Sinonasal, genital and acrolentiginous melanomas show distinct characteristics of KIT expression and mutations.

Abstract:
KIT aberrations predict the outcome of targeted therapies in acrolentiginous (ALM) and mucosal (MM) melanoma patients. KIT immunoreactivity and mutation status was assessed in 41 ALM and 25 MM patients. Of these, 19 ALM and 15 MM patients had matched primary and metastatic lesions. P-ERK was investigated in a subset of 9 ALM and 7 MM matched primary/metastatic pairs by immunohistochemistry. Heterogeneous KIT immunoreactivity was observed in both primary and metastatic lesions. Mutations were present in four of 41 ALM (10%) and five of 25 MM (20%) patients. Only vulvar mucosal samples carried KIT mutations in contrast to sinonasal lesions (p = 0.0109). In KIT-mutated tumours, the mutations were present in KIT expressing as well as KIT negative cells, as shown by Laser Capture Microdissection (LCM). P-ERK expression was preferentially found in metastases. KIT mutations predict treatment outcome with KIT inhibitors. Therefore, especially vulvar melanoma patients should be screened for activating KIT mutations.