Population Screening of K-ras Gene and Genetic Counselling for Patients Affected with Ampulla of Vater in Tamil Nadu

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ABSTRACT Carcinoma of the ampulla of Vater is a relatively infrequent neoplasm, approximately six percent of periampullary tumours. The aim of the study is to identify the chromosomal alterations and the K-ras mutations in the familial and sporadic carcinomas of the ampulla of Vater. A totally of 21 samples were selected which included 18 familial and 3 sporadic cases which were categorized based on their age group (group I < 50 years; group II>50 years). Techniques such as the GTG-banding and PCR-RFLP were used to identify the genetic alterations. The result revealed a high frequency of chromosomes 1p- and 12p+ involved in the poorly differentiated (PD) tumor grade and an increased prevalence of the K-ras mutations at the codon 12 associated with > 2cm tumor size in the familial carcinomas of the ampulla of Vater. The researchers concluded that the chromosomes 1p- and 12p+ region may play a vital role for the development of a high grade tumor and the K-ras gene mutation is an early molecular event leading to an abnormal proliferation of the cells.