

## BRAF GENE STATUS IN FINE-NEEDLE ASPIRATES OF METASTATIC MELANOMA

Maria Gabriela Gasparinho, Daniela Proença, Ricardo Fonseca, Saudade André  
Department of Anatomic Pathology, Portuguese Institute of Oncology Francisco Gentil,  
Lisbon, Portugal

**BACKGROUND:** BRAF mutations in metastatic melanoma are now being recognized as a putative target in novel therapeutic regimens, namely BRAF inhibitors. About 40-60% of cases harbour mutations involving the BRAF gene (V600E -70-90% of cases; V600K -6-30% of cases). Detection of BRAF mutations becomes of the utmost importance for the selection of patients.

**OBJECTIVES:** To evaluate cytology as a means of sampling material for the determination of BRAF mutation status.

**MATERIAL-METHODS:** Fine-needle aspirates from melanoma cutaneous metastases were prospectively obtained. Cells were collected from smears and/or from cell suspensions (PBS). DNA was extracted and amplified. Hybridization of DNA to a test-strip containing a V600E mutation probe (ViennaLab Diagnostics GmbH ®) was performed. Results were confirmed by Sanger sequencing.

**RESULTS:** Ten samples were collected. Extractions yielded good quality DNA. The DNA amount obtained from scratched smears (two cases) was 7,37 and 8,60µg/ml; when obtained from PBS (eight cases), it ranged between 18,50 and 177,30µg/ml (mean: 61,95µg/ml). Only one case harboured a V600E mutation by both test-strip and sequencing. The remaining nine cases were wild-type by test-strip, but sequencing highlighted two heterozygous cases for V600K mutation.

**CONCLUSIONS:** Fine needle cytology is a reliable way of obtaining good quality samples for testing BRAF mutations in melanoma. Collection to saline yielded a more significant DNA amount, when compared to smears. Commercially available tests, such as the one conducted in these samples, effectively detect the V600E mutation, seeming suitable as selection tests, overcoming the need for the time-consuming genomic sequencing.

**Keywords:** Braf mutations testing, fine-needle aspiration cytology