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OPINION ARTICLE

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Molecular Diagnostics and Its Various Techniques

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About the Study

A group of methods known as "molecular diagnostics" applies molecular biology to medical testing and looks at biological markers in the genome and proteome as well as how cells express their genes as proteins. In medicine, the technique is used to identify risks, diagnose and monitor diseases, and choose the treatments that will be most effective for specific patients. In agricultural biosecurity, the technique is used to monitor crop and livestock diseases, assess risks, and choose the appropriate quarantine measures.

Techniques

Development from research tools: It is now feasible to apply molecular biology assay instruments in clinics as a result of their industrialization. Medical diagnostics may be brought into the clinic, the office, or the home thanks to miniaturisation into a single handheld device. High requirements of dependability are necessary for the clinical laboratory, whereas accreditation or medical device restrictions may apply to diagnostics. Throughput is increased through automation and sample barcoding, and the likelihood of error or contamination during manual handling and results reporting is decreased. There are now single devices that can complete the entire assay.

Assays: *In vitro* biological assays used in molecular diagnostics include PCR-ELISA and fluorescence in situ hybridization. In a patient sample, the assay finds a molecule that is a sign of disease or danger, frequently in low amounts. Prior to analysis, the sample must be preserved. Reduce manual labour as much as possible. RNA is a delicate molecule that presents some difficulties. It provides a measure of gene expression as a result of the biological process of expressing genes as proteins, but it is susceptible to hydrolysis and destruction by constantly active RNAse enzymes. Samples can be cultured with preservation chemicals or immediately frozen in liquid nitrogen.

Prenatal: Traditional prenatal screening methods for chromosomal abnormalities like Down syndrome rely on examining the karyotype, which is an analysis of the number and arrangement of the chromosomes. Because plasma contains cell-free DNA, molecular diagnostic procedures like microarray comparative genomic hybridization, which analyse a DNA sample instead, may be less invasive.

Treatment: Pharmacogenomics is the study of how certain single nucleotide polymorphisms in a patient's DNA can be used to predict how rapidly they will metabolise specific medications. The anti-clotting medication Clopidogrel, for instance, is metabolised by the enzyme CYP2C19 into its active forms. Doctors can screen for these polymorphisms to determine if a patient will completely benefit from the medication. Some people have polymorphisms in specific locations on the 2C19 gene that make them poor metabolizers of those medications.

Some syndromes that were previously thought to be a single disease are now known to be multiple subtypes with wholly diverse causes and treatments because to advancements in molecular biology. Molecular diagnostics can assist in determining the subtype of a disease with an inherited component, such as Silver-Russell syndrome, or the genetic analysis of a disease with an inherited component, such as infections and malignancies.

Disease risk management: A patient's genome may have a hereditary or accidental mutation that influences the likelihood that they may get a disease in the future. For instance, early discovery of Lynch syndrome,

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a genetic condition that increases the risk of colorectal and other cancers in affected individuals, might result in attentive monitoring, increasing the patient's chances of a successful outcome. Biological indicators that indicate cardiovascular risk can be used in screening to determine the likelihood that a child would be born with a hereditary condition like cystic fibrosis. Patients might not want the stress of knowing their risk, which makes genetic testing ethically challenging. A known danger may increase insurance costs in nations without universal healthcare.