

Abstract

Low Incidence of Germline and Somatic Alterations in the E-Cadherin Gene in Lobular Breast Carcinoma

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Germline mutations of E-cadherin have been identified in families with gastric cancer. Moreover, E-cadherin has been shown to be inactivated in the majority of human invasive lobular breast cancers. E-cadherin is a tumor suppressor gene located in 16q22.1, a region which is frequently deleted in a variety of tumors. We therefore analysed 15 families with lobular breast cancer or breast and gastric cancer for germline mutations and 20 sporadic lobular

breast carcinomas for somatic mutations in the E-cadherin gene. Screening of the lobular breast cancer families revealed two germline mutations of the missense type (A592T, P692L). One of these mutations (A592T) has previously been identified in a sporadic papillary thyroid tumor. No germline mutations were found in breast and gastric cancer associated families. Only 2 mutations could be identified in the tumor panel. Both were of the frameshift type and associated with LOH causing inactivation of E-cadherin. We here report for the first time germline mutations in the E-cadherin gene in families with lobular breast carcinomas. In addition, the frequency of E-cadherin mutations in our panel of lobular breast carcinomas is lower than published in former studies.

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