Case Report

T1 Breast Cancer Associated with Von Recklinghausen’s Neurofibromatosis


A rare case of breast cancer associated with von Recklinghausen’s neurofibromatosis is reported. This case and review of the literature illustrate the problems of clinical diagnosis.

A 66-year-old woman who had undergone sigmoidectomy for sigmoid colon cancer two years previously, was admitted to the hospital because of a left breast skin retraction in October, 1998. The patient had von Recklinghausen’s disease (neurofibromatosis type 1). The TNM clinical staging was T1cN0M0. Modified radical mastectomy was performed. The histopathological diagnosis of the breast tumor was invasive ductal carcinoma and the skin tumor was neurofibroma. The pTNM pathological staging was pT1cN1aM0.

Among patients similar to our case, almost all were staged higher than T2. This may be because multiple neurofibromas obscure breast mass at palpation, leading to delayed detection of the cancer. Systemic and careful exploration is essential for patients with von Recklinghausen’s neurofibromatosis to detect breast cancer at an early stage.


Key words: Breast cancer, Von Recklinghausen’s disease, Neurofibromatosis

Von Recklinghausen’s disease has been often seen in association with some nonepithelial malignant tumors such as neurofibrosarcoma, however, an association with breast cancer has been rarely reported. In this paper a rare case of T1 breast cancer in a patient with von Recklinghausen neurofibromatosis is reported and the patient literature is reviewed.

Case Report

A 66-year-old woman was admitted with a left breast skin retraction in October, 1998. She had undergone sigmoidectomy for sigmoid colon cancer two years previously at Nerima General Hospital, with a histopathological diagnosis of well differentiated adenocarcinoma. There was a history of having numerous tumors and café-au-lait spots over the whole body since the age of 46. There was no family history of breast cancer or neurofibromatosis. On physical examination, skin retraction was observed at the upper outer quadrant of her left breast (Fig 1), and tumor was palpated beneath it. No lymphadenopathy was appreciated. Mammography revealed an irregularly shaped, spiculated mass, 1.7×1.5 cm in size, without microcalcifications (Fig 2). Ultrasonography revealed a 1.5×1.2 cm hypoechoic lesion (Fig 3). Fine needle aspiration cytology yielded a diagnosis of breast cancer. Left modified radical mastectomy was performed.

Fig 1. Local findings. Skin retraction [arrow] and neurofibromas are observed.
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performed on November 11, 1998. Macroscopically, the tumor was located on the left lateral side of the left breast, measured $2.0 \times 1.5 \times 1.5$ cm, and showed no invasion of the skin or nipple. Histopathologically, the breast tumor was invasive ductal carcinoma (Fig 4) with lymph node involvement (one of 13 nodes were positive). The skin tumor adjacent to the retraction was neurofibroma (Fig 5). Postoperative staging was pT1cN1a (1/13) M0, stage IIA, according to the TNM classification. The tumor was negative for estrogen receptor and positive for progesterone receptor. The postoperative course was uneventful, and she is currently healthy without recurrence.

Discussion

Von Recklinghausen's disease was first described in 1882. Formerly, it was considered a single disease, but it is now known to be two distinct diseases, neurofibromatosis type 1 (NF-1) and neurofibromatosis type 2 (NF-2). NF-1 is readily diagnosed by the presence of six or more café-au-lait spots and multiple neurofibromas. NF-2 has an onset characterized on the development of tinnitus or hearing loss due to bilateral acoustic