

## CHARACTERIZATION OF THE GENOMIC PROFILE IN NON-SMALL CELL LUNG CANCER

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**Abstract:** The present study aims to discuss the importance of characterizing the genomic profile in Non-Small Cell Lung Cancer (NSCLC), since lung cancer is one of the most common types of tumor and one of the main causes of mortality in Brazil. Early diagnosis through genetic mapping has proven to be an ally in the therapeutic proposal. Initially, we found 15 articles associated with the descriptors: Lung Neoplasia; Biomarkers; Genomics. Established criteria were selected, and 8 of these articles were chosen for study and analysis. The publications were detected in the following databases: SCIELO, BVS and PUBMED.

**Keywords:** Lung Neoplasia; Biomarkers; Genomics; Thematic Area; Free Themes.

## **INTRODUCTION**

Cancer is a pathology that begins as a consequence of multiple alterations in the DNA of a single cell, changing its genomic constitution. Lung cancer is the most common neoplasm in the world, according to estimates made by the National Cancer Institute (INCA). For each year of the 2023/2025 triennium, 32,560 new cases of lung, trachea and bronchial cancer will be diagnosed in Brazil, being the third most common in men (18,020 new cases) and the fourth in women (14,540 new cases). It is known that smoking is an important risk factor for the development of lung cancer and approximately 85% of diagnosed cases are associated with the consumption of tobacco derivatives, being responsible for approximately 22% of all deaths related to neoplasia. With the advancement, the realization of a complete genomic profile can lead to early detection and consequently the choice of the best therapeutic proposal, where it will be possible to identify molecular subtypes of the neoplasia. The molecular alterations found in Non-Small Cell Lung Cancer (NSCLC) through genomic profiling

are expression, amplification, inversion/fusion and mutations detected in EGFR, KRAS, BRAF, TP53, HER2, RH, MET and ALK where these altered pathways interfere with the normal function of cells and play a fundamental role in the carcinogenesis and progression of lung cancer, therefore, molecular diagnosis can determine the choice of the best therapy, through a personalized monitoring plan.

## **METHODOLOGY**

This is an integrative review; whose main objective is to investigate the importance of characterizing the genomic profile in lung cancer. The inclusion criteria were articles with full text and available in Spanish, English and Portuguese, published between 2013 and 2023. The databases used were: Scientific Electronic Library Online (SCIELO), Virtual Health Library (VHL) and National Library of Medicine (PUBMED). The exclusion criteria for the research were duplicate articles that did not address the characterization of the genomic profile of neoplasia and lung cancer and those that deviated from the line of study, thus leaving 7 articles to compose this research.

## **THEORETICAL BASIS OR RESULTS AND DISCUSSIONS**

### **RESULTS**

Based on the precepts established for this review, 7 articles were selected, their main results are characterized below:

### **DISCUSSIONS**

According to research, the delay in diagnosing lung cancer is one of the main challenges faced in Brazil, as there is insufficient information on the time required for an individual with suspicious symptoms to receive a diagnosis and begin treatment.

In view of this, genomic testing has proven to be essential to improve therapeutic outcomes; however, accessibility and incorporation of molecular tests has been challenging, especially in low- and middle-income countries. It has been observed that lack of information on the part of physicians and patients' lack of knowledge, failure to access specific therapy, and logistics can negatively contribute to the care and treatment of these individuals, in addition to approximately two-thirds of tests being performed in the private sector, and only one-third in public institutions. The prevalence of pathogenic alterations in the EGFR gene, whose function is associated with cell growth and survival, in Brazil varies (25-30%), but data on alterations in the Brazilian population are still scarce.

According to a Brazilian retrospective study, alterations in the EGFR gene presented a prevalence of 32.7%; translocations in the ALK gene 4% of patients; and variants of the KRAS, BRAF, HER2, ROS1 genes were present in 20%, 11.8%, 14.3% and 9% of patients respectively. It is also worth highlighting the importance of genetic counseling, which is defined as an interactive process of help and assistance to the individual or family who presents or is at risk of developing, or even transmitting, a genetic condition.

## **FINAL CONSIDERATIONS**

Given the results, we conclude that characterizing the genomic profile and understanding the molecular alterations involved in the pathogenesis of Non-Small Cell Lung Cancer (NSCLC) is essential for the management of patients with this type of cancer. It is clear that strategies for accessing and incorporating molecular tests need to be expanded, since reducing the time to diagnosis is extremely important in the fight against lung cancer, which is usually diagnosed late.

Title	Author:/year	Main goals	Type of study
Lung Cancer Genomics	Parikh, 2019	Understanding the phenomenon of lung cancer treatment evolution using genomic testing to identify specific and resistant mutations.	Narrative literature review
Epidemiological study of biomarkers in non-small cell lung cancer	Melo, 2011	Detailed study on the epidemiology of lung cancer, staging and treatment of non-small cell lung cancer.	Retrospective cohort study.
Nurses' performance in oncology from the perspective of genetics and genomics.	Flória et al, 2013	The work aims to reflect on the role of nurses in oncology, on genetics and genomics, their role in the multidisciplinary team and ontological genetic management, based on a precise reading of the literature in the area at stake.	Review article, the result of a thorough reading of the area
Epidemiological and molecular profile in patients with lung cancer and adenocarcinoma in Ceará	Rocha, 2015	Epidemiology of lung cancer, providing data on the estimated number of tumors in Brazil and by region, lung cancer worldwide and cell signaling pathways.	Observational, prospectus and analytical review.
Lung cancer	Ministry of health, 2022.	Proposal for educational material on how to prevent, signs and symptoms and treatment of lung cancer.	Educational material
Lung cancer in Brazil	Araujo et al, 2018	Review not only of the current situation of lung cancer through research of relevant data on prevention, diagnosis and treatment in the country, but also of issues regarding smoking control and late diagnosis.	Literature review article
Cancer Progress and Priorities: Lung Cancer	Matthew B. Schabath	Epidemiological study on the process and priorities of lung cancer.	Descriptive epidemiology

Source: own elaboration

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