Characterizing Genetic Mutations in Familial Lung Cancer: Insights & Implications
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Introduction

Background:
- Lung cancer (LC) is the leading cause of cancer-related deaths worldwide.
- First-degree relatives of LC patients have a 1.51 times higher risk of developing LC.
- Less than 20% of smokers develop LC, suggesting that genetics play a key role.
- Difficult to pinpoint a list of specific genetic mutations that cause LC.
- Caused by a combination of both genetic and environmental risk factors.
- Multiple genes are responsible.
- Identifying genetic mutations aids in determining the best treatment options.
- Precision Medicine (PM): specific drugs can target specific genetic mutations.
- PM can help LC patients avoid generalized harmful treatments like chemotherapy.

Gaps:
- Identified genetic mutations linked to LC susceptibility need further validation.
- More research is required to confirm findings across different populations.

Objective:
- Characterize genetic mutations in familial LC cases.

Methods

Interpretations of Results
- Prevalence of the EGFR and ALK gene mutations amongst study participants varied by about 5% each from what they were reported to be in literature.
- Those with EGFR mutation:
  - Higher number of relatives with LC.
  - Earlier age of onset.
  - Different types of mutations were observed: exon 19 deletion, exon 20 insertion, and p.T790M mutation.
- No participants tested positive for the ALK mutation despite being one of the most common genes tested for.

Limitations
- No standard screening panels for mutational analysis.
- Only included Caucasians.
- Only 21 participants had mutational analysis.

Future Directions
- How PM can be applied in the presence of multiple genetic mutations.
- Whether somatic or germline mutations of a gene more commonly occur.
- How the presence of certain gene mutations affects LC prognosis.
- Average age of LC onset in patients with a germline mutation.
- Common genetic mutations in never smokers.
- How patients can possess certain germline mutations but never develop LC.

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