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A challenging case of triple negative, *BRCA* mutated breast cancer with unusual histology

Disclosure

I have no actual or potential conflict of interest
in relation to this presentation

Mrs. A.G., 35 y



- ◉ Paternal aunt with ovarian cancer at 57 y BRCA 1-mutated
- ◉ Menarche 12 y, oral contraception for 10 years, 2 sons, breastfeeding, small breast
- ◉ March 2016: clinical evidence of a left breast nodule, 6 x 5.5 cm, with inflamed skin. Ultrasound evidence of a left breast nodule, 3.5 x 2.2 cm, and suspected left axillary lymph nodes, max 1.2 x 0.8 cm.
- ◉ **Breast core needle biopsy:** metaplastic carcinoma ER neg, PgR neg, HER2 neg, Ki67 80%. **FNA left axillary lymph node:** no mets. cT4Nx.
- ◉ Total body CT scan: no distant mets.
- ◉ *Multidisciplinary discussion* among oncologists, surgeons, radiotherapists, radiologists, etc: *indication to neoadjuvant chemotherapy.*

- ◉ March-July 2016: **LH-RH analogue** as ovarian protection + **primary systemic therapy** (doxorubicin + docetaxel x 2, FEC x 4 because of infusion reaction to docetaxel). Local response to chemotherapy: breast nodule 3.2 x 2.4 cm (vs previous 3.5 x 2.2cm); left axillary lymph node max. 0.7 x 0.6 cm (vs previous 1.6 x 0.8 cm). Subsequent **modified radical mastectomy, axillary lymph nodes dissection and breast reconstruction**, metaplastic carcinoma with invasive ductal carcinoma foci and squamous carcinoma foci, ypT2N0 G3, ER neg, PgR neg, HER2 neg, Ki67 60%. **Adjuvant RT (50.4 Gy)**.
- ◉ Negative follow up until
- ◉ July 2017: total body CT scan evidence of a lung nodule. *Multidisciplinary discussion* among oncologists, radiologists and chest surgeons: **solitary lung nodulectomy**, lung metastasis of metaplastic carcinoma of the breast.
- ◉ September 2017 - January 2018: **carboplatin + paclitaxel** x 6.
- ◉ January - September 2018: NED.

A genetic counseling has been performed: **BRCA1 exons 16-17 deletion**; concomitant BRCA2 mutation c.8881G>A (p.Gly2961Ser) of uncertain significance. We discussed with the patient about prophylactic mastectomy and salpingo-oophorectomy and surveillance options.

CONCLUSIONS

- ◉ Triple negative breast cancer is still a therapeutic challenge
- ◉ Unusual histology: how to manage
- ◉ Importance of the MDT: when and how to consider metastasectomy
- ◉ BRCA mutation and its implications
- ◉ PARP inhibitors might enhance therapeutic options in this setting
- ◉ A role for multi-gene panel assay?

Thank you for your attention!