INSIDE A CLINICAL MOLECULAR DIAGNOSTICS LABORATORY

DNA Sequencing In Cancer

WHO ARE WE?
We are molecular professionals — specialist physicians, doctoral scientists, and medical technologists that develop, perform and/or interpret molecular tests.

1. PATIENT SAMPLE IS COLLECTED FOR ANALYSIS
- Tissue (e.g., tumor, normal)
- Sterile body fluids (e.g., blood)
- Other sources (e.g., saliva)

DNA (deoxyribonucleic acid)
DNA is the hereditary material that encodes the information that produces the structural materials and molecules that perform the functions necessary for cell and organ function.
DNA is made up of individual bases (A, C, G and T) and their order, or sequence, determines the instructions encoded by the DNA. The DNA bases pair up together: A always with T, C always with G.

2. PATIENT SAMPLE IS PREPARED FOR MOLECULAR TEST
- The sample will be prepared for testing by:
  - Carefully breaking down the tissue/fluid sample into the individual components, including DNA, RNA, and proteins.
  - The necessary isolated component needed (e.g., DNA) is purified and cleaned to remove any contaminants.

3. MOLECULAR TEST IS PERFORMED
- The sample (e.g., the patient’s DNA) is assessed using a variety of molecular testing methods.
  - DNA is “sequenced” when a molecular professional uses specialized machines and laboratory techniques to “read” the bases along the DNA strand. This information is then stored in a computer file.
  - Depending on the unique case for each person, and the specific type of testing being performed, either small specific sections of DNA can be sequenced or all the DNA (the entire genome) can be sequenced in pieces.

4. MOLECULAR TEST RESULT IS ANALYZED AND INTERPRETED THEN COMPREHENSIVE REPORT IS GENERATED
- The DNA is analyzed and the key findings are reported back to the ordering physician and patient.
  - A specialist physician or a doctoral scientist analyzes the data generated through sequencing and prepares a comprehensive results report that includes the key findings, such as detections of alterations or biomarkers that may assist the patient and their clinician to decide on the best treatment or management plan.

5. OPTIMIZED CLINICAL TREATMENT FOR PATIENT

GERMLINE TESTING (also known as genetic or inherited disease testing)
- Sequencing the DNA in normal tissue (such as blood or saliva) to identify alterations that exist in all tissues of the patient’s body. These alterations can be passed on to the individual’s children.
- These alterations can have implications for:
  - A patient’s predisposition to developing certain types of cancer,
  - A patient’s response to specific drugs and dosages
- These results can help assess a patient's risk for particular diseases/disorders, lead to specific management of patient’s symptoms, or determine an ideal drug regimen and dosage.

CANCER BIOMARKER TESTING (also known as cancer genetic testing, molecular testing, and tumor profiling)
- A molecular test is performed on a sample of tumor tissue to identify genetic alterations, such as mutations or other biomarkers.
  - Unique biomarkers can inform:
    - Diagnosis
    - Directed therapy or matched therapy (such as an FDA-approved immunotherapy)
  - Prognosis
  - Clinical trial eligibility
  - Patient management
- Some cancer biomarkers may not currently have an FDA-approved biomarker-driven therapy, and other treatment options, such as chemotherapy, radiation, or participation in a clinical trial may be more appropriate.

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