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“Characterizing Genetic Mutations in Familial Lung Cancer: Insights & Implications”

Lung cancer (LC) is the leading cause of cancer-related deaths worldwide, often linked to smoking tobacco. However, less than 20% of smokers develop LC. It has been shown in literature that there is both an environmental component (smoking) and a genetic component that play a significant role in the development of LC, as indicated by disease aggregation and higher mortality rates in individuals with affected relatives with LC. Identifying genetic mutations is crucial for developing novel therapies to treat previously untreatable diseases. Researchers have identified significant genetic mutations that may be linked to susceptibility to LC in specific populations, yet more research is needed to confirm these findings. The goal of this study is to characterize the genetic mutations in individuals with LC who have a family history of the disease.

The research study of genetics of lung cancer recruited approximately 2,500 individuals with LC from a network of 30 Louisiana hospitals and from multiple states across the country. After screening, 800 participants were confirmed to have at least two cases of LC in their family. Out of those 800 study participants, only 21 people had previous mutation analyses by the certified laboratories, 18 of which received somatic mutation screening and three of which received germline mutation screening. Medical and pathology reports containing mutation analyses of the 21 study participants were obtained from hospitals in addition to demographic and environmental information from the families.

The analyses of the results found that approximately 24% of study participants from these familial LC families tested positive for the EGFR mutation and 0% tested positive for the ALK mutation. The four who tested positive for the EGFR mutation had a higher number of family members affected by LC compared to those who tested negative.

Limited information can be concluded due to the lack of a standardized list of mutations that LC patients are tested for. The mutation screening among individuals with LC helps with determining to administer the most appropriate therapies. This research confirms the findings of previous research, highlighting the need for continued research on the genetics of LC, and the need for a standardized mutation screening panel for all individuals with LC.