C-kit protein expression correlated with activating mutations in KIT gene in oral mucosal melanoma

Type:
Article

Abstract:
C-kit is a trans-membrane receptor tyrosine kinase (RTK) encoded by the proto-oncogene KIT located at 4q11-12. Gain-of-function mutations arising to c-kit activation independent of its ligand were observed in various tumors related to germ cells, mast cells, and interstitial cells of Cajal. C-kit also participates in melanocyte development; hence, its involvement in oral mucosal melanoma (OMM) tumorigenesis was investigated. Immunohistochemistry and mutation analysis were performed using 18 cases of human primary OMM. Results revealed 16 cases positive to c-kit protein. Atypical melanocytes expressed c-kit. All in situ components expressed c-kit, but only four cases exhibited intense expression in the invasive component. Missense mutations were observed in four cases, and two of those correlated with increased protein expression. C-kit expression in atypical melanocytes suggests the role of c-kit in the early stage of OMM tumorigenesis. C-kit protein expression correlated with activating mutations indicating the pertinent role of the proto-oncogene KIT in the tumorigenesis of OMM.

Keyword:
oral mucosal melanoma, C-kit mutation, immunohistochemistry, gastrointestinal stromal tumors, endothelial growth-factor, amplicon, melting analysis, malignant-melanoma
imatinib mesylate, metastatic, melanoma, melanocytic lesions, clinical-efficacy, cd-117 expression, kinase domain

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