

# Mutation Profiling: A Key Tool in Precision Medicine

Miguel A. Torres-Luna\*

Department of Translational Cancer Research, Universidad del Pacífico Médico, Chile

**Received:** 02-Sep-2025, Manuscript No: ipacr-25-15835, **Editor Assigned:** 04-Sep-2025, Pre QC No: ipacr-25-15835 (PQ), **Reviewed:** 17-Sep-2025, QC No: ipacr-25-15835, **Revised:** 20-Sep-2025, Manuscript No: ipacr-25-15835 (R), **Published:** 30-Sep-2025

**Corresponding author:**  
Miguel A. Torres-Luna

✉ luna746@gmail.com

Department of Translational Cancer Research, Universidad del Pacífico Médico, Chile

**Citation:** Torres-Luna MA (2025) Mutation Profiling: A Key Tool in Precision Medicine. Archives Can Res, Vol. 13 No. 5: 80.

## Introduction

Mutation profiling is the systematic analysis of genetic alterations in DNA to identify mutations associated with diseases, particularly cancer. By examining specific genes or whole genomes, mutation profiling provides critical insights into the molecular mechanisms driving disease progression, prognosis, and therapeutic response. This approach has become a cornerstone of precision medicine, allowing clinicians to tailor treatments based on the genetic characteristics of a patient's disease. Advances in next-generation sequencing and bioinformatics have significantly enhanced the speed, accuracy, and accessibility of mutation profiling.

## Discussion

Mutation profiling enables the identification of various genetic changes, including point mutations, insertions, deletions, copy number variations, and chromosomal rearrangements. These mutations can influence disease behavior, treatment response, and resistance mechanisms. In oncology, for instance, profiling tumor DNA can reveal driver mutations that promote cancer cell growth and identify actionable targets for therapy. Examples include EGFR mutations in lung cancer, BRAF mutations in melanoma, and KRAS mutations in colorectal cancer. Such information allows clinicians to select targeted therapies or enroll patients in clinical trials for precision treatments.

Beyond cancer, mutation profiling is valuable in diagnosing hereditary disorders, understanding genetic risk factors, and guiding preventive strategies. In rare genetic diseases, identifying causative mutations aids in accurate diagnosis, family counseling, and treatment planning. In infectious diseases, mutation profiling can track viral evolution, detect drug-resistant strains, and inform public health strategies.

Techniques for mutation profiling range from targeted gene panels, which focus on a select set of clinically relevant genes, to whole-exome or whole-genome sequencing, which provide

a broader view of genetic alterations. Emerging approaches, such as liquid biopsy, allow non-invasive mutation profiling using circulating tumor DNA, enabling real-time monitoring of disease progression and treatment response.

Despite its promise, mutation profiling faces challenges. The interpretation of variants of unknown significance, tumor heterogeneity, and sequencing errors can complicate analysis. Data management and the need for specialized computational tools further add complexity. Ethical considerations, including patient consent and genetic privacy, are also critical when handling genomic information.

## Conclusion

Mutation profiling is a powerful tool that underpins modern precision medicine by revealing the genetic basis of disease. Its applications in cancer, genetic disorders, and infectious diseases allow personalized diagnosis, treatment, and monitoring, improving patient outcomes. While challenges related to interpretation, technical complexity, and ethics remain, advances in sequencing technology and bioinformatics continue to enhance its clinical utility. Mutation profiling is poised to remain a central component of personalized healthcare, guiding targeted therapies and shaping the future of medicine.