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BRCA mutation carrier patient: How to manage?

Clinical Case Presentation

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DISCLOSURE

No potential conflicts of interest

CASE PRESENTATION

- Jan 2010: Healthy, single, 34 year old woman with a known *BRCA1* mutation (185delAG) was being followed in high risk clinic
- Obstetric history: G0P0
- Age at menarche: 11
- Pre-menopausal

CASE PRESENTATION

- *BRCA1* status was known because her mother was a *BRCA1* carrier
- The mother was diagnosed with ovarian cancer at 43, was successfully treated, and later diagnosed with breast cancer at 60
- The patient had known about her *BRCA1* status from the age of 30

Q1. What is the optimal screening regimen for a healthy *BRCA1/2* carrier?

1. Clinical breast examination every 6-12 months from the age of 25 or 10 years before the youngest breast cancer onset in the family
2. Annual breast MRI started from the age of 25 with addition of annual mammography from the age of 30
3. Gynaecological exam with transvaginal US and CA125 level every 6 months from the age of 30
4. Breast ultrasound every 4 months
5. Options 1, 2, 3 and 4
6. Options 1, 2 and 3

(one answer)

Q2. The patient would like to know what are the best options for breast cancer risk reduction/prevention. What would you recommend?

1. Lifestyle modification, including breastfeeding, regular exercise with maintaining healthy body weight and limited alcohol consumption
2. Avoidance of hormone replacement therapy (HRT)
3. Use of oral contraceptive
4. Bilateral risk-reducing mastectomy
5. Options 1, 2 and 4
6. Options 1, 2, 3 and 4

(one answer)

CASE PRESENTATION

- Jan 2010: On annual screening MRI, a suspicious 1cm lesion is detected in the left breast
- Biopsy reveals a ER/PR/HER2-negative breast cancer
- Clinical stage is cT1N0

MRI, magnetic resonance imaging; ER, oestrogen receptor; PR, progesterone receptor, HER2, human epidermal growth factor 2

Q3. What surgical options would you recommend?

1. Mastectomy
2. Breast-conserving treatment (BCT)
3. Unilateral mastectomy with reconstruction
4. Bilateral mastectomy with reconstruction

(one answer)

CASE PRESENTATION

- The patient underwent bilateral mastectomy and reconstruction
- Her final diagnosis was a T-1.9cm N0, Grade 3 IDC, ER/PR/HER2-negative, Ki67-high
- In summary: **pT1cN0 triple negative breast cancer**
- Prior to commencing chemotherapy she is referred for fertility preservation counselling
- She is planned to receive dose dense AC-T

IDC, invasive ductal carcinoma; ER, oestrogen receptor; PR, progesterone receptor; HER2, human epidermal growth factor 2; AC-T, doxorubicin and cyclophosphamide, followed by paclitaxel or docetaxel

Q4. Which of the following is false about fertility in *BRCA* mutation carriers?

1. *BRCA* mutation is associated with reduced ovarian reserve and fertility
2. Women with *BRCA* mutation should be encouraged to complete childbearing before risk-reducing salpingo-ovariectomy (RRSO)
3. There are some fertility preservation options with oocyte and embryo cryopreservation for women planning RRSO
4. *BRCA* mutation carriers diagnosed with cancer should be informed about methods of fertility preservation before starting oncology treatment

(one answer)

CASE PRESENTATION

- June 2013: The patient is now 37 years old and is 3 years after the completion of adjuvant chemotherapy
- She is still single but very much wants a child
- She returns to you a year later and has not yet conceived

Q5. What do you recommend to her about reducing her risk of ovarian cancer?

1. Active surveillance with 6-monthly transvaginal US and serum CA125
2. Chemoprevention with use of oral contraceptives
3. Risk-reducing surgery with removal of both ovaries and fallopian tubes that should be performed at age 35-40
4. Risk-reducing salpingectomy at any age
5. Options 1 and 3

(one answer)

CASE PRESENTATION

- Despite the advice to complete risk reducing oophorectomy by 40, she persisted with her desire to conceive
- 2016: At her annual follow up she is 12 weeks pregnant
- Jan 2017: She delivers a healthy baby boy by cesarean section, she is 41 years old
- At her 6 week post-partum follow-up – there is a suspicious mass in pelvic ultrasound
- She underwent a laparotomy and was diagnosed with stage IIIC ovarian cancer
- Following optimal debulking surgery, she commenced six cycles of taxol-carboplatin