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Genetic alterations are frequent and hetrogenously distributed in HPV-related carcinoma of different anatomical sites

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Next generation sequencing analysis allowed to identify previously unrecognized hot spot mutations in coding and regulatory regions of human genome. Hot spot mutations in the promoter region of the telomerase reverse transcriptase gene (TERT) have been identified in several tumor types and have been found associated with specific pathological features and poor prognosis. The rs2853669 SNP in some tumors has shown to modulate the expression induced by TERT promoter mutations. To determine the frequency and the role of these mutations and rs2853669 SNP in TERT promoter region among HPV-related cancers at different body sites, we have analyzed 244 tumors including penile cancer, cervical carcinoma and head and neck cancer. Activating mutations in TERT promoter were detected in 30.4% of penile cancers and 26.1% of cervical cancer. Mutations were more frequent in HPV-negative (66.7%) than HPV-positive penile cancers (27.3%) and predominant in the verrucous squamous cell carcinoma (100%) histotype. On the other hand, all TERT mutated cases of cervical cancer were positive for high risk HPVs. We found no association between TERT promoter mutations and the rs2853669 polymorphism in HPV-related genital tumors. No mutations were detected among the 53 HPV-negative and 7 HPV-positive head and neck cancers analyzed in this study suggesting that in these tumors there was an alternative activation mechanism of telomerase expression, likely related to TP53 mutations and consequent abrogation of p53-dependent TERT repression. In conclusion, mutations in TERT promoter represent a frequent event in specific subgroups of HPV-related genital cancers. These findings raise the possibility that TERT expression in HPV-positive cancers may be activated either by the expression of E6 and E7 HPV oncogenes or by activating mutations in TERT promoter region. TERT mutations have an impact on prognosis and response to therapies in different HPV-related cancer histotypes needs to be analyzed in investigative clinical trials.

Biography

Maria Lina Tornesello has completed her graduation in 1988 at the Faculty of Science of University "Federico II"-Napoli, Italy and obtained Board Certification in Microbiology and Virology in 1994 at the “Federico II” Medical School-Napoli. She is the Head of Viral and Tumor Marker Lab, at the Istituto Nazionale Tumori Fond Pascale. She has published more than 130 articles in peer reviewed journals and has been serving as an Editorial Board Member of *PLoS One, Infect Agent* Cancer, Clinical and Vaccine Immunology and Canadian Journal of Infectious Diseases and Medical Microbiology.

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