AMSER Case of the Month September 2021

72-year-old female presents for annual mammogram

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Patient Presentation

- HPI:
  - Patient presents for clinical breast exam and right breast screening – 5 years follow up left IDC s/p mastectomy
  - Patient denies palpable findings, nipple discharge, nipple inversion, breast pain, or erythema of the breast
- PMH:
  - Left DCIS s/p partial mastectomy and adjuvant radiation (2009)
  - Left IDC (ER-/PR-/HER2-) s/p simple mastectomy and adjuvant chemotherapy (2016)
  - *Other relevant medical history temporarily withheld*

What imaging should be ordered according to ACR breast cancer screening guidelines?

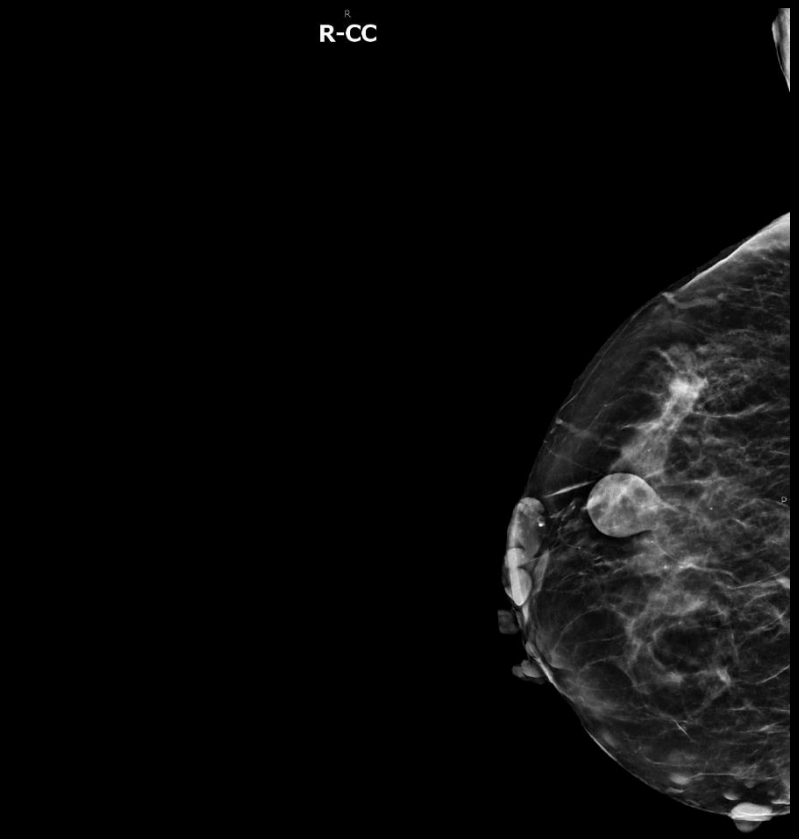
ACR Appropriateness Criteria

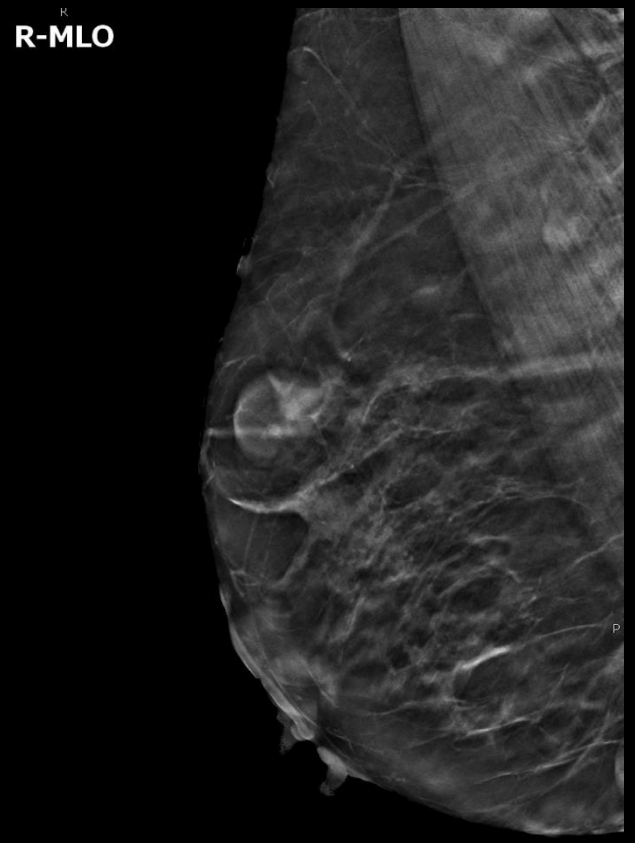
Variant 2: **Breast cancer screening. Intermediate-risk women: women with personal history of breast cancer, lobular neoplasia, atypical ductal hyperplasia, or 15% to 20% lifetime risk of breast cancer.**

Procedure	Appropriateness Category	Relative Radiation Level
Mammography screening	Usually Appropriate	☢☢
Digital breast tomosynthesis screening	Usually Appropriate	☢☢
MRI breast without and with IV contrast	May Be Appropriate	O
US breast	May Be Appropriate	O
FDG-PET breast dedicated	Usually Not Appropriate	☢☢☢☢
Sestamibi MBI	Usually Not Appropriate	☢☢☢
MRI breast without IV contrast	Usually Not Appropriate	O

This imaging modality was ordered by the primary physician

Findings (unlabeled) – Screening Mammogram



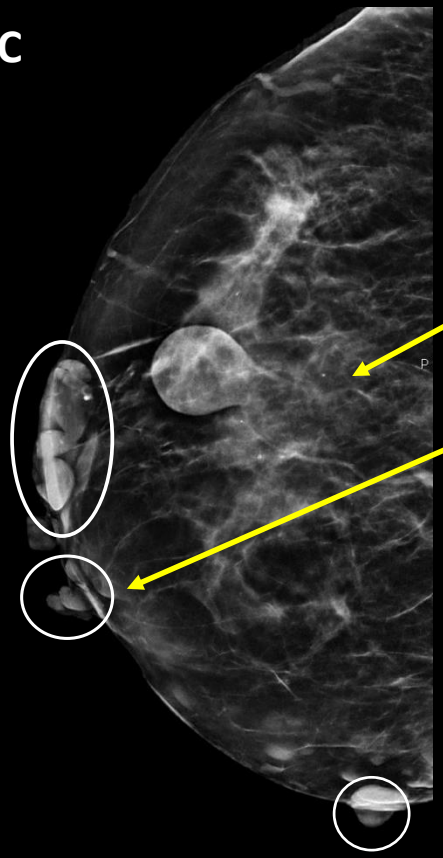


Did you spot any interesting skin findings?

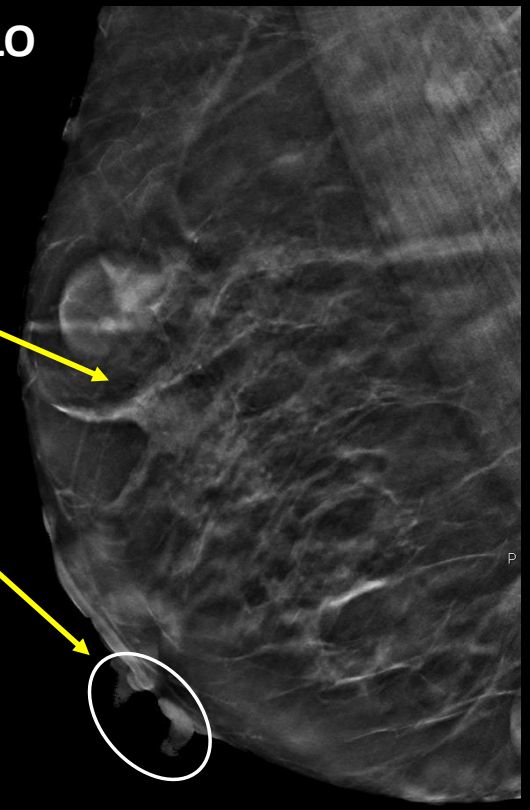
Additional Imaging – Prior CT

Findings (labeled) – Screening Mammogram

R-CC



R-MLO



FINDINGS:

Scattered areas of fibroglandular density

Numerous superficial peri-areolar <u>neurofibromas</u>

**circled in white*

No suspicious masses, areas of architectural distortion or calcifications

Left breast surgically absent

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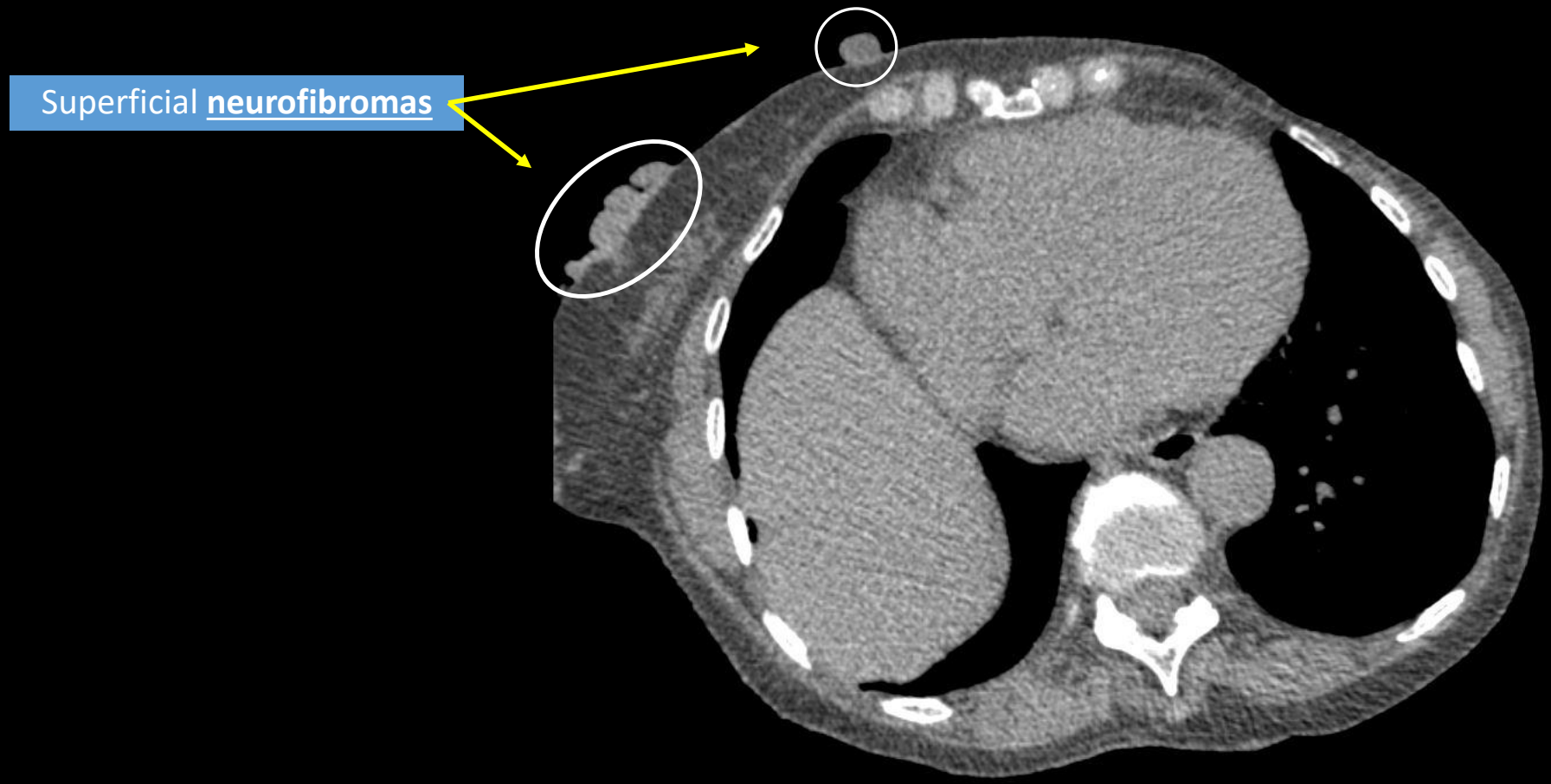
Additional Imaging (labeled) – Prior CT



Final Diagnosis:

Neurofibromatosis 1

Final Impression of Screening Mammogram: BI-RADS 2 – Benign

Case Discussion – Neurofibromatosis 1 (NF1)

- Autosomal dominant multisystem neurocutaneous disorder
- Affects 1:2500-3000 individuals
- Mutation of tumor suppressor gene → loss of function → uninhibited cell growth with neurofibroma development
- Clinical manifestations:
  - Neurofibromas
  - Café au lait spots
  - Lisch nodules
  - Axillary and inguinal freckling
  - Skeletal abnormalities
    - Sphenoid wind dysplasia
    - Leg bowing
  - Optic nerve glioma

Teaching Point - Breast radiographic features of neurofibromas in NF1

Mammography

- Well-defined benign-appearing masses
- Classically peri-areolar
- May mimic and partially obscure breast lesions

Ultrasound

- Well-defined hypoechoic masses with posterior acoustic enhancement
- *Features similar to fibroadenoma*

References:

- Williams VC, Lucas J, Babcock MA et-al. Neurofibromatosis type 1 revisited. Pediatrics. 2009;123 (1): 124-33. doi:10.1542/peds.2007-3204

- Lu-Emerson C, Plotkin SR. The Neurofibromatoses. Part 1: NF1. Rev Neurol Dis. 2009;6 (2): E47-53.

- Hillier JC, Moskovic E. The soft-tissue manifestations of neurofibromatosis type 1. Clin Radiol. 2005;60 (9): 960-7. doi:10.1016/j.crad.2005.02.008

- Gokalp G, Hakyemez B, Kizilkaya E et-al. Myxoid neurofibromas of the breast: mammographical, sonographical and MRI appearances. Br J Radiol. 2007;80 (958): e234-7. doi:10.1259/bjr/33539044