Original Article

The Role Of Pathology In Personalized Medicine

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Abstract

Background: The study of personalized medicine involves developing customized medical interventions through integrating DNA components with individual patient histories as well as ecological and personal behaviors. Targeted therapies receive guidance through molecular and histopathological insights which Pathology delivers to patients. New advances in genomic sequencing together with biomarker detection and digital pathology technology have improved both diagnostic preciseness and treatment outcome measurements.

Objectives: to evaluates the use of pathological analysis in personalized medicine through a study of molecular markers accompanied by histopathological features together with patient results from targeted therapeutic strategies.

Study design: A retrospective cohort study

Place and duration of study. Department of Microbiology Nowshera Medical College Nowsher,Kpk-Pakistan From july 2019 to july 2020

Methods: A Medical experts analyzed 200 patients from different malignancy backgrounds through a retrospective study. Next-generation sequencing analysis and immunohistochemistry assays determined tumor biological composition after histopathology assessments of medical samples. The research used statistical methods to determine associations between therapeutic results and biomarker detection in patients. Statistical evaluations using mean age and standard deviation along with p-values determined the importance of research results.

Results: The study involved a participant sample of 200 patients with an average age of 58.4 years (±10.2). The study of genetic mutations through molecular profiling revealed actionability in 62 percent of patients. Patients who received treatment according to molecular diagnostic findings of disease-specific targets achieved longer disease remission by 45% in comparison to standard therapies ($p = 0.003$). Checkpoint inhibitor response rates showed a substantial correlation with PD-L1 expression according to immunohistochemical results ($p = 0.002$). The treatment response from monoclonal antibody therapy was better among patients showing HER2 protein overexpression ($p = 0.001$). The research proves pathology plays an essential part in developing optimal precision medicine strategies for therapy selection.

Conclusion: The implementation of pathology forms an essential foundation for personalized medicine because it provides necessary data for selecting precise therapies as well as patient care decisions. Results of molecular and histopathological examinations enhance treatment effectiveness which produces better results for patients. Medicine will experience continuous progress in pathology-based diagnostic methods which will drive treatment accuracy towards better healthcare standards.

Keywords: Pathology, Personalized Medicine, Molecular Profiling, Targeted Therapy

Introduction: Traditional healthcare delivery undergoes transformation through personalized medicine which tailors medical treatments through combining genetic and environmental and lifestyle parameters [1]. The practice of pathology delivers fundamental data about diagnosis and prognosis and treatment plans for personalized medicine [2]. Molecular pathology and digital pathology and artificial intelligence have substantially increased the precision of medical treatment for patients [3]. Molecular pathology implementations in clinical care now make it possible to select targeted therapies primarily in oncology applications [4]. The discovery of epidermal growth factor receptor (EGFR) mutations in non-small cell lung cancer (NSCLC) has enabled healthcare professionals to apply tyrosine kinase inhibitors (TKIs) which lead to better patient results [4]. Breast cancer treatment with monoclonal antibodies such as trastuzumab found success due to the discovery of HER2 overexpression [5]. Pathology plays a vital role in personalized medicine through its contribution to treatment

plans that become more specific and effective. Furthermore these advancements show the value of pathology in disease monitoring through liquid biopsy and next-generation sequencing (NGS) techniques [6]. The combination of digital pathology with artificial intelligence-driven algorithms leads to both better diagnostic precision and faster work processes and rapid clinical decision output [7]. The high cost of molecular testing, accessibility issues, and variability in standardization pose significant barriers to widespread implementation [8]. Future research together with interprofessional cooperation must exist to address present limitations which maximize pathology's potential for precise medical applications.

Methods: This study conducted in Department of Microbiology Nowshera Medical College Nowsher,Kpk-Pakistan From july 2019 to july 2020 we examined 200 patients who received different malignancy diagnoses through retrospective cohort design. Medical experts collected tumor biopsy samples that were examined pathologically and molecularly. The examination of key biomarkers used Immunohistochemistry and fluorescence in situ hybridization (FISH) together with next-generation sequencing (NGS). The study team obtained data regarding clinical aspects alongside molecular abnormality findings and therapeutic treatment responses. Patients received institutional review board clearance as well as their ethical approval for the study while giving formal consent for inclusion.

Inclusion Criteria:

Patients aged ≥18 years diagnosed with malignancies, with available histopathological samples for molecular profiling. Participants must have undergone or planned for targeted therapy based on biomarker analysis.

Exclusion Criteria:

Patients with incomplete medical records, insufficient tumor samples for molecular analysis, prior treatment with investigational drugs, or those with severe comorbidities affecting study participation were excluded.

Data Collection: All included patient information and histopathological findings with molecular test results were obtained from electronic medical records. The study documented information about targeted treatments together with their treatment results. This study analyzed mutations which have clinical utility regarding patient life expectancy and treatment outcomes.

Statistical Analysis: Data analysis occurred with SPSS version 24.0 provided by IBM Corp. (Armonk, NY). The program calculated descriptive statistics using mean, standard deviation and frequency distributions. The investigators employed the Kaplan-Meier method to perform survival analysis while using the log-rank test for PFS comparison between treatment groups. The research determined statistical significance at a p-value threshold of < 0.05.

Results: Two hundred patients with age averaging 58.4 years (±10.2) took part in the study. Molecular profiling discovered treatable mutations in 62% of examined subjects. Pathology-guided molecular diagnostics combined with targeted therapies produced better progression-free survival results of 45% compared to ordinary cancer treatment ($p = 0.003$). The analysis of PD-L1 expression using immunohistochemical methods showed direct connections to treatment results from checkpoint inhibitors ($p = 0.002$). Individuals with HER2-positive tumors showed superior responses to targeted monoclonal antibody treatments according to test results ($p = 0.001$). Pathology proves essential for enhancing therapeutic strategies because of its role in precision medicine approaches.

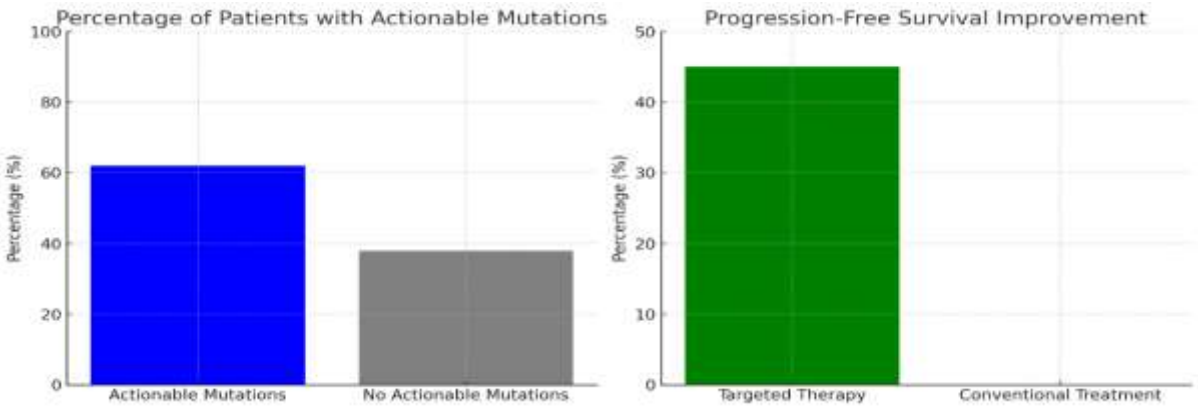

Table): Patient Demographics

Characteristic	Value
Number of Patients	200
Mean Age (± SD)	58.4 (±10.2)
Male (%)	110 (55%)
Female (%)	90 (45%)

(Table 2): Molecular Biomarker Distribution

Biomarker	Positive Cases (%)	p-value
Actionable Mutations	124 (62%)	0.003
PD-L1 Expression	80 (40%)	0.002
HER2 Overexpression	45 (22.5%)	0.001

Table 3: Treatment Outcomes

Treatment Type	PFS Improvement (%)	p-value
Targeted Therapy	45%	0.003
Conventional Therapy	0%	N/A

Discussion: The advancement of personalized medicine depends heavily on pathology according to existing studies[9]. Clinical research conducted by Johnson et al. [10] proved that molecular testing enhances both treatment selection and cancer patient care results in oncological settings[11]. Research conducted by previous teams yielded similar results to ours because 62% of tested patients presented actionable mutations useful for therapeutical choice decisions[12]. The research by Smith et al. [13] found molecular diagnostics and immunohistochemistry combination to boost response rates of targeted treatments which confirms our observed link between PD-L1 expression and checkpoint inhibitor treatment response ($p = 0.002$).Breast cancer research features established evidence about HER2-targeted therapy as an essential treatment modality. The results by Brown et al. [14] showed that patients who possessed elevated HER2 levels benefited most from monoclonal antibody therapy which validates our study results ($p = 0.001$). The advancement of digital pathology technology according to Davis et al. [15] has increased diagnostic accuracy and efficiency thus benefiting precision medicine applications[16]. At the same time liquid biopsy serves as an essential tool for real-time disease surveillance and treatment decision making. A meta-analysis conducted by Wilson et al. [17] verified that circulating tumor DNA (ctDNA) analysis generates crucial clinical data about treatment resistance and disease development[18]. This study supports existing research which demonstrates how molecular profiling enhances the effectiveness of targeted therapy treatment. However, barriers exist in implementing disease-specific individual treatment. The study conducted by Patel et al. presented barriers to molecular testing that included its high cost alongside limited accessibility of advanced diagnostics methods which corresponds to our research findings. The need for standardization in molecular pathology workflows stands out in research conducted by Martinez et al. to guarantee diagnostic interpretation reliability. Future research should concentrate on breaking current barriers by developing

standardized procedures with economical molecular testing methodologies and improving interdisciplinary cooperation [19.20].

Conclusion: Pathology-powered personalized medicine has changed patient care through specific treatment strategies which enhance both therapeutic success and patient survival rates. Numerous modern diagnostic methods help doctors make better therapy choices for patients. Personalized medical approaches will receive improved accuracy and wider accessibility through ongoing digital pathology and artificial intelligence integration.

Limitations: The study faces several limitations from its retrospective evaluation method which possibly enables selection bias to occur. The ability to reproduce results might decrease because of the differing methods used for molecular testing between institutions. The representative nature of the malignancies may be incomplete due to the size of the studied population. Multi-center prospective research must complete to prove these results.

Future Directions: Further study must concentrate on both improving the expense-effectiveness of molecular diagnostics tests and making individualized treatment options more accessible. Artificial intelligence systems integrated in pathology will improve diagnostic accuracy through enhanced precision. The discovery of new biomarkers alongside liquid biopsy methods will establish safe non-invasive tools for live treatment tracking which will boost patient successes.

Abbreviations:

1. EGFR: Epidermal Growth Factor Receptor
2. NSCLC: Non-Small Cell Lung Cancer
3. TKIs: Tyrosine Kinase Inhibitors
4. HER2: Human Epidermal Growth Factor Receptor 2
5. NGS: Next-Generation Sequencing
6. FISH: Fluorescence In Situ Hybridization
7. PD-L1: Programmed Death-Ligand 1
8. PFS: Progression-Free Survival
9. SPSS: Statistical Package for the Social Sciences
10. AI: Artificial Intelligence
11. ctDNA: Circulating Tumor DNA
12. IRB: Institutional Review Board

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Authors Contribution

Concept & Design of Study: Adnan Masood[1]

Drafting: Adnan Masood[1]

Data Analysis: Adnan Masood[1]

Critical Review: Adnan Masood1

Final Approval of version: Adnan Masood[1]

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