## Familial Breast Cancer

#### **Breast Cancer**

- Breast cancer is the commonest cancer in women, affecting 1 in 7 women during their lifetime
- The aetiology of breast cancer is multifactorial, including lifestyle, environmental, hormonal and genetic factors
- Identifying women who are at risk of familial breast cancer is important as they may be managed differently to the general population

#### Familial Breast Cancer

- Up to one-third of all breast cancer cases may be due to an inherited genetic susceptibility
- Only about 5% of breast cancer is due to the inheritance of highly penetrant breast cancer susceptibility gene (e.g. BRCA1 or BRACA2)
- The remaining inherited susceptibility is now recognised to be due to polygenic inheritance,
- 60-70% of familial risk remains unexplained and research is continuing to fully elucidate familial risk

### Task in pairs

- You see Ann in your surgery. She is a 37 year old lady who is upset as her sister has recently been diagnosed with breast cancer and she is worried that she might be at risk.
- What information do you want to find out from Ann?

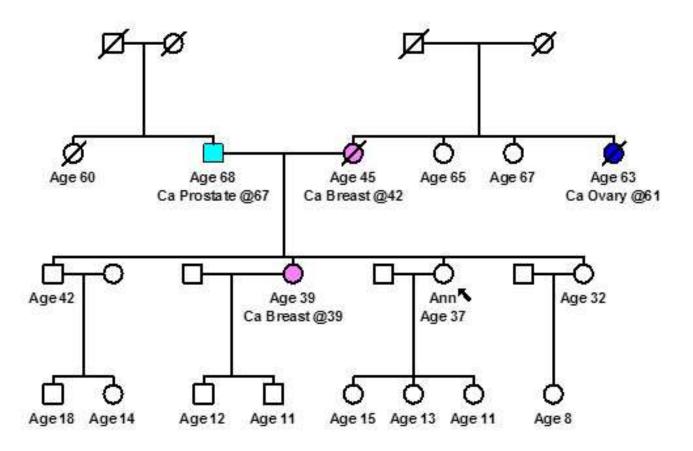
#### Information from Ann

- NICE recommends when a person presents with concerns about their family history a first and second-degree family history should be taken (including paternal as well as maternal relatives) in primary care
- Age of her sister
- Any other relatives affected with breast cancers (both paternal and maternal sides) and their ages at diagnosis
- Any male breast cancer
- Any family history of other cancers, in particular ovarian cancer, sarcomas in under 45s, childhood cancers, etc
- Any Jewish ancestry
- Any bilateral breast cancer

# Ann's Family History

- Ann has 2 sisters (32 and 39) and a brother (42) her one sister is 39 at diagnosis her other two siblings are unaffected.
- Her mother was diagnosed with breast cancer at age 42 and died when she was 46, her father is still alive aged 73 with prostate cancer diagnosed aged 67.
- Her father has one sister who died, she had no children.
- Her mother was one of 4 sisters the surviving sisters are aged 65, 67
  who are well with no significant PMH. A third maternal sister died aged 63 having been diagnosed with ovarian cancer at 61.
- Ann's brother has two children, a boy aged 18 and a girl aged 14. Ann's affected sister has two boys aged 11 and 12, her unaffected sister has a girl aged 8. Ann herself has three girls aged 11, 13 and 15. This generation of the family is unaffected.

# Ann's Family History



#### NICE GUIDELINES

NICE guidance on familial breast cancer identifies three levels of risk:

- Women at population risk, who can be managed and reassured in the primary care setting.
- Women at moderate risk who meet criteria for referral to secondary care (family history clinic) for increased breast surveillance.
- Women at high risk who meet criteria for referral to tertiary care (specialist cancer genetics clinic) for increased breast surveillance and assessment of eligibility for genetic testing.

### Referral to Secondary Care

- One first-degree relative under 40
- One first degree male relative
- One first degree relative with bilateral breast cancer, first diagnosed under 50
- Two first-degree or one first-, one second-degree relatives at any age
- One first- or second-degree relative diagnosed at any age and one first- or second-degree relative diagnosed with ovarian cancer at any age
- Three first-degree or second-degree relatives diagnosed at any age

#### Seek advice if

- bilateral breast cancer
- ovarian cancer
- Jewish ancestry
- sarcoma in a relative younger than age 45 years
- glioma or childhood adrenal cortical carcinomas
- complicated patterns of multiple cancers at a young age
- paternal history of breast cancer (two or more relatives on the father's side of the family)

#### Low risk

- Reassure that their risk is closer to the population risk
- However, if their family history changes (e.g. new diagnosis in another relative, find out more information) then they should come back for review
- Discuss
  - Breast awareness info
  - Lifestyle advice (diet, alcohol, smoking)
  - HRT and Contraception
  - May want to discuss other factors (breast feeding, family size and timing of pregnancies)
- Advice to attend national screening programmes when invited

#### **Moderate Risk**

- Lifetime risk of developing breast cancer is at 17-29%
- Give advice on risk reduction
- Eligible for additional screening (annual mammograms age 40-49)
- Consider chemoprevention

## High Risk

- Lifetime risk is greater than 30%
- Potentially eligible for genetic testing
- Eligible for extra screening including annual mammography age 40-59 and also MRI screening
- Chemoprevention
- Risk-reducing bilateral mastectomy /oophrectomy

### Chemoprevention

- Offer Tamoxifen for 5 years to pre-menopausal women at high risk
- Offer anastrazole to postmenopausal women (unless severe osteoporosis) at high risk
- Do not prescribe if increased risk of thromboembolism or endometrial cancer
- Do not offer with the woman has had a bilateral mastectomy.
- You can consider use in women at moderate risk

#### Task

 In pairs practice explaining to a patient who has come to see you after their mother was diagnosed with breast cancer at the age of 68 that they are at low risk

### Summary

- Concern about family history of breast cancer is common presentation to GP
- NICE (CG164) give guidelines on how to manage women presenting with a family history of breast cancer
- Low risk patients can be managed in primary care with advice
- Moderate to high risk patients will need onward referral