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Ampullary Carcinoma – A Genetic Perspective

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Abstract

Ampulla of vater carcinoma (AVC) is a rare gastrointestinal tumour that is associated with a high mortality rate and it's often diagnosed at later stages due to lack of clinical symptoms. Early diagnosis of this condition is essential to effectively treat patients for better prognosis. A significant amount of advancement has been made in understanding the molecular nature of cancer in the past decade. A substantial number of mutations and alterations have been detected in various tumors. Despite the occurrence of AVC across the globe, the number of studies conducted on this tumor type remains low; this is largely due to its rare occurrence. Moreover, AVC tissues are complex and contain mutations in oncogenes, tumour suppressors, apoptotic proteins, cell proliferation proteins, cell signaling proteins, transcription factors, chromosomal abnormalities and cellular adhesion proteins. The frequently mutated genes included KRAS, TP53 and SMAD4 and are associated with prognosis. Several molecules of the PI3K, Wnt signaling, TGF-beta pathway and cell cycle have also