72-year-old female presents for annual mammogram

Savannah Gorisek, MS4
Drexel University College of Medicine

Sarah Thomas, DO
Matthew Hartman, MD
Allegheny Health Network
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?

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Appropriateness Category</th>
<th>Relative Radiation Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mammography screening</td>
<td>Usually Appropriate</td>
<td>☀️ ☀️</td>
</tr>
<tr>
<td>Digital breast tomosynthesis screening</td>
<td>Usually Appropriate</td>
<td>☀️ ☀️</td>
</tr>
<tr>
<td>MRI breast without and with IV contrast</td>
<td>May Be Appropriate</td>
<td>☀️ ☀️</td>
</tr>
<tr>
<td>US breast</td>
<td>May Be Appropriate</td>
<td>☀️ ☀️</td>
</tr>
<tr>
<td>FDG-PET breast dedicated</td>
<td>Usually Not Appropriate</td>
<td>☀️ ☀️ ☀️ ☀️</td>
</tr>
<tr>
<td>Sestamibi MBI</td>
<td>Usually Not Appropriate</td>
<td>☀️ ☀️ ☀️ ☀️</td>
</tr>
<tr>
<td>MRI breast without IV contrast</td>
<td>Usually Not Appropriate</td>
<td>☀️ ☀️ ☀️ ☀️</td>
</tr>
</tbody>
</table>

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

FINDINGS:

- Scattered areas of fibroglandular density
- Numerous superficial peri-areolar neurofibromas *circled in white
- No suspicious masses, areas of architectural distortion or calcifications
- Left breast surgically absent
Additional Imaging (labeled) – Prior CT

Superficial neurofibromas
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:


