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MOLECULAR DIAGNOSTIC

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Course

CLS615

Date

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MOLECULAR DIAGNOSTICS

① You have been given the task of setting up a molecular diagnostics laboratory to provide pharmacogenetic testing. ② What factors would you influence the scale and repertoire of tests you offer, and give two specific examples of genetic tests used to determine drug therapy? Various factors may influence the scale of tests being offered in a molecular diagnostic lab. One of them is the disease being searched for. Clinical presentation and symptom panels may help choose the sample type that is most appropriate for the condition being studied. An assortment of infections may be detected using laboratory-developed tests that have been around for a long time. Sputum, blood, fecal samples, aspirates, urine, swabs, and cerebrospinal fluid are the most often used specimens. A wide range of tests will be moved from the high-complexity lab to the bedside with this plan. Currently, sample loading is being done in a method that is as straightforward as possible. For respiratory and sexually transmitted diseases, swabs have been used to extract samples into an elution, vortex them (if necessary), and then introduce them into an automated system. Gram-negative and gram-positive bacteria and yeast may also be detected using blood culture testing. Fecal samples may be added to a system to test for bacteria, parasites, and viruses. Genetic testing has become very widely recognized, and pharmaceutical companies, patients' advocacy organizations, and patients support genetic screening efforts (Lindy, et al., 2018). The first example is the foundation of fitting blindness that sponsors a genetic testing program for people who have inherited retinal diseases. Patients with these diseases undergo genetic testing and are given genetic counseling. The major purpose of this program is helping in accelerating the pace of clinical trial research by identifying large groups of patients who can qualify for genetic interventional trials and rug trials. Prescription drug treatment may be improved, and pharmaceutical side effects avoided using pharmacogenetic testing. Individual changes in a protein or enzyme expression may alter how a medication is metabolized in a clinical setting. Changes in the levels of active and inactive metabolites may necessitate the administration of various drugs or doses due to these effects.

② Describe how fluorescence in situ hybridization (FISH) is used in cancer diagnosis and prognosis. Many malignancies are caused by a malfunction in our cells' DNA. Tobacco use and other environmental circumstances, such as parental indifference, may also induce a kid to make the same mistake as their parents. What's more, knowing cancer cells' genetic make-up may lead to improved patient treatment, regardless of the disease's origin. "Often referred to as FISH, fluorescence in situ hybridization is a laboratory-based test that helps flesh up the whole picture of a cancer diagnosis by focusing on chromosomes in the cell" (Huber, et al., 2018). Breast cancer, lymphoma, and certain leukemias are among the most prevalent conditions for which FISH is employed. We use blood or bone marrow aspiration samples to screen for blood malignancies. Biopsy tissue preserved in wax is used to treat solid tumors. Probes are manufactured DNA segments to which fluorescent molecules have been added. They've been designed in the laboratory to target certain chromosomal regions. We combine the patient's cells with the probe. This signifies that a strand of the probe binds to the corresponding strand of DNA in the patient's specimen. While binding, the probe emits light to indicate the presence of an anomaly in the chromosomes it was designed to investigate. For example, if a gene ordinarily exists on chromosome one's long arm, but there are additional copies of that area, we'll detect more glowing signals than we would see in a normal cell. For diagnosis- Some chromosomal mistakes can be traced in many individuals. Some types of leukemia are identified by translocations between the 15th and 17th chromosomes, such as acute promyelocytic leukemia. For prognosis - the genetic make-up of a tumor may influence how rapidly it develops and how easily it spreads. FISH data allow us to forecast the behavior of tumors better. When a myelodysplastic syndrome patient develops a tumor that demonstrates the deletion of chromosome 7, it generally advances swiftly. MDS patients with tumors that reveal chromosome 20 depletion tend to have their illness advance more slowly. ③ Molecular techniques are being used increasingly in the diagnosis. Briefly describe how DNA technology is changing the practice of the disorder's diagnosis. In recombinant DNA technology, certain sequences may be isolated, altered, or manipulated to achieve desired outcomes (Natarajaseenivasan, 2018). Recombinant DNA technique is more often referred to as biotechnology than PCR. An interesting aspect of recombinant DNA technology is how quickly it has grown into a multi-billion-dollar business and altered agriculture and pharmaceuticals.

Recombinant DNA molecules are DNA sequences that originate from molecular replications, integrating genetic material from biological species. Because all creatures have the same chemical structure, just differing nucleotides, the process of joining DNA molecules is conceivable. The molecular structure of cancer may be used to determine the underlying causes of the disease. Cancer is an inherited illness that worsens over time and destroys vital organs and tissues. In biotechnology and basic research, recombinant DNA is used extensively to discover and map impacted genes and define the function of each. In molecular research, recombinant DNA methods may be used in two ways: direct and indirect study detection by linkage analysis (Natarajaseenivasan, 2018). A genetic defect may be diagnosed via an indirect investigation of the responsible structural gene is unknown. Analysis is performed using the polymorphisms analysis method, in which fragment length polymorphisms are restricted. Recombination events are more likely to occur if they are closer together. Direct detection of a mutant gene may also be used, but only if the gene is known and its structure is well described. This method can accurately diagnose diseases with molecular abnormalities that show small discrepancies. Restriction fragment length polymorphisms analysis is utilized in conjunction with this method. ② DNA-based molecular techniques have influenced screening for genetic diseases. ③ Give two unrelated examples of how the use of molecular diagnostic has facilitated screening and how this has made a difference. One of the major ways the molecular diagnostic has facilitated screening is through nucleic acid amplification. Nucleic acid amplification methods for viral disease diagnosis and treatment are widespread nowadays. ④ In the clinical virology laboratory, this technology has decreased the utilization of other techniques, such as viral culture-based approaches and serological tests. In addition, nucleic acid amplification techniques are currently the most widely used diagnostic tests for a wide range of illnesses, including cancer. Patients will benefit from adopting these innovative methods and their automation in the clinical laboratory. Consequently, the primary goals of nucleic acid testing in this profession include providing prompt findings that are beneficial for high-quality patient care at a reasonable cost. ④ There are various benefits to employing amplification methods such as polymerase chain reaction, real-time polymerase reaction, or nucleic acid sequence-based amplification for viral detection, genotyping, or quantification (Platts-Mills, et al., 2018). Another way is through Syndromic Molecular Testing for Infectious Disease Diagnostics, which has greatly helped manage pneumonia. In managing patients with pneumonia, timely delivery of suitable medicines is essential, yet conventional, sluggish, culture-based procedures remain the gold standard for microbiological diagnosis. Only 23-40% of individuals with clinically diagnosed pneumonia have a pathogen detected by culture, and that number is even lower after antibiotics have been administered. Pneumonia fast syndromic molecular diagnostics have been developed in recent years. Real-time information on the organisms present and their anticipated antibiotic susceptibility, as well as genotypic indications of resistance, might revolutionize therapy. Multiple studies have shown that PCR-based systems for identifying bacterial infections in the sputum outperform traditional culture in terms of diagnostic accuracy.

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