

Dr. Andrea Otilia Onisim

Institute of Oncology “Prof.Dr.Ion Chiricuta” Cluj-Napoca, Romania

A BRCA2-Mutant, Triple-Negative, Early Breast Cancer Case

Case at presentation


- T.M., 35 year-old caucasian woman
- Patient characteristics:
 - one pregnancy → one birth at 28 years
 - no personal history of malignancies or other diseases
 - no family history of breast or ovarian cancer
- Physical exam (May 2014):
 - ECOG PS= 0
 - post-lumpectomy right upper breast scar
 - no breast or axillary palpable lesions

Primary treatment

- the patient presented to our Institute (**May 2014**) after **breast-conserving surgery (BCS)**
- the **histopathological report** revealed a 14 mm invasive medullary carcinoma in the upper external quadrant of the right breast :
 - resection margins less than 1 mm;
 - grade III Nottingham;
 - important lymphocyte infiltrate;
 - no tumor necrosis, no lympho-vascular or perineural invasion;
 - **HER2 - negative; ER, PR - negative; ki 67= 70%.**
- **Chest, abdomen, pelvis CT scan** – no distant metastases
- **re-excision and right axillary lymph node dissection (June 2014) :**
 - no tumor remnant
 - 23 axillary lymph nodes negative for tumor

 **Triple negative invasive medullary breast carcinoma pT1cN0M0, stage IA**

Postoperative treatment

- Adjuvant chemotherapy (Jun-Nov 2014):
 - **EC**: Epirubicin 100 mg/m² IV + Cyclophosphamide 600 mg/m² IV

on day 1 every 3 weeks for **4 cycles**
 - **CMF**: Cyclophosphamide 100 mg/m² PO on days 1-14 +
Methotrexate 40 mg/m² IV on days 1 and 8 +
5-FU 600 mg/m² IV on days 1 and 8 every 4 weeks for **4 cycles**
- Adjuvant radiotherapy (Jan 2015) → 3D-CRT of 50.40 Gy in 28 fractions

Follow-ups

Regular follow-ups every 4 months → 26 months disease-free interval

BRCA 1/2 testing (March 2016)



carrier of a class 5 BRCA2 mutation
(chr13; exon 24; BRCA2 - c.9253delA)

Recommendations:

- regular visits
- annual ipsilateral and contralateral mammography alternating with MRI
- gynecological consultation

Discussion

<i>Lifetime risk</i>	<i>BRCA1 mutation</i>	<i>BRCA2 mutation</i>
Breast cancer	65% [CI: 44–78%]	45% [CI: 31–56%]
Ovarian cancer	39% [CI: 18–54%]	11% [CI: 2.4–19%]

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- Up to 20% of women with TNBC carry a BRCA1/2 gene mutation
- BCS vs. Mastectomy in mutation carriers
- BRCA-mutant TNBC: - prediction of chemosensitivity (DNA-damaging agents)
 - benefit from platinum-based chemoT (neoadjuvant /adjuvant)
 - future perspectives → PARP inhibitors in the adjuvant setting

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- Risk reduction strategies to be discussed :
 - bilateral/ contralateral prophylactic mastectomy
 - prophylactic bilateral salpingo-oophorectomy