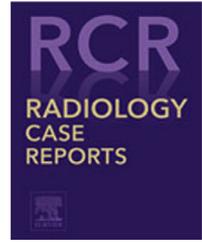


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Case Report

Dynamic contrast-enhanced MRI and sequential CT findings of metaplastic breast carcinoma in neurofibromatosis type 1: A case report [☆]

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ABSTRACT

Metaplastic breast carcinoma in neurofibromatosis type 1 is extremely rare. There are few reports about dynamic contrast-enhanced MRI findings and sequential CT findings of metaplastic breast carcinoma in neurofibromatosis type 1. Herein, we report imaging findings, including dynamic contrast-enhanced MRI and sequential CT, of metaplastic breast carcinoma in an 82-year-old woman with neurofibromatosis type 1. Short tau inversion recovery image revealed an oval mass with a circumscribed margin that exhibited moderate intensity with partially hyperintense area inside, and T1-weighted imaging revealed a spotty hyperintense area. The solid component of the mass showed heterogeneous enhancement and the time-intensity curve had a fast/washout pattern with restricted diffusion. In addition, multiple neurofibromas were observed. Sequential CT revealed that the diameter of the mass doubled in 3 months without apparent lymph node metastasis. Because detection of metaplastic breast carcinoma in neurofibromatosis type 1 tends to be delayed due to multiple neurofibromas, characteristic MRI findings suggestive of metaplastic breast carcinoma and sequential CT findings are important for early treatment of metaplastic breast carcinoma in patients with neurofibromatosis type 1.

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Introduction

Metaplastic breast carcinoma (MBC) is a rare subtype of breast cancer, accounting for 0.1%-0.5% of all breast cancers [1]. MBC rapidly grows without lymph node metastasis [2,3] and has worse prognosis than other subtypes of breast cancer [4]. Although breast cancer in patients with neurofibromatosis type 1 (NF-1) is a relatively common manifestation, breast cancers in NF-1 are usually invasive ductal carcinoma pathologically [5] and only eight cases of MBC in NF-1 have been reported to date [6]. Here, we report imaging findings, including dynamic contrast-enhanced magnetic resonance imaging (MRI) and sequential computed tomography (CT), of MBC in NF-1.

Case report

An 82-year-old woman clinically diagnosed as NF-1 was admitted to our hospital with a lump in her left breast. On physical examination, an elastic hard mass approximately 3 cm

in diameter was detected in the upper area of her left breast and multiple nodules were observed on her skin. Her medical history was hypertension and right breast cancer. Approximately 15 years ago, her right breast was resected in another hospital (specific pathology was unknown). Her parents and siblings had no history of breast cancer. Her laboratory findings were not significant. On mammography, an oval mass approximately 3 cm in diameter showed high density and zonal pleomorphic calcification in the left middle (M) region of mediolateral oblique (MLO) view and left inner (I) and outer (O) regions of craniocaudal (CC) view (Fig. 1A, arrow). On Doppler ultrasonography, a hypo-echoic mass with irregular margins was detected (Fig. 1B). The mass showed indistinct anterior border and abundant blood flow signals. CT revealed a heterogeneous low-density mass (Fig. 1C), and the diameter of the mass increased from 3 to 6 cm in 3 months (Figs. 1C and D) during preoperative examination. There was no evidence of metastasis. Dynamic contrast-enhanced MRI of the breast was performed preoperatively. Short tau inversion recovery (STIR) imaging showed a well-circumscribed oval mass with a moderate-intense and partially hyperintense area inside (Fig. 2B). On T1-weighted imaging, the mass included

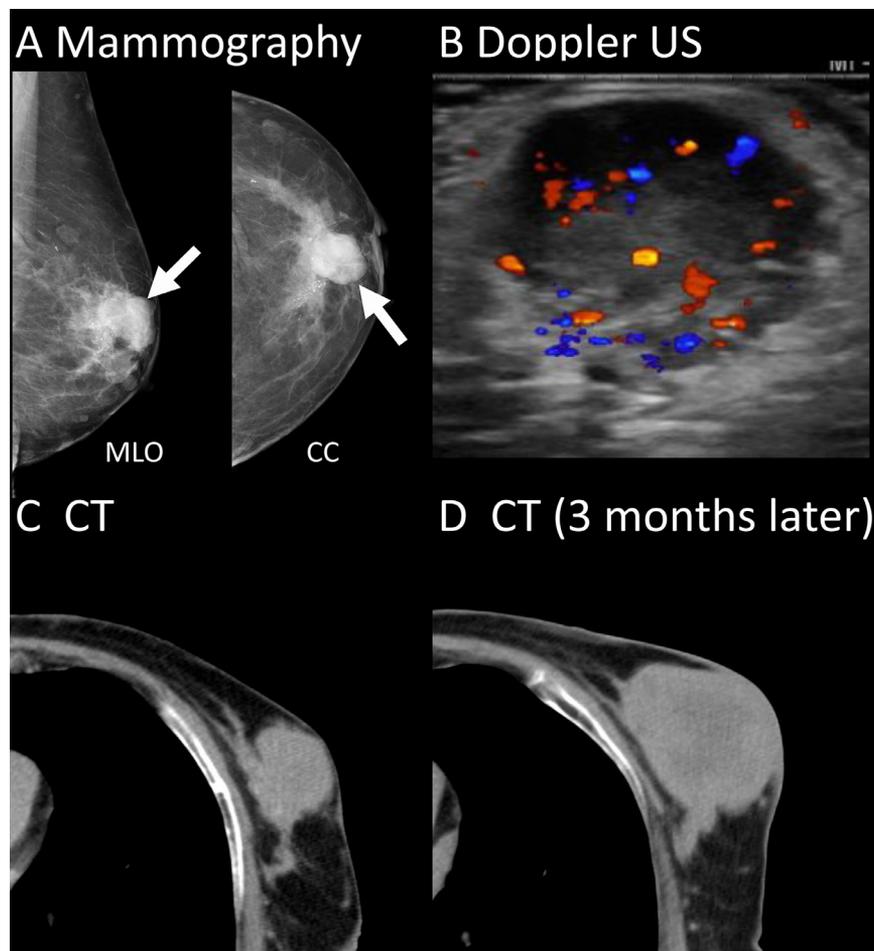


Fig. 1 – Mammography of the left breast showed a high-density oval mass of approximately 3 cm in diameter with zonal pleomorphic calcification (A, arrow). Doppler ultrasonography (US) demonstrated a hypo-echoic mass with irregular margins and detectable flow signal (B). Non-contrast enhanced CT showed a heterogeneous low-density mass, which rapidly increased from 3 cm to 6 cm in diameter in 3 months (C and D).

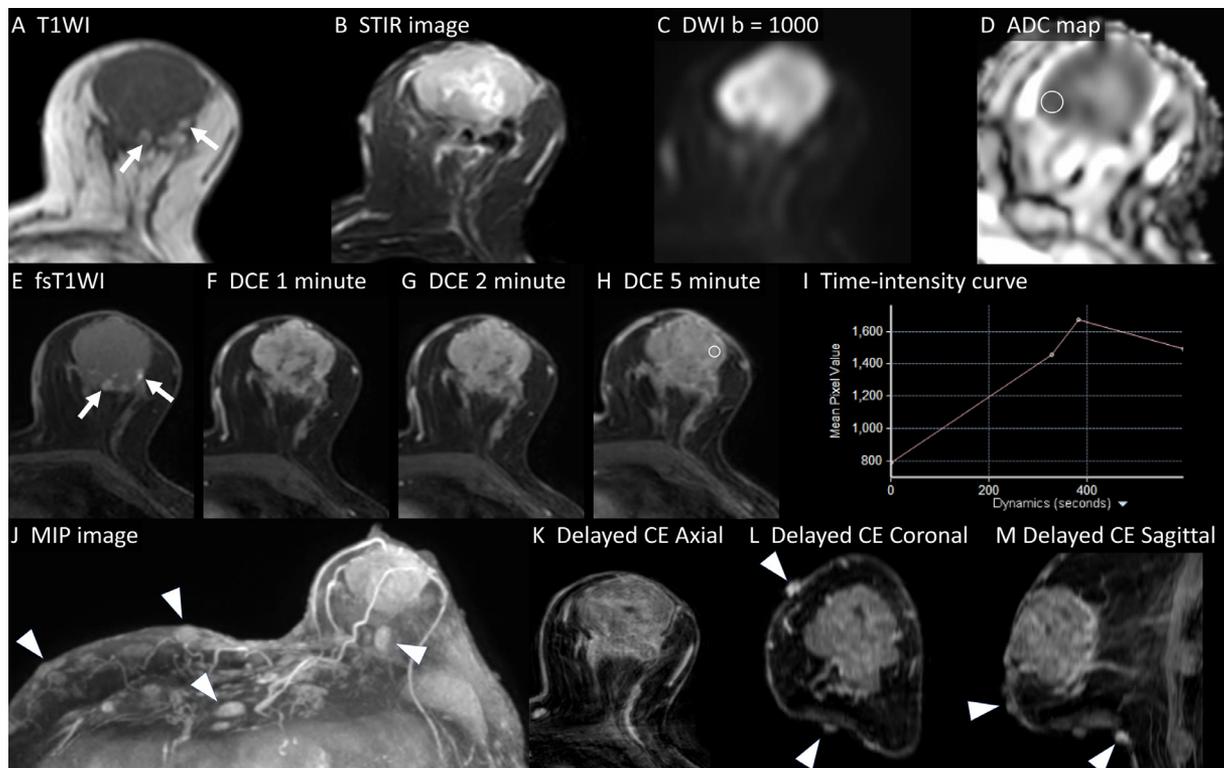


Fig. 2 – An oval mass with circumscribed margin is hypo-intense on T1-weighted imaging with spotty hyper-intense area (A and E, white arrows), which is considered to be hemorrhagic degeneration. The mass has a partially hyper-intense area on short tau inversion recovery (STIR) imaging (B). The solid portion of the mass exhibits restricted diffusion (C, DWI b = 1000; D, apparent diffusion coefficient [ADC] map) and its ADC value was $0.63 \times 10^{-3} \text{ mm}^2/\text{s}$ (D, measured within the circle). On dynamic contrast-enhanced MRI (E, fat-suppressed T1-weighted imaging [fsT1WI] before injection of gadolinium contrast medium; F, 1 minute after injection; G, 2 minutes after injection; and H, 5 minutes after injection), the mass shows heterogeneous enhancement and the time-intensity curve shows fast/washout pattern (I, measured within the circle in H). Maximum intensity projection imaging visualized the overall picture, including the resected right breast (J). Multiplanar delayed contrast-enhanced fsT1WI demonstrates a heterogeneous mass at high resolution (K–M). In addition, characteristic neurofibromas are visualized as nodules on the skin (J, L, and M, white arrowhead).

spotty hyperintense areas (Figs. 2A and E, white arrow). On dynamic contrast-enhanced study, the solid component of the mass showed heterogeneous enhancement (Figs. 2E–H), and the time-intensity curve showed a fast/washout pattern (Fig. I, measured within the circle). The mass had restricted diffusion and its apparent diffusion coefficient value was $0.63 \times 10^{-3} \text{ mm}^2/\text{s}$ (Figs. 2C and D, measured within the circle). In addition, multiple enhanced nodules were observed in the skin (Figs. 2J, L, and M, yellow arrow). Based on the imaging findings, invasive ductal carcinoma or malignant lobular tumor were suspected. Preoperative needle biopsy revealed spindle cells in the tumor, which suggested malignant phyllodes tumor or metaplastic breast carcinoma, and left mastectomy was performed. Pathological examination revealed that the tumor consisted of spindle cell carcinoma (Figs. 3B, triangle; and 3C and D), invasive ductal carcinoma with apocrine differentiation (Figs. 3B, circle; and 3E), and ductal carcinoma in situ. Moreover, resected specimen included some neurofibromas (Fig. 4). The final diagnosis was MBC in NF-1. The resection margin was negative. There was no recurrence 6 months after surgery.

Discussion

MBC is considered a rare subtype of breast cancer, accounting for only 0.1%–0.5% of all breast cancers [1]; however, a review article about breast cancers in NF-1 patients reported that 3 of 29 cases (10.3%) had MBC [5]. Furthermore, an association between mutation in the neurofibromin 1 (NF1) gene and MBC has been recognized [7–9]. These findings suggest that the frequency of MBC in NF-1 may be higher than expected.

Dynamic contrast-enhanced MRI revealed that the well-circumscribed mass showed heterogeneous enhancement and the time-intensity curve showed a fast/washout pattern in the solid component. On diffusion-weighted imaging, the solid component showed restricted diffusion. The solid component may reflect rich spindle cells with small amount of stromal component. STIR imaging revealed a hyperintense area inside the mass. Similar to previous reports, this hyperintense area was correlated with necrotic tissue [2,10]. In addition, the mass included hyperintense spots on T1-weighted imaging, which correlated with hemorrhagic degeneration

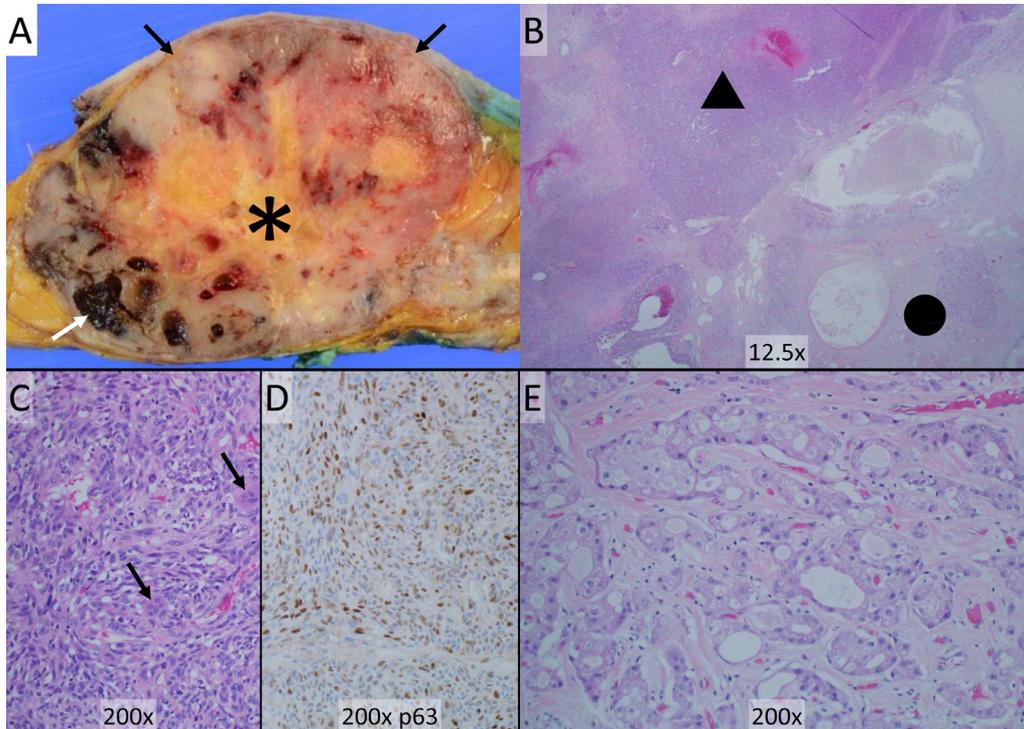


Fig. 3 – Macroscopic examination of the specimen reveals an oval mass of approximately 6 cm in diameter, which included whitish solid component (A, black arrows), hemorrhagic degeneration (A, white arrow), and necrotic tissue in the center of the tumor (A, asterisk). Based on histological examination, the whitish solid component consisted of spindle cell carcinoma (B, triangle), invasive ductal carcinoma with apocrine differentiation (B, circle), and ductal carcinoma in situ (hematoxylin-eosin staining, 12.5x). The spindle cell carcinoma includes characteristic osteoclast giant cells (hematoxylin-eosin staining, 200x, C, arrow). Immunostaining of p63 highlights the spindle cells (200x, D). The invasive ductal carcinoma with apocrine differentiation shows ductal structure (hematoxylin-eosin staining, 200x, E).

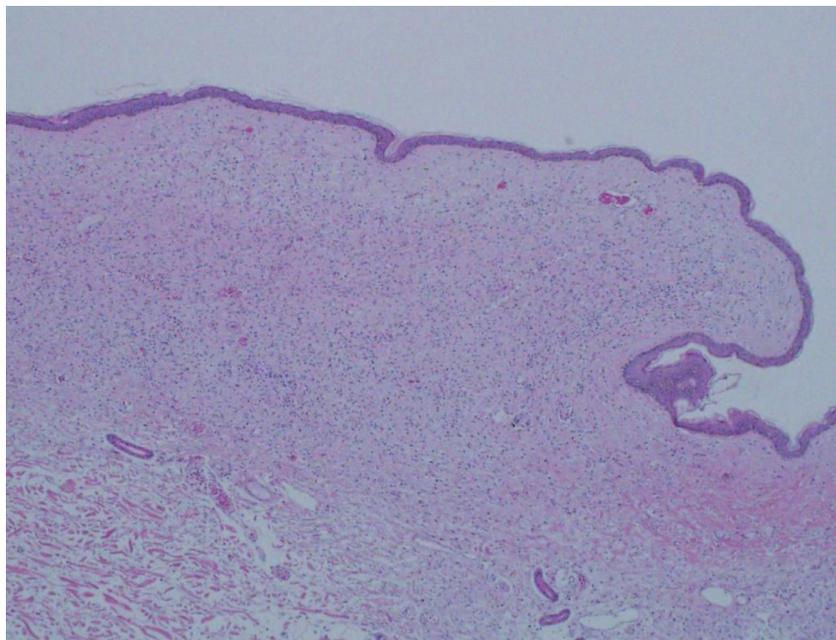


Fig. 4 – Multiple skin nodules in the resected specimen are neurofibromas on pathological examination.

pathologically. A previous study reported hypo-intense rim on T2-weighted imaging [11]; however, this was unclear in the present case.

Rapid tumor growth without lymph node metastasis was observed on sequential CT. Compared with invasive ductal carcinoma, MBC often grows rapidly, but lymph node metastasis is rare [2]. In the present case, the diameter of the mass doubled in 3 months during preoperative examination without apparent lymph node metastasis. Although MBC grows rapidly, detection of MBC in patients with NF-1 may be delayed due to multiple neurofibromas [12]. Therefore, characteristic MRI findings and sequential imaging findings suggestive of MBC are important for early treatment of MBC in patients with NF-1.

In summary, imaging findings of MBC in NF-1, including those on dynamic contrast-enhanced MRI and sequential CT, were found. Although imaging findings suggestive of MBC in NF-1 are limited, taking into account the recently reported association between NF1 mutation and MBC, MBC should be considered for differential diagnosis in patients with NF-1.

Patient consent

Written and informed consent was received from the patient.

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