MUTATIONAL PROFILE AND PROTEIN EXPRESSION OF PTCH1, SMO, SUFU AND TP53 GENES IN RECURRENT LESIONS OF BASAL CELL CARCINOMA

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INTRODUCTION

Basal cell carcinoma (BCC) is the most common malignant neoplasm in humans[1]. Tumors often develop in chronically sun exposed skin. BCC is a slow-growing, locally invasive tumor with a high morbidity rate due to invasion and local destruction of the skin and adjacent structures[2]. It particularly affects the face, head and neck, causing deformities, with greater local damage in cases where there is tumor recurrence[3]. BCC is primarily driven by the Sonic Hedgehog (Hh) pathway. Approximately 50% of BCCs show a *TP53* mutation. Although the most common driver pathways in BCC is known, tumors display great variability in aggressiveness, morphology and response to treatment[4]. Local recurrences, despite apparent complete excision, and occurrence of synchronic and metachronous tumors, bring the question, also discussed in other neoplasms, of the presence of molecular alterations existing in a surgical margin being responsible for the return of the tumor[5].

OBJECTIVES

This study sought to determine whether the gene expression profile and protein expression in the margin of the tumors differs between recurrent and non-recurrent BCCs lesions.

METHODS

An observational nested case-control study of 587 cases with confirmed diagnosis of BCC, from 2009 to 2012 will be assessed using information from Brazilian Cancer Registry and Pathology Department of Instituto Nacional do Câncer (INCA). The frequency of mutations in Hh pathway genes (*PTCH1, SMO*, and *SUFU*) and gene *TP53*, and the expression of PTCH1, p53 proteins by immunohistochemistry (IHC) staining in the tumor and margins will be analized in 184 cases (recurrents) and 184 controls. This study will be conducted according to the Resolution 466/2012.

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