ETV6 gene rearrangement in a mammary analogue secretory carcinoma of the skin

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Mammary analogue secretory carcinoma is a relatively recently recognized entity that harbors the recurrent ETV6 gene rearrangement. To date, only a few cases of mammary analogue secretory carcinoma of the skin have been reported; however, to the best of our knowledge, the presence of ETV6 gene rearrangement has not been observed in the aforementioned cutaneous cases. A 57 year-old man presented with a 6.0 cm cystic mass in the axilla involving the dermis and superficial subcutis. Microscopically, the tumor was composed of nodular aggregates of tubular and microcystic structures embedded in a dense fibrotic and hyalinized stroma. Characteristic "colloid-like" eosinophilic secretions were present within the intraluminal spaces. Many tumor cells contained vesicular nuclei with conspicuous central nucleoli, surrounded by pink vacuolated cytoplasm. By immunohistochemistry, the tumor cells were intensely positive for AE1/AE3, Cam 5.2, and CK7, while Ber-EP4 and CEA were completely negative. The dual color break-apart fluorescence in situ hybridization probe identified a rearrangement of the ETV6 gene locus on chromosome 12. The patient is alive, with no evidence of recurrent disease or metastasis three years after surgery. In conclusion, we report the first case of mammary analogue secretory carcinoma of the skin with ETV6 gene rearrangement. Awareness of this rare cutaneous tumor and thus reporting of additional cases is necessary for further delineating its full clinicopathologic spectrum.