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Opinion Article

Molecular Pathology: Study and Diagnoses of Diseases at Molecular Level

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DESCRIPTION

A subspecialty of pathology called molecular pathology is concerned with studying and diagnosing diseases at the molecular level. It uses techniques such as DNA sequencing, Polymerase Chain Reaction (PCR), microarray, *in situ* hybridization, etc., to detect and analyse genetic and epigenetic alterations in cells and tissues that are associated with various diseases, especially cancer. Molecular pathology can provide more accurate and personalized diagnosis, prognosis, and treatment options for patients, as well as insights into the mechanisms and pathways of disease development and progression.

Molecular pathology has several applications in different fields of medicine, such as: Oncology which can identify specific mutations, amplifications, deletions, translocations, or expression changes in oncogenes and tumour suppressor genes that drive the growth and survival of cancer cells. These molecular markers can help classify tumours into subtypes, predict their response to targeted therapies or immunotherapies, monitor their resistance or recurrence, and guide the development of new drugs. For example, molecular pathology can detect Estimated Glomerular Filtration Rate (EGFR) mutations in lung cancer, V-Raf Murine Sarcoma Viral Oncogene Homolog B1 (BRAF) mutations in melanoma, Human Epidermal Growth Factor Receptor 2 (HER2) amplification in breast cancer, KRAS mutations in colorectal cancer, etc., and select appropriate inhibitors for these targets.

Infectious diseases can detect and identify various pathogens, such as bacteria, viruses, fungi, or parasites that cause infections in humans or animals. Molecular pathology can also determine the antibiotic resistance or susceptibility of the pathogens, their virulence factors, their strain types, their transmission routes, and their evolutionary relationships. For example, molecular pathology can detect Severe Acute Respiratory Syndrome Coronavirus 2 (SARS-CoV-2) infection by PCR or sequencing, identify its variants by genomic analysis, monitor its spread by phylogenetic analysis, and evaluate its vaccine efficacy by serological testing.

Inherited diseases can diagnose various genetic diseases that result from mutations or rearrangements in single genes or chromosomes. Molecular pathology can also perform carrier testing or prenatal testing for couples or families at risk of having a child with a genetic disease. For example, molecular pathology can diagnose cystic fibrosis by detecting mutations in the Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene, haemophilia by detecting mutations in the F8 or F9 genes, Down syndrome by detecting trisomy 21 by karyotyping or Non-Invasive Prenatal Testing (NIPT), etc.

Transplantation in molecular pathology can perform tissue typing or Human Leukocyte Antigens (HLA) typing for donors and recipients of organ or bone marrow transplantation. Molecular pathology can also monitor the graft function and rejection by measuring the levels of Donor-Derived Cell-Free DNA (ddcfDNA) in the recipient's blood. For example, molecular pathology can match the HLA alleles of a kidney donor and recipient by PCR-based methods, detect acute rejection by measuring the increase of ddcfDNA by digital PCR or Next-Generation Sequencing (NGS) methods, etc. It is an interdisciplinary field that integrates knowledge and skills from various disciplines, such as pathology, molecular biology, biochemistry, genetics, bioinformatics, etc. Molecular pathologists are trained to perform and interpret molecular tests in clinical laboratories or research settings. They also collaborate with other clinicians and scientists to provide comprehensive and evidence-based care for patients.

It is a rapidly evolving field that faces many challenges and opportunities. Some of the current challenges include standardizing and validating molecular tests across different platforms and laboratories; ensuring the quality control and quality assurance of molecular tests; interpreting complex and heterogeneous molecular data; integrating molecular data with other clinical data; communicating molecular results to patients and physicians; addressing ethical and legal issues related to molecular testing; educating and training molecular pathologists and other health professionals; etc. Some of the current opportunities include developing new molecular technologies

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and methods; discovering new molecular biomarkers and targets; implementing precision medicine and personalized medicine; advancing translational research and clinical trials; improving public health and global health; etc. Molecular pathology is a dynamic and exciting field that has a significant impact on human health and disease. It offers a unique perspective on understanding the molecular basis of diseases and providing better diagnosis and treatment for patients.

CONCLUSION

Molecular pathology has various applications in different fields of medicine, such as oncology, infectious diseases, inherited diseases, and transplantation. It can provide more accurate and personalized diagnosis, prognosis, and treatment options for patients, as well as insights into the mechanisms and pathways of disease development and progression. Molecular pathology is an interdisciplinary field that integrates knowledge and skills from various disciplines, such as pathology, molecular biology, biochemistry, genetics, bioinformatics, etc. Molecular pathologists are trained to perform and interpret molecular tests in clinical laboratories or research settings. They also collaborate with other clinicians and scientists to provide comprehensive and evidence-based care for patients.