Familial cancer guidelines
Cancer genetics referral guidelines

The following family histories are suggestive of an inherited cancer predisposition and would be appropriate for referral. This list is a guide only, and is not comprehensive. If you are unsure whether to refer, please telephone 01223 216446 between 9am and 5pm and speak to the on-call team.

- Referrals can be made for individuals affected by cancer or for their close relatives. If possible, refer the affected person in the first instance.
- Close relatives are father/mother, brother/sister and son/daughter.
- Affected relatives must be close relatives of each other, through either the maternal or the paternal side.
- In breast/ovarian cancer families, women related through unaffected men are considered close relatives (i.e., a paternal family history may still be significant).
- Few families have a mutation in a known cancer gene. For many it will not be appropriate to offer genetic testing.
- In general, a living relative affected by cancer is tested prior to offering testing to unaffected family members. This maximizes the chance of finding the family gene mutation if one exists.
- Not all individuals referred will be eligible for additional screening.
- Not all individuals referred will be offered a clinic appointment; some may receive a risk assessment by letter.

Breast/ovarian cancer families

- Breast cancer under 40
- Grade 3 triple negative breast cancer under 50
- Two cases of breast cancer (including bilateral breast cancer), average under 50
- Three cases of breast cancer, average under 60
- Four or more cases of breast cancer
- Male breast cancer at any age plus breast cancer (male or female) under 60
- Breast cancer and ovarian cancer in a single individual
- High grade serous papillary ovarian cancer under 60
- Ovarian cancer at any age plus breast cancer under 60
- Two or more cases of ovarian cancer
- A woman with Ashkenazi Jewish ancestry and any family history of breast or ovarian cancer
• Families in whom a BRCA1 or BRCA2 mutation has been identified

Colorectal cancer families

• Colorectal cancer under 50
• Two cases of colorectal cancer, average under 60
• Colorectal cancer plus one gastrointestinal, endometrial, ovarian, renal or urinary tract cancer, average under 60
• Three or more cases of gastrointestinal, endometrial, ovarian, renal or urinary tract cancers
• Multiple gastrointestinal polyps
• Families in whom a Lynch syndrome (HNPCC) or familial adenomatous polyposis (FAP) mutation has been identified

Prostate cancer families

• Prostate cancer under 60
• Two cases of prostate cancer, at least one under 65
• Three or more cases of prostate cancer
• Prostate cancer under 70, with a strong family history of breast/ovarian cancer

Pancreatic cancer families

• Pancreatic cancer under 50
• Pancreatic cancer with a previous malignancy (eg breast cancer)
• Two or more cases of pancreatic cancer at any age

Other families

• Three or more primary cancers in a single individual
• Three or more cases of cancer at the same site
• Any two of: sarcoma, breast cancer, brain tumour, leukaemia or adrenal cortical tumour, one under 45
• Childhood cancer plus one close relative with cancer
• Any individual or family with an unusual pattern of cancer e.g rare tumours or young ages at diagnosis
• Families with a known cancer predisposition syndrome e.g. Li-Fraumeni, MEN1,
MEN2, Von Hippel-Lindau
Breast and ovarian cancer guidelines

Approximately 1 in 8 women in the United Kingdom develop breast cancer over their lifetime, and around 5-10% of these have a hereditary cause.

All women are eligible for 3-yearly mammography in the NHS Breast Screening Programme and will be called for their first mammogram between the ages of 47 and 50. For women with a strong family history of breast and ovarian cancer, additional screening is recommended.

Referral for additional screening is only made following accurate assessment of family history, please see referral guidelines and documents below.

Genetic testing for BRCA1 and BRCA2 is only appropriate in a small number of families, and usually begins in a relative who has been affected by breast or ovarian cancer. Testing is currently offered when there is a >10% chance of finding a mutation, please see document below.
Colorectal cancer guidelines

Approximately 1 in 20 people in the United Kingdom develop colorectal cancer (CRC). Five to 10% of CRCs are due to an underlying genetic condition. The two most common genetic conditions associated with a significantly increased risk of CRC are familial adenomatous polyposis (FAP) and Lynch syndrome (hereditary non polyposis colonic cancer, HNPCC).

In FAP the risk of CRC is up to 100% and in Lynch syndrome the risk is up to 80%. Identification of those with these two conditions allows cost effective screening and a reduction in cancer related morbidity and mortality.

There are also a number of rare conditions and genes leading to a moderately increased risk of CRC for which it may be possible to offer genetic testing and/or additional screening.

For more information download the guidelines below.