Familial ovarian cancer: a guide for primary care

Up to 20 per cent of women with ovarian cancer have a family history of the disease which can be conferred via the maternal or paternal line.

Ovarian cancer susceptibility genes

- Mutations in the BRCA1 and BRCA2 genes are commonly associated with ovarian cancer.
- A BRCA mutation significantly increases an individual's lifetime risk of developing ovarian cancer from two per cent (general population) to between 30–50 per cent for BRCA1 and
- 10–25 per cent for BRCA2.
- Mutations linked to Lynch Syndrome (also known as hereditary non-polyposis colorectal cancer or HNPCC) can also predispose individuals to ovarian cancer.
- Mutations in genes including Rad51C, Rad51D, STK11 and BRIP1 have also been shown to increase the risk of developing ovarian cancer.
- A proportion of familial cases are currently unexplained but are likely due to polygenic inheritance.

Establishing a family history: key questions

Maternal and paternal lines should be considered and evaluated in isolation. Cancer diagnoses from both sides of the family should not be combined to establish a case for genetic assessment.

The following questions help facilitate rapid assessment and determine whether a referral to clinical genetics services is necessary:

1 Has anyone on either side of your family had ovarian and/or breast cancer?

First-degree female relatives from the maternal and paternal side with a history of breast and/or ovarian cancer should be considered.

Ovarian cancer and breast cancer often develops under the age of 50 years in individuals who carry a BRCA1 or BRCA2 mutation. Mutation in either gene can predispose male relatives to breast cancer.

2 How big is your family? How many men vs. women in the family?

In small families or families with a larger proportion of men it is likely to be more difficult to establish a family history.

3 Hs there any Ashkenazi Jewish history (where appropriate)?

BRCA1 and BRCA2 mutations are more common in individuals of Ashkenazi Jewish descent.

Did you know?
Ovarian cancer susceptibility genes can be transmitted by either parent. Assess the paternal as well as the maternal line.

Guidelines

Local referral guidelines will vary, but broadly speaking the following warrants referral to clinical genetics services.

Type of cancer	Relatives affected	Age at diagnosis
Female breast cancers only	1 first-degree relative	Under 40
Female breast cancers only	2 first-degree relatives 1 first-degree and 1 second-degree relative 3 first-degree relatives 3 second-degree relatives	Any age
Male breast cancer	1 first-degree male relative	Any age
Bilateral breast cancer	1 first-degree relative	Under 50 for diagnosis of first cancer
Breast and ovarian cancer	1 first-degree relative with breast cancer and 1 first-degree relative with ovarian cancer 1 first-degree relative with breast cancer and 1 second-degree relative with ovarian cancer 1 second-degree relative with breast cancer and 1 first-degree relative with ovarian cancer	Any age

First-degree relatives include mother, father, daughter, son, sister, brother.

Second-degree relatives include grandparent, aunt, uncle, niece, nephew, half-sister, half-brother

Your local clinical genetics service may provide a template form for patients to take home and complete. Forms can be returned to clinical genetics services for assessment and appointments issued directly to the patient.

Reducing risk

For women at high risk, the current medical advice is to have their ovaries and fallopian tubes removed after having completed their families.

Surveillance using CA125 and/or pelvic ultrasound for women at high risk of developing ovarian cancer is not currently supported by clinical evidence, and is not recommended for routine use. GPs should advise caution to women seeking the services of private providers.

How Target Ovarian Cancer can help you

- Access our accredited online learning modules at **targetovariancancer.org.uk/cpd**
- Order our booklet Genetic testing and hereditary ovarian cancer written for women with ovarian cancer wanting to find out more about genetic testing at **targetovariancancer.org.uk/orderguides**
- Women concerned about hereditary ovarian cancer and genetic testing can access support and information through our nurse-led support line. Call us on **020 7923 5475** or ask a question at **targetovariancancer.org.uk/supportline**

Target Ovarian Cancer