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22976 | Molecular Profiling of Prognostic Biomarkers in Melanoma

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Background & Aim: Cutaneous melanoma (CM) is the most aggressive and deadliest form of skin cancer (1). Its development is influenced by the genetic background, genetic mutations and environmental factors. The three most frequent mutations of CM appear in *BRAF* gene, which leads to uncontrolled cell growth, in *NRAS* gene, that disrupts normal cell signaling, and in *TERT* promoter, that increases telomerase activity and play a crucial role in tumor progression, influencing melanoma aggressiveness and patient outcome (2, 3). Despite advances in treatment, the prognosis of patients with advanced-stage remains poor (4). This work aims to improve prognosis determination by correlating identified molecular alterations with clinicopathological data. **Methods:** The analysis is being conducted on primary tumors collected at Hospital dos Capuchos. The study currently includes 29 samples collected between 2023 and 2024, 18 from male subjects and 11 from female subjects, with an average age of 69. Most of the melanomas were located on the trunk, and the most common diagnoses were nodular melanoma and superficial spreading melanoma. The mean Breslow thickness of the series is 3.78 mm, and 46% of samples have a mitotic index greater than 1. The samples were subjected to histopathological evaluation, followed by PCR and sequencing techniques, to identify the three mutations. Subsequent statistical analysis will be performed to assess the correlation between these genetic changes and clinicopathological features, with a focus on determining their impact on patient outcomes. **Results:** The work is still ongoing, but the results will provide further information about the three mutations and their corresponding aggressiveness, as well as how the coexistence of different mutations influences patient prognosis. **Conclusions:** This study underlines the significance of molecular profiling in the prognosis assessment of melanoma patients, and it will provide a foundation for further research on molecular-based prognosis models.

Keywords: Melanoma, mutations, *BRAF* gene, *NRAS* gene, *TERT* promoter.

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References:

[1] Timis T, Bergthorsson JT, Greiff V, Cenariu M, Cenariu D. Pathology and Molecular Biology of Melanoma. Curr Issues Mol Biol. 2023;45(7):5575-97.

[2] Helena Pópulo PSJML. Insights into melanoma: targeting the mTOR pathway for therapeutics. 2012.

[3] Martins F, Martins C, Fernandes MS, Andrade MI, Lopes JM, Soares P, et al. TERT Promoter Mutational Status in the Management of Cutaneous Melanoma: Comparison with Sentinel Lymph Node Biopsy. Dermatology. 2022;238(3):507-16.

[4] Domingues B, Lopes JM, Soares P, Populo H. Melanoma treatment in review. Immunotargets Ther. 2018;7:35-49.