

Referral criteria for Cancer Genetics service

1. Patient meets criteria for constitutional/germline genetic testing as set out in the NHS National Genomic Test Directory england.nhs.uk/publication/national-genomic-test-directories*
2. Genetic testing has been undertaken and the patient, or their blood relative, has a likely pathogenic or pathogenic variant in a cancer predisposition gene.
3. Individual with personal history of multiple malignant/ benign tumours suggestive of an inherited cancer predisposition (particularly where one or more cancers are diagnosed < age 50).
4. There is a strong family history of cancer suggestive of inherited cancer predisposition, or family history of breast or bowel cancer, meeting one of the family history criteria below.

Family history criteria

These criteria guide referrals but are not exhaustive. Please provide full details for complex family histories or where inherited cancer predisposition is suspected but criteria are not clearly met.

These criteria may apply to a patient affected with a relevant cancer themselves, or to their close relatives where the family history meets the criteria below.

Family history suggestive of inherited cancer predisposition criteria

1. 3 or more individuals on the same side of the family diagnosed with cancers at the same site.
2. 3 or more individuals diagnosed with cancer under age 50, on the same side of the family.
3. 3 or more individuals on the same side of the family with one of these cancers, all diagnosed under age 75: breast, epithelial ovarian, colorectal, prostate, pancreas, melanoma, thyroid, renal, sarcoma, or non-melanoma skin tumours.

Breast/ovarian cancer family history criteria

1. 1 female diagnosed with breast cancer under age 40.
2. 1 male diagnosed with breast cancer at any age.
3. 1 female diagnosed with bilateral breast cancer.
4. 2 females diagnosed with breast cancer at any age, including at least 1 first-degree and 1 second-degree relative, on the same side of the family.
5. 2 individuals on the same side of the family: 1 female diagnosed with breast cancer and 1 individual diagnosed with epithelial ovarian cancer.
6. 2 individuals diagnosed with epithelial ovarian cancer, on the same side of the family.

A single female relative with breast cancer diagnosed at age 40 or over, with no other relevant family history, does NOT meet referral criteria.

Colorectal cancer family history criteria

1. 1 individual diagnosed with colorectal cancer under age 50.
2. 2 individuals diagnosed with colorectal cancer, at any age, on the same side of the family.
3. 3 or more individuals diagnosed with colorectal cancer, at any age, on the same side of the family.
4. 1 individual diagnosed with colorectal or endometrial cancer and 2 other individuals on the same side of the family with Lynch-related** cancers, all diagnosed under age 75.
5. 1 first-degree relative diagnosed with serrated polyposis or multiple bowel polyps.

* Test eligibility includes all living affected individuals diagnosed with breast cancer age <40, or with colorectal/ endometrial cancer age <40, or with epithelial ovarian cancer at any age

**Lynch-related cancers include endometrial, epithelial ovarian, urothelial, TCC renal pelvis, cholangiocarcinoma, small bowel, upper GI, glioblastoma, endocervical, sebaceous carcinoma, prostate, gastric, and pancreatic cancers.

Important notes

- Close relative means first- or second-degree relative: parent, child, sibling, aunt, uncle, or grandparent.
- Wherever possible, genetic testing should be carried out in someone who has themselves been diagnosed with the relevant cancer. The affected person should therefore be referred first rather than unaffected family members.
- Urgent referrals should be clearly marked as urgent where the outcome may affect immediate management. Please include the reason for urgency, the relevant treatment or surgical decision, and any key dates such as operation date.
- Meeting referral criteria does not necessarily mean we are able to offer an appointment; we may give risk assessment and management advice by letter.
- Patients offered an appointment are not automatically offered genetic testing.
- Genetic testing is very unlikely for a family history limited to lung, oesophageal, or cervical cancer without other significant features.
- The national genomic test directory sets out eligibility criteria for genetic testing. Some specialists can request these tests directly through current NHS genomic medicine mainstreaming pathways. Please contact our team for further information.
- **Sending a sample for genetic testing to the genetics laboratory is not a referral to Clinical Genetics. If Clinical Genetics input is required, please submit a separate referral to us with relevant clinical and family history information.**

Contacting the team

For Clinical Genetics advice or referral queries, please contact the team by email at clinical.genetics@ouh.nhs.uk or by telephone on **01865 226 024**.

The Clinical Genetics team has an on-call advisory service for urgent queries. The team can be contacted by telephone Monday to Friday, 09:30–16:00.