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# Associations between telomere attrition, genetic variants in telomere maintenance genes, and non-small cell lung cancer risk in the Jammu and Kashmir population of North India

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## Abstract

**Background** Telomeres are repetitive DNA sequences located at the ends of chromosomes, playing a vital role in maintaining chromosomal integrity and stability. Dysregulation of telomeres has been implicated in the development of various cancers, including non-small cell lung cancer (NSCLC), which is the most common type of lung cancer. Genetic variations within telomere maintenance genes may influence the risk of developing NSCLC. The present study aimed to evaluate the genetic associations of select variants within telomere maintenance genes in a population from Jammu and Kashmir, North India, and to investigate the relationship between telomere length and NSCLC risk.

**Methods** We employed the cost-effective and high-throughput MassARRAY MALDI-TOF platform to assess the genetic associations of select variants within telomere maintenance genes in a population from Jammu and Kashmir, North India. Additionally, we used TaqMan genotyping to validate our results. Furthermore, we investigated telomere length variation and its relation to NSCLC risk in the same population using dual-labeled fluorescence-based qPCR.

**Results** Our findings revealed significant associations of TERT rs10069690 and POT1 rs10228682 with NSCLC risk (adjusted  $p$ -values = 0.019 and 0.002, respectively), while TERF2 rs251796 and rs2975843 showed no significant associations. The TaqMan genotyping validation further substantiated the associations of TERT rs10069690 and rs2242652 with NSCLC risk (adjusted  $p$ -values = 0.02 and 0.003, respectively). Our results also demonstrated significantly shorter telomere lengths in NSCLC patients compared to controls ( $p$  = 0.0004).

**Conclusion** This study highlights the crucial interplay between genetic variation in telomere maintenance genes, telomere attrition, and NSCLC risk in the Jammu and Kashmir population of North India. Our findings suggest that TERT

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