St James's Hospital
Cancer Genetics Service
Clinical Instruction
Author: Rosie O' Shea & Aoife O'Shaughnessy-Kirwan
Authorised by: Prof Karen Cadoo & Prof David Gallagher
Date: 31.03.25
Date of issue: 31.03.25

St. James's Hospital
Cancer Genetics - HOPe

Referral criteria for the Cancer Genetics Service at St James's Hospital

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Background

The following referral criteria act as a guide only and cannot comprehensively cover all clinical scenarios. If you are unsure whether your patient meets our referral criteria please contact us on (01) 4103890/ (01) 4103759 or e-mail cancergenetics@stjames.ie to discuss the referral further with a member of the Cancer Genetics team. Pending implementation of a national test directory as planned by the National Genetics and Genomics Office, the NHS genomic test directory may be a useful reference.

Fulfilling the referral criteria does not automatically mean a person will be seen in the Cancer Genetics clinic or offered genetic testing. Some individuals may receive an assessment by letter, with the assessment being informed by the information provided in the Family History Questionnaire (FHQ). If possible, we would recommend that a family member who is affected with cancer is referred in the first instance, as in most cases, genetic testing needs to start in an affected family member.

If your patient does not meet our referral guidelines, you may wish to consider referral to the relevant screening service, to assess whether or not they might be eligible for enhanced surveillance. Please consult the relevant guidelines.

It is important to be aware that guidelines for genetic assessment are changing overtime and this guide will be updated periodically to reflect this.

Referring a patient

Direct all referrals through our online referral form via e-mail cancergenetics@stjames.ie or to:
Prof. Karen Cadoo/ Prof. David Gallagher,
Consultant Medical Oncologist & Cancer/Medical Geneticist
Cancer Genetics Service
St. James's Hospital
James's Street
Dublin 8.

Please note, assessments are made on the basis of the information provided and it is the responsibility of the referrer to ensure the information is accurate and complete, insofar as possible.

Your patient will be triaged into one of three categories; urgent (1-4 weeks for an appointment), as soon as possible (1-16 weeks for an appointment) or routine (wait time of approximately 18-24 months currently).

Please note, we are a cancer genetics service only; patients requiring general genetics assessment should be referred to the appropriate service.

Urgent and ASAP referrals

The urgent and ASAP categories are for patients where a genetic test result will modify current cancer management. We will try to see these patients in a timely manner, however it is important to note that this is not something we will always be able to achieve within the desired time frames. It is important that any referral highlight how genetic testing could affect care if urgent or ASAP assessment is required.

Direct order *BRCA1/2* genetic testing is available for certain cancer types. Additional information on this is available from the National Cancer Control Programme (NCCP).

Referral criteria

First Degree Relative (FDR) = mother, father, sibling, or child.
Second Degree Relatives (SDR) = parent's siblings, nephew/niece or grandparents.

Referral type	Referral indications
Known cancer predisposition syndrome	• Pathogenic/likely pathogenic gene variant already identified in a family member
Breast Cancer	An individual or family with: • Invasive breast cancer at/under 45 years • Triple negative breast cancer (ER-, PR-, HER2-) at/under 60 years • Bilateral breast cancer • Male breast cancer at any age • Breast cancer and has a family history of breast and/or ovarian cancer or adopted or with limited family structure • Multiple cases of breast cancer, for example, two affected family members who were diagnosed at a younger age, or who have triple negative or bilateral diagnoses. Or where there are three breast cancer affected relatives. CanRisk may be used to determine eligibility for testing in this scenario. • Both breast and ovarian cancer; a single individual who develops both cancers. • Ashkenazi Jewish ancestry or ethnicities where there are known founder pathogenic variants. Depending on personal/family history CanRisk may be used to determine eligibility for testing in this scenario. • Individuals without a breast cancer diagnosis who have a significant family history as above and no living relative available to test and who meet a CanRisk probability of ≥ 10% to identify a cancer predisposition variant
Ovarian Cancer	An individual or family with: • A diagnosis of (non-mucinous) ovarian cancer

	• One FDR with both breast and ovarian cancer • Two relatives diagnosed with ovarian cancer • Rare ovarian tumours, such as; Ovarian sex cord tumour with annular tubules, Sertoli-Leydig cell tumour, Small cell carcinoma of the ovary hypercalcaemic type, ovarian fibroma/ leiomyoma
Colorectal and Endometrial Cancer	An individual with: • Colorectal or endometrial cancer diagnosed under age 50 years • Colorectal or endometrial cancer diagnosed with abnormal tumour immunohistochemistry for mismatch repair proteins. In cases of MLH1/PMS2 loss BRAF and/or MLH1 hypermethylation status should also be assessed prior to referral. Colorectal and/or endometrial cancer with a family history of colorectal and/or endometrial cancer, and/or a family history of gastrointestinal, renal, urinary tract, or ovarian cancer at any age • Significant personal or family history of polyps • One close relative with colorectal cancer and/or endometrial cancer diagnosed under 50 years AND a family history of ovarian, urothelial, gastric or hepatobiliary cancer • Two or more close relatives with colorectal cancer and/or endometrial cancer • Individuals with a significant family history as above and no living relative available to test • Familial Adenomatous Polyposis (FAP) or *MutYH*-associated polyposis (MAP) diagnosis in the family • Lynch syndrome confirmed/suspected
Prostate Cancer	An individual or family with: • Prostate cancer at/under 50 years • Two close relatives with prostate cancer younger than 60 years • Three or more FDRs or SDRs with prostate cancer at any age • Prostate cancer (Gleason score >7) and two or more cases of breast, ovarian, and/or pancreatic cancer in close relatives • Metastatic castrate-resistant prostate cancer • Prostate cancer and Ashkenazi Jewish ancestry
Melanoma	An individual or family with: • Three or more cases with melanoma or pancreatic cancer in close relatives • Multiple separate melanomas in one person • Melanoma and pancreatic cancer in the same person
Pancreatic Cancer	An individual or family with: • Metastatic pancreatic cancer • Pancreatic cancer under 60 years • Pancreatic cancer with a previous malignancy (e.g. breast cancer) • Pancreatic cancer and a strong family history of breast, ovarian, prostate cancer, or melanoma

	• Two or more cases of pancreatic cancer at any age in close relatives
Unusual patterns of Cancer	An Individual or family with: • Multiple primary cancers in one individual • Multiple cancers or sarcomas at a young age (<45 years) on the same side of the family • Renal cancer diagnosed below 45 years or multiple renal cancers in close relatives • Renal cancer and cutaneous/uterine leiomyomata (fibroids) • One or more cases of: medullary thyroid cancer, adrenocortical cancer, phaeochromocytoma, paraganglioma, Wilm's tumour, retinoblastoma • Unusual pattern of multiple cancers, especially with young age of diagnoses • MEN1 – two cases of pancreatic cancer, parathyroid cancer/hyperplasia or pituitary adenoma • MEN2 – two cases of thyroid cancer, parathyroid adenoma/carcinoma or adrenal phaeochromocytoma • Two or more cases of papillary or follicular thyroid cancer • Diffuse gastric cancer under age 50 years • Diffuse gastric cancer and lobular breast cancer in the same person, or in two people who are first degree relatives to each other

For all the above criteria, referrals for individuals or families with the following are important;

- a pathogenic/likely pathogenic variant in a cancer predisposition gene requiring post-test genetic counselling
- a variant of uncertain significance (VUS) for genetic counselling
- a negative test result BUT with a personal or family history that raises significant concern for a familial cancer syndrome