient for quickly interrogating particular genomic regions of interest.
- B. Provides a deeper coverage of genomic regions of interest.
- C. Can be utilized in deciding a therapeutic plan of action for both germline and somatic cancers.
- D. Detects and quantifies low-frequency variants such as rare drug-resistant viral mutations (e.g., HIV, hepatitis B virus, or microbial pathogens).

E. All of the above.

3. Applications of NGS in medicine include the following:

- A. Detecting mutations that play a role in diseases such as cancer.
- B. Identifying genes responsible for inherited skin diseases.
- C. Determining RNA expression levels.
- D. Identifying novel virulence factors through sequencing of bacterial and viral species.

E. All of the above.

ANSWERS

- 1. B.
- 2. E.
- 3. E.