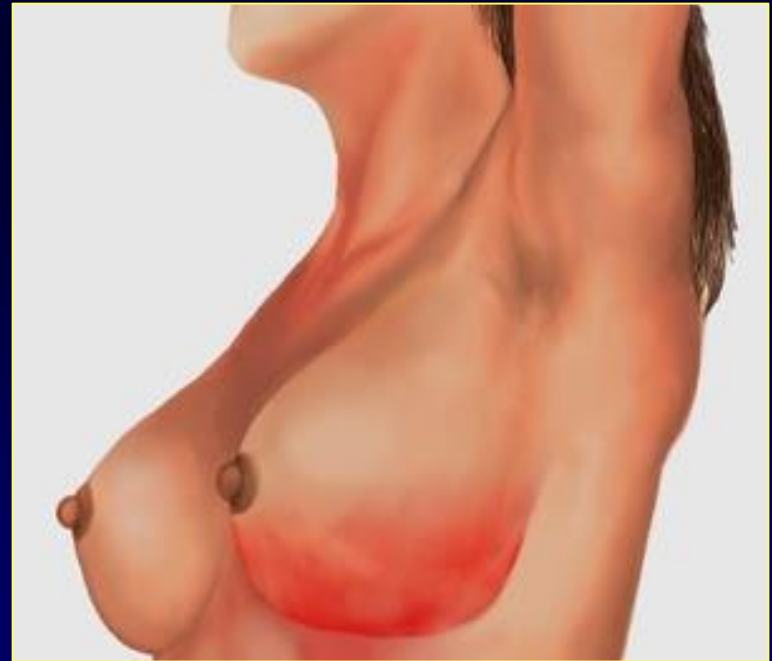


Anamnesi

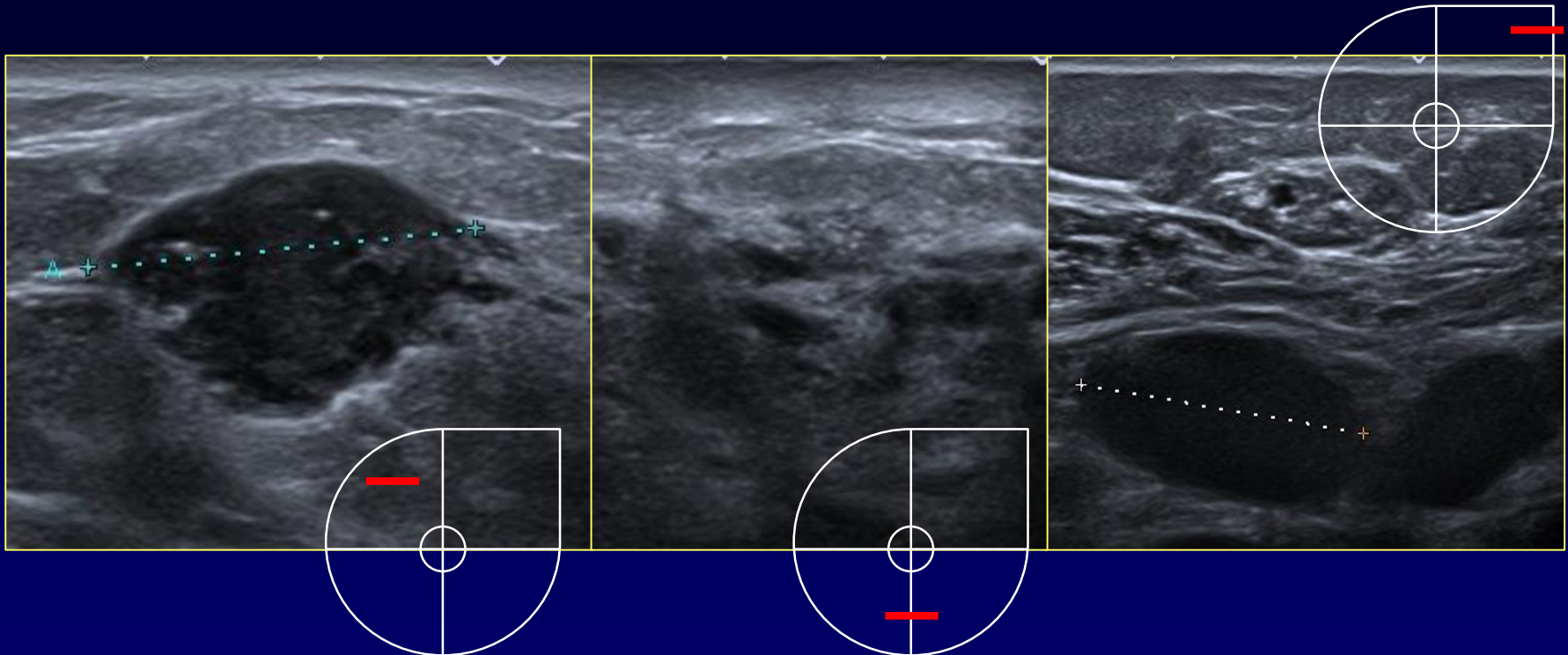
- ✓ 31 anni
- ✓ Non familiarità per neoplasia mammella / ovaio

- ✓ **Mastite sinistra**
- ✓ Non correlata all'allattamento
- ✓ Antibiotico senza beneficio



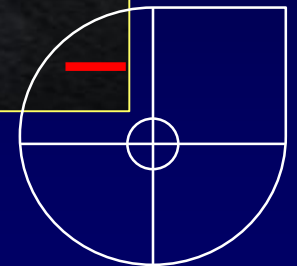
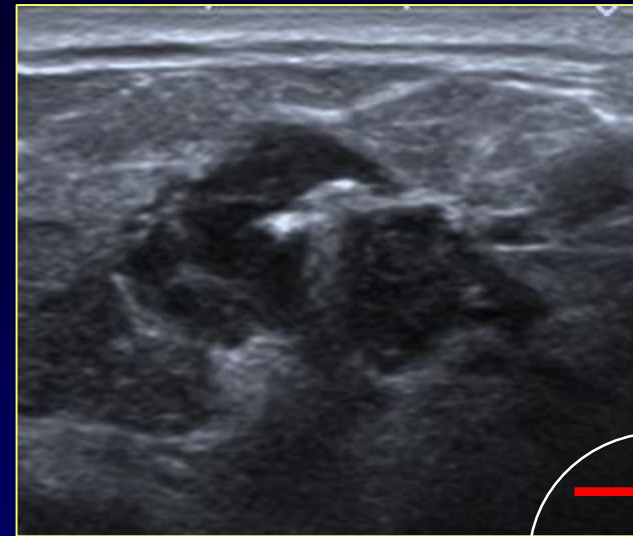
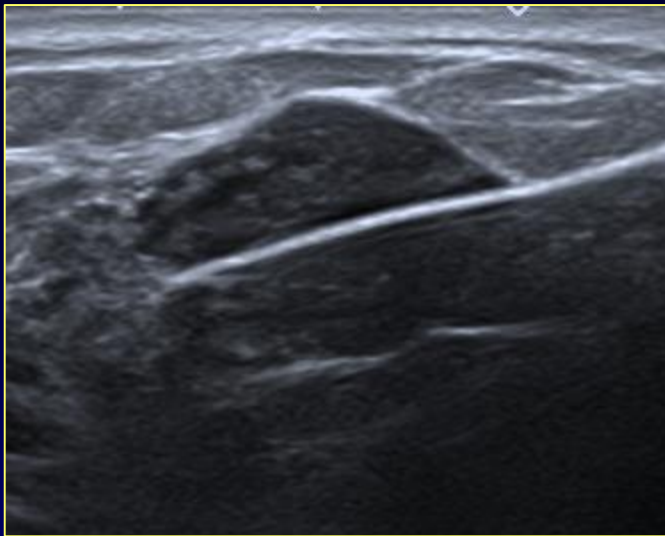
Ecografia mammaria

- ✓ **Formazione nodulare ipoecogena sospetta QSM sinistra**
- ✓ **Ispessimento di cute e sottocute della mammella sinistra**
- ✓ **Tumefazioni linfonodali di aspetto patologico in sede ascellare sinistra**



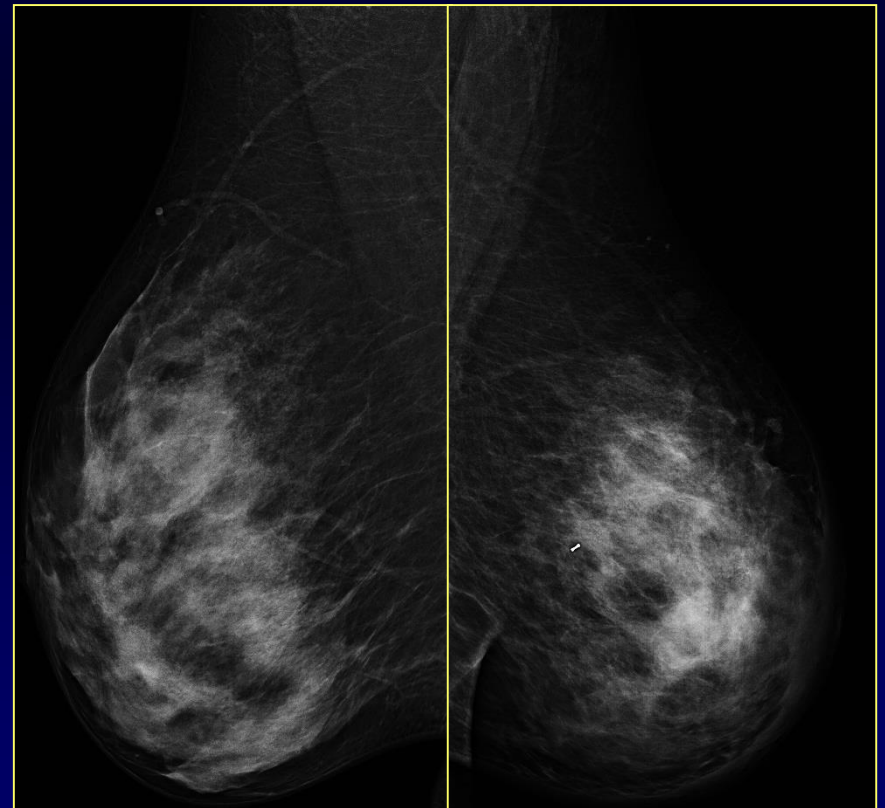
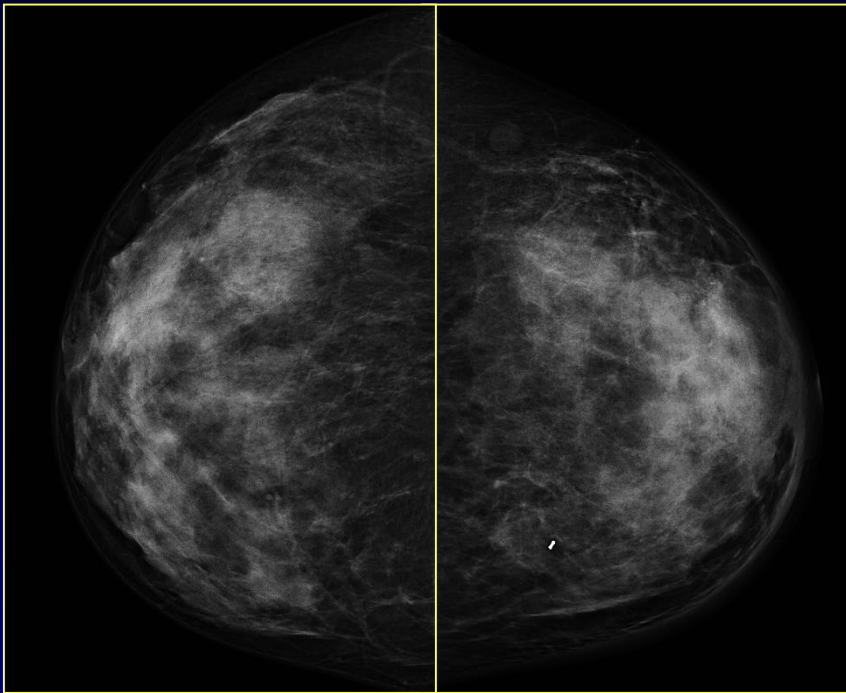
Agobiopsia ecoguidata

- ✓ Esame Istologico QSM sinistra
Carcinoma Duttale Infiltrante G3
ER 10% PgR 1% ki67 46% HER2+ 90% 3+



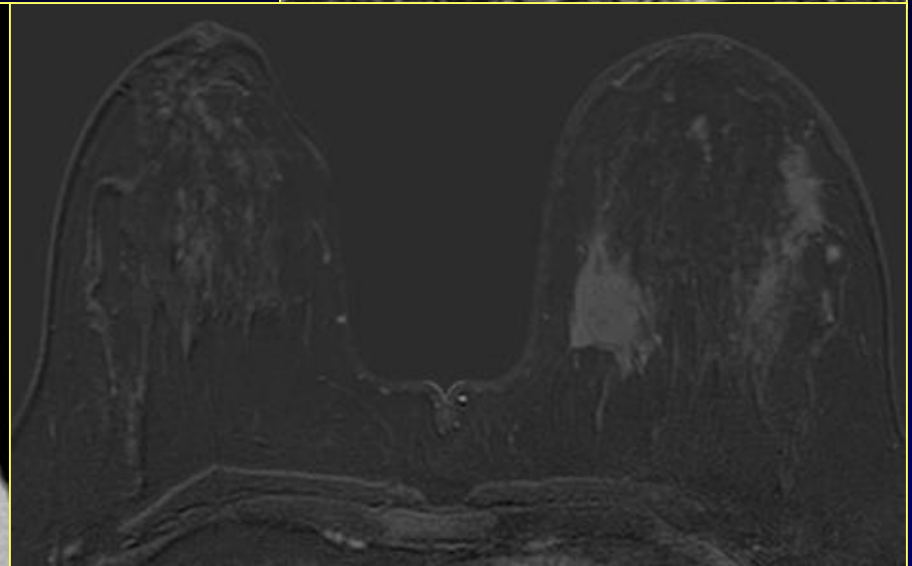
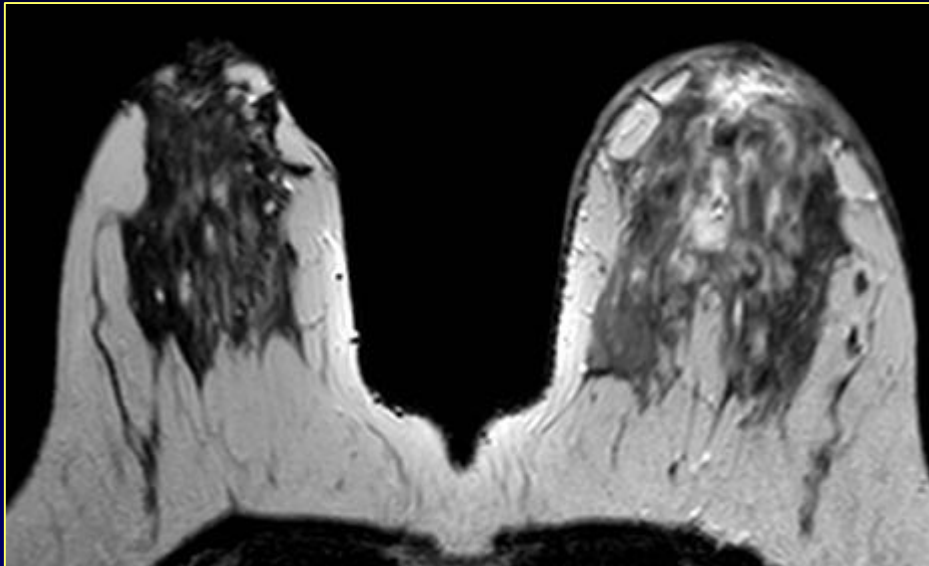
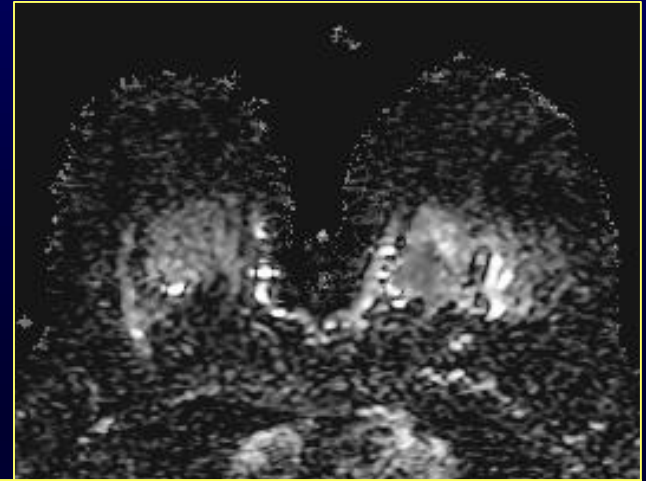
Mammografia

- ✓ Mammelle diffusamente e disomogenamente radiopache
- ✓ Tenue opacità QSM sinistra (contenente clip)
- ✓ Ispessimento cutaneo a sinistra



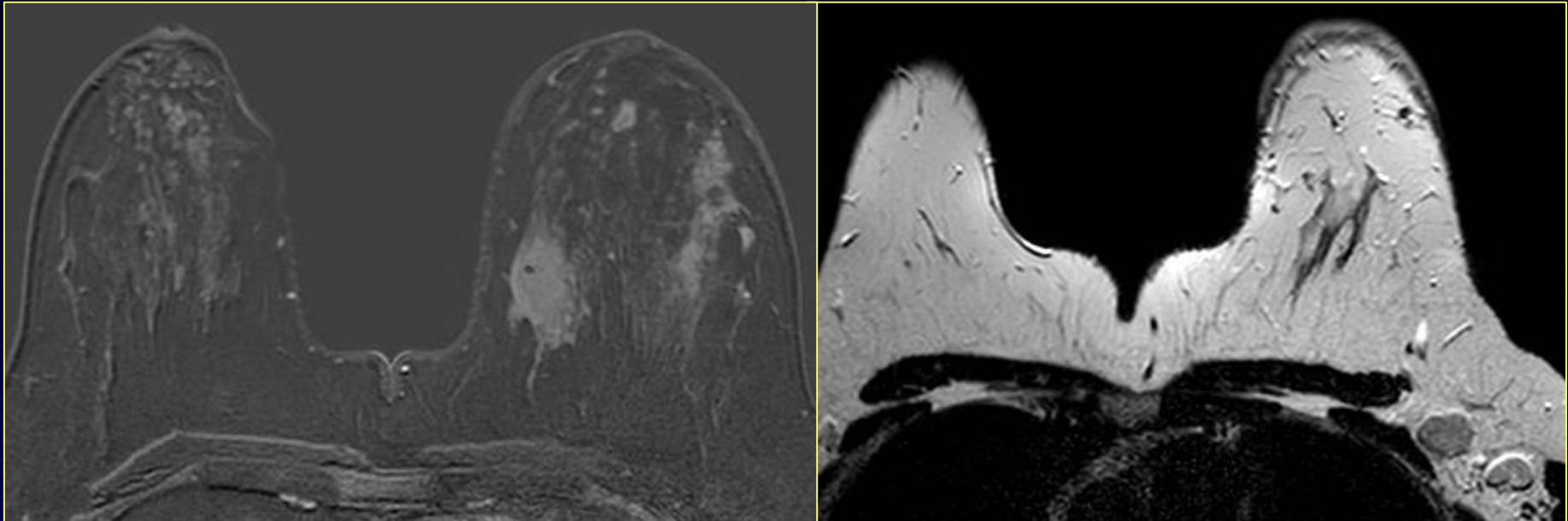
Risonanza Mammaria

- ✓ Enhancement mass like QSM sinistra
- ✓ Ulteriori aree di enhancement QSE sinistra
- ✓ Ispessimento dei piani cutanei ed edema del parenchima a sinistra



Risonanza Mammaria

- ✓ Tumefazioni linfonodali di aspetto patologico in sede ascellare sinistra
- ✓ Estesa area di non mass-like enhancement al passaggio tra i quadranti superiori di destra

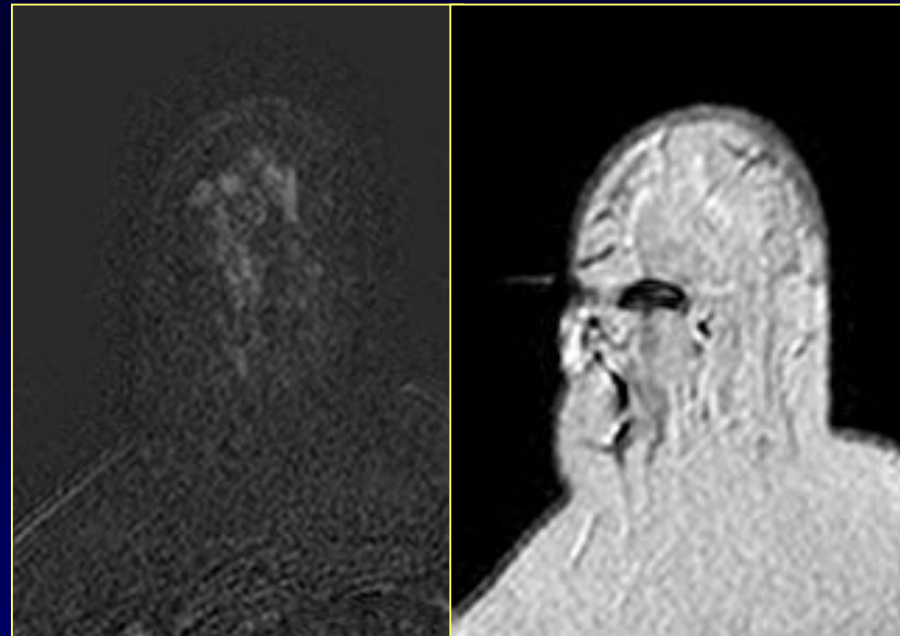


Biopsia RM-guidata

- ✓ Second look ecografico QQSS destra: disomogeneità ecostrutturale, non corrispondente per morfologia ed estensione all'area non mass RM

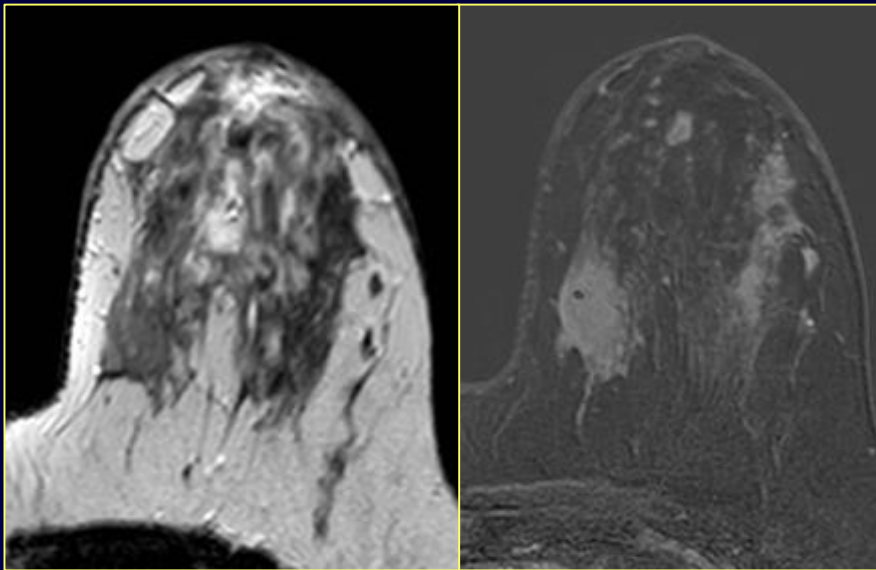
- ✓ Agobiopsia RM-guidata:

Esame istologico QQSS destra
CDIS solido G2



Punch biopsy

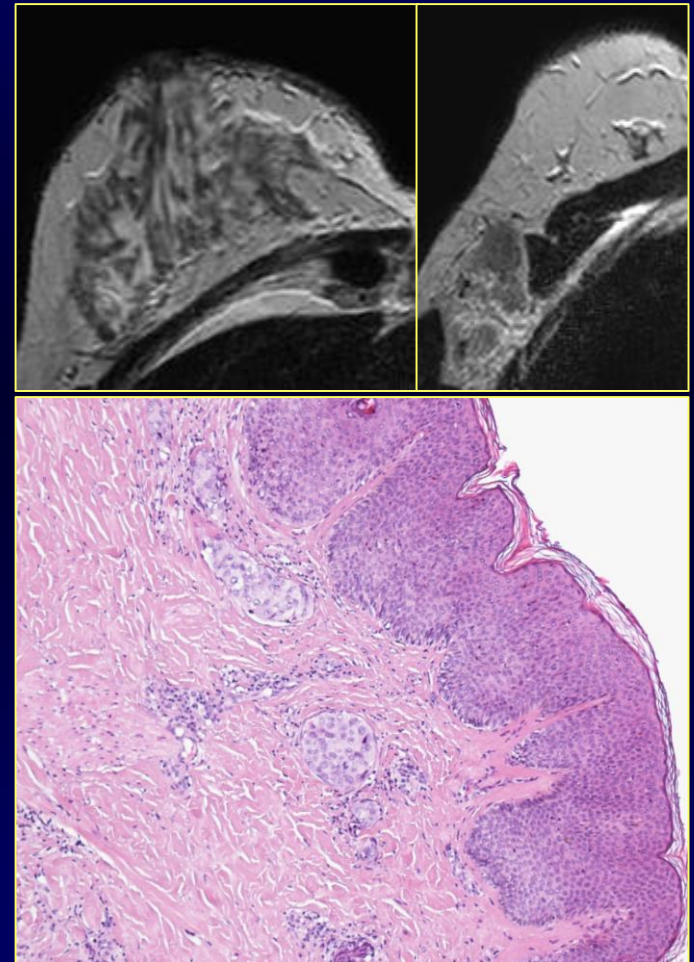
✓ Biopsia cutanea: **negativa**



Carcinoma infiammatorio

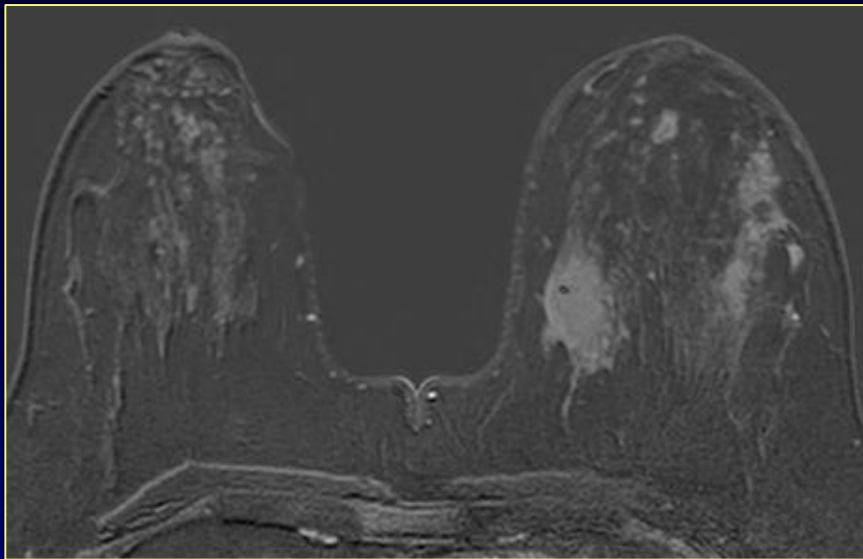
Edema mammario focale può essere associato al carcinoma infiammatorio

- ✓ **Aumento della permeabilità dei vasi formati per neoangiogenesi**
- ✓ **Ostruzione e dilatazione delle vie di drenaggio linfatico da emboli neoplastici**



Storia clinica

- ✓ 31 anni
- ✓ Non familiarità per neoplasia mammella / ovaio
- ✓ Mastite sinistra
- ✓ Neoplasia mammaria bilaterale



- ✓ Test genetico: **BRCA2**
(de novo?)
- ✓ Chemioterapia primaria
- ✓ Mastectomia bilaterale
- ✓ Esame istologico:
CDI G2 ypT1aN0 sinistra
CDIS multifocale G2 destra

SHORT COMMUNICATION

Breast and ovarian cancer predisposition due to *de novo* *BRCA1* and *BRCA2* mutations

L Golmard^{1,2}, C Delnatte³, A Laugé¹, V Moncoutier¹, C Lefol¹, K Abidallah¹, H Tenreiro¹, F Copigny¹, M Giraudeau¹, C Guy¹, C Barbaroux¹, G Amorim¹, A Briaux¹, V Guibert³, J Tarabeux^{1,2}, S Caputo¹, A Collet¹, P Gesta⁴, O Ingster⁵, M-H Stern^{1,2}, E Rouleau¹, A de Pauw¹, M Gauthier-Villars¹, B Buecher¹, S Bézieau^{3,6}, D Stoppa-Lyonnet^{1,2,7} and C Houdayer^{1,2,8}

Table 1. *BRCA1* and *BRCA2* *de novo* mutations reported in the literature and in this study

Gene	Mutation	Clinical features (age at diagnosis)	Breast/Ovarian cancer family history	Reference
<i>BRCA1</i>	c.3769_3770delGA	BC (< 40)	None	Tesoriero <i>et al.</i> (Australia) ⁵
<i>BRCA1</i>	c.5332+1G>A	Bilateral IDBC (38, 43)	Maternal aunt, BC (54)	Edwards <i>et al.</i> (UK) ⁶
<i>BRCA1</i>	Exons 1–12 deletion	IDBC (30)	None	Kwong <i>et al.</i> (China) ⁷
<i>BRCA1</i>	Gene deletion	Bilateral BC (28, 37)	None	Garcia-Casado <i>et al.</i> (Spain) ⁸
<i>BRCA1</i>	c.3494_3495delTT	BC (52), OC (53)	None	De Leener <i>et al.</i> (Belgium) ⁹
<i>BRCA1</i>	Mosaic exon 16 deletion	Bilateral BC (39,47)	Paternal cousin, BC (30)	Delon <i>et al.</i> (UK) ¹⁰
<i>BRCA1</i>	c.5468-2 A>G	OC (39)	Two paternal aunts, BC (57,75)	Case 1, this report
<i>BRCA1</i>	c.2296_2297delAG	BC (31)	None	Case 2, this report
<i>BRCA1</i>	Mosaic exons 1–13 deletion	IDBC (41)	Two sisters, BC (62,63) Paternal grandmother, BC (75)	Case 3, this report
<i>BRCA2</i>	c.2808_2811del	Multifocal BC (39)	Paternal cousin, BC (54)	Van der Luijt <i>et al.</i> (Netherlands) ¹¹
<i>BRCA2</i>	c.7260insA	Bilateral IDBC (35)	None	Robson <i>et al.</i> (USA) ¹²
<i>BRCA2</i>	c.8754+1G>A	IDBC (40)	Mother, BC (59)	Hansen <i>et al.</i> (Denmark) ¹³
<i>BRCA2</i>	c.5301insA	IDBC (35)	Paternal grandmother, BC (42)	Marshall <i>et al.</i> (USA) ¹⁴
<i>BRCA2</i>	c.51dupA	Bilateral IDBC (27, 37)	None	Diez <i>et al.</i> (Spain) ¹⁵
<i>BRCA2</i>	c.6082_6086delGAAGA	Bilateral BC (40,48)	Daughter, BC (31)	Case 4, this report

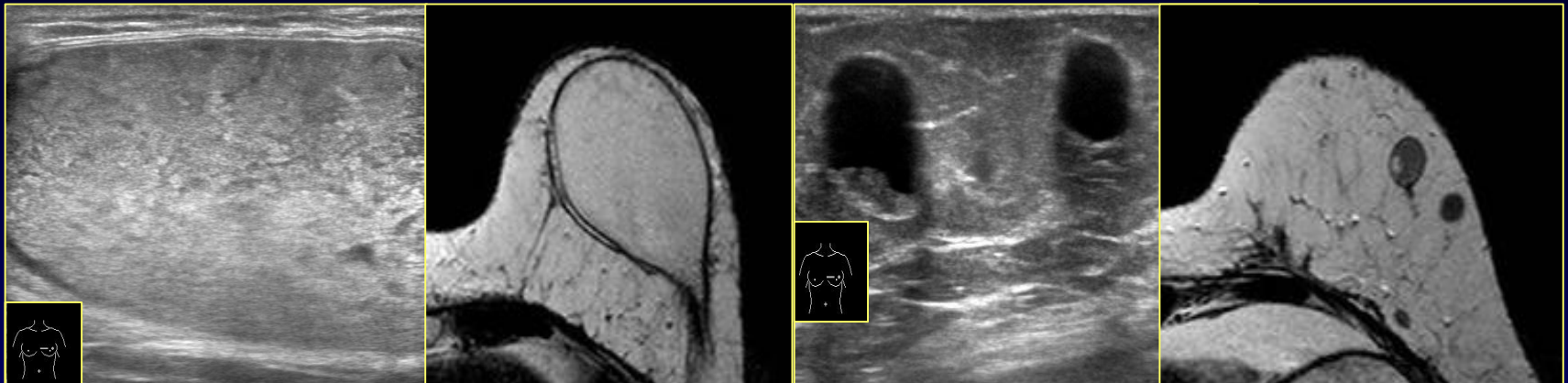
Abbreviations: BC, breast cancer; IDBC, invasive ductal breast carcinoma; OC, ovarian cancer.

Anamnesi

- ✓ 1996 (19 anni) Linfoma Hodgkin
RT mediastinica e chemioterapia
- ✓ 2007 (30 anni) neoplasia mammaria sinistra
Chemioterapia neoadiuvante
Mastectomia e linfadenectomia ascellare sinistra
Espansore
- ✓ E.I. Carcinoma duttale infiltrante (8 mm) ypT1N0
- ✓ Radioterapia

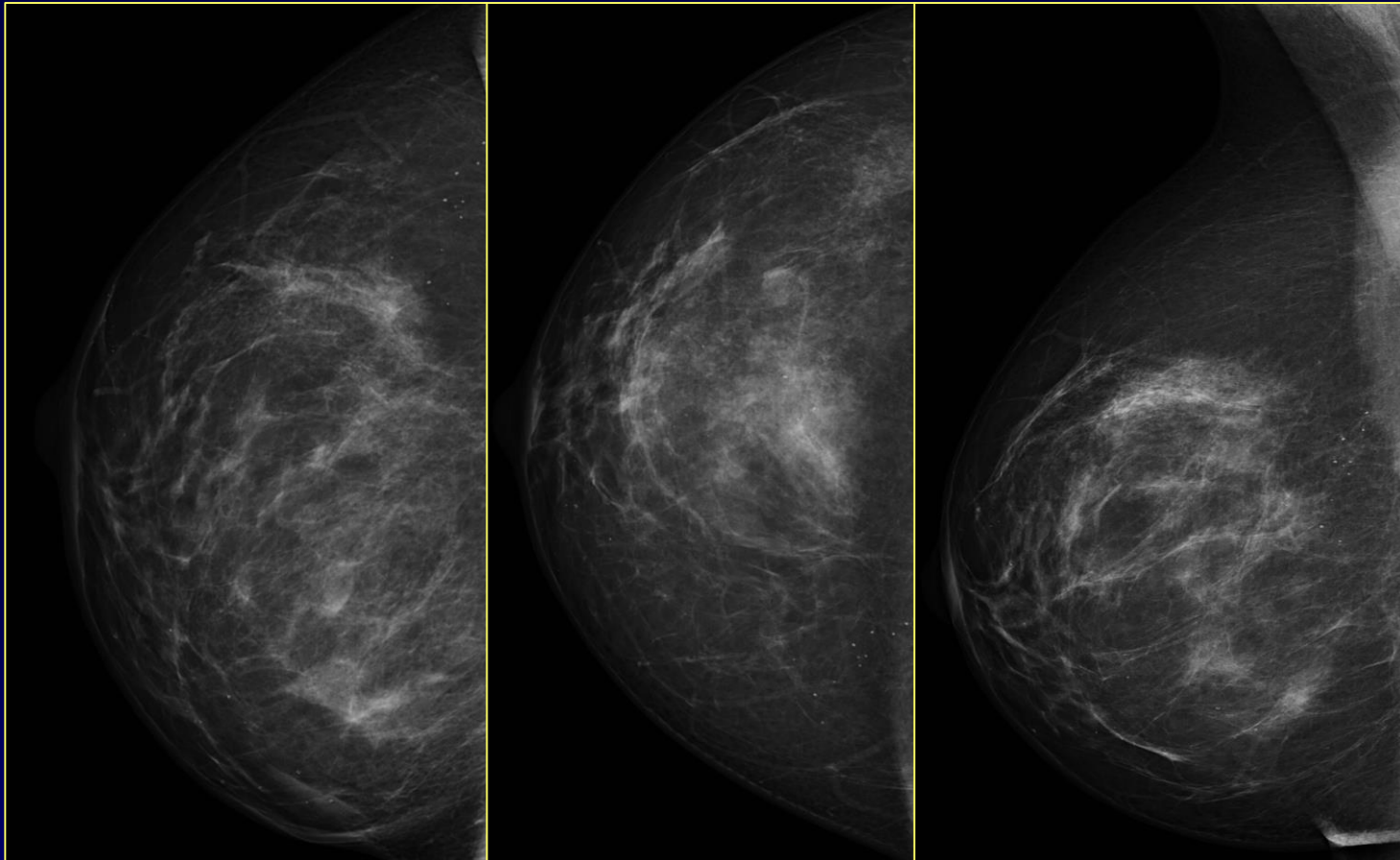
Anamnesi

- ✓ **Ottobre 2009** protesi mammaria sinistra
Mastopessi destra
- ✓ **Novembre 2009** rimozione protesi mammaria
sinistra per rigetto e ricostruzione con lipofilling



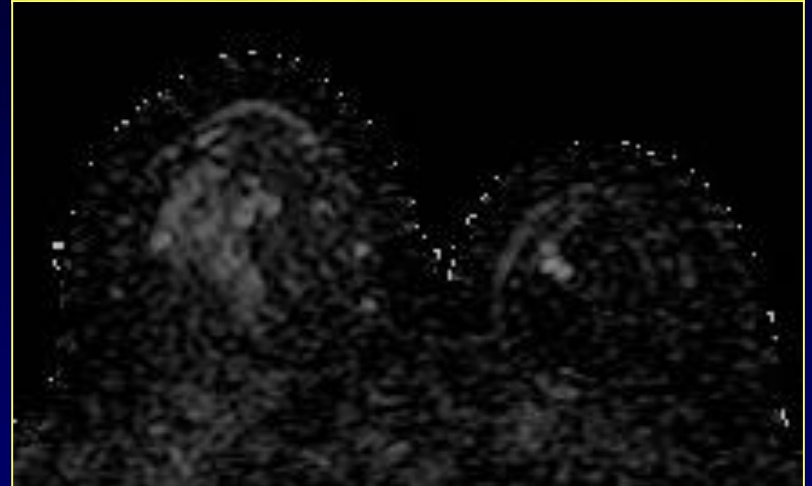
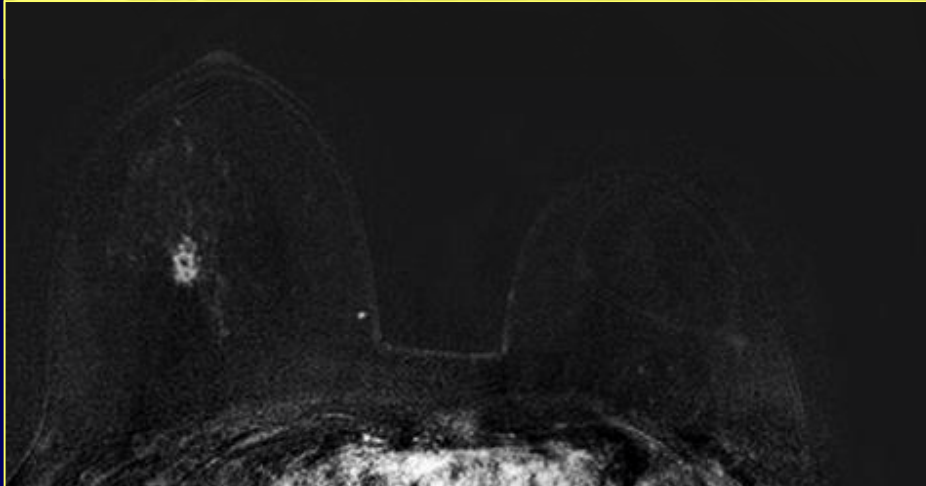
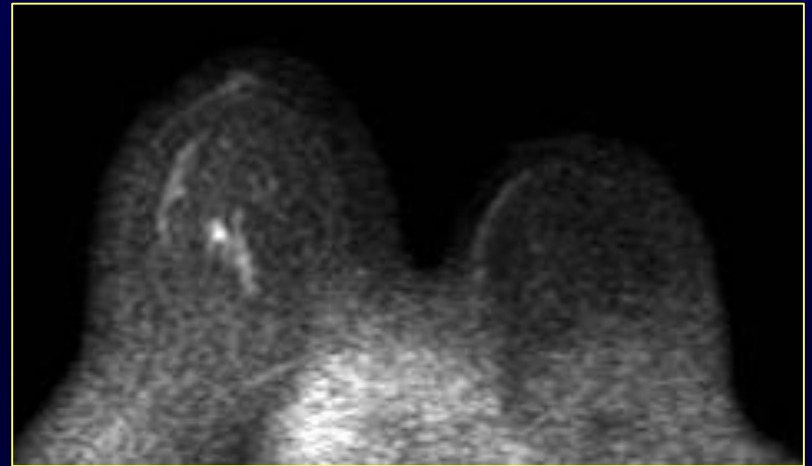
Mammografia – Ecografia

2015 (38 anni) Quadro mammografico ed ecografico invariato



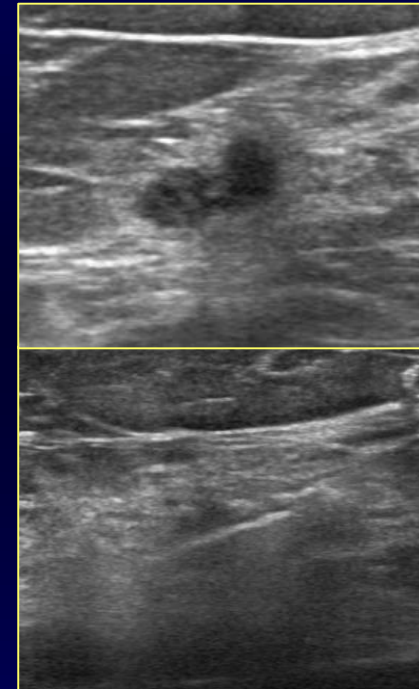
Risonanza magnetica mammaria

2015 (38 anni) Area di enhancement sospetta QSE destra



Second look ecografico

2015 (38 anni) Area di enhancement sospetta QSE destra



**E.I. Carcinoma duttale
infiltrante G3 TN**

Grazie per l'attenzione!

Radioterapia mediastinica

Prima dei 30 anni incremento del rischio di neoplasia mammaria

- ✓ **Anamnesi per valutare profilo di rischio individuale**
- ✓ **Mammografia annuale (o DBT)**
- ✓ **RM mammaria cmc a partire da 25 anni o da 8 dal termine del trattamento radiante**