

Cancer Genetics Referral Criteria

Clinical Genetics Unit

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Family history form: <http://tinyurl.com/goshcafhq>



North East Thames
Regional Genetics Service

Please e-mail all referrals to us from an nhs.net account to: gosh-tr.clinicalgenetics@nhs.net.

If you wish to seek advice about a patient, please telephone or e-mail us.

Meeting our referral criteria does not necessarily mean that your patient will be offered a genetic test.

Patients diagnosed with cancer:

See below criteria.

Please send histology for your patient's cancer diagnosis with the referral.

Please include who has had cancer in the family, the types of cancer, and the ages of diagnosis. Alternatively, include a completed [family history form](#) from the patient.

Patients not diagnosed with cancer:

All individuals from a family with a *confirmed* cancer predisposition syndrome should be referred, with the name and date of birth of a relative who carries the familial mutation, if possible.

Genetic testing, when appropriate, is most informative when performed first in a relative who has had the type of cancer under investigation.

IF YOUR PATIENT HAS NOT DEVELOPED CANCER AND NO GENETIC TESTING HAS TAKEN PLACE IN THE FAMILY, PLEASE INFORM THE PATIENT THAT THEIR AFFECTED RELATIVE NEEDS TO BE ASSESSED FIRST.

If all affected relatives are deceased, refer with a completed [family history form](#).

Genetic testing criteria are set nationally by NHS England and may change over time. For more detailed criteria and testing recommendations please see: <https://www.england.nhs.uk/publication/national-genomic-test-directories/>

Patient has breast or ovarian cancer	Manchester Scoring System	
	Cancer and age at diagnosis	Score
<ul style="list-style-type: none">Breast cancer diagnosed at 30 years or youngerTriple positive breast cancer diagnosed at 35 years or youngerBilateral breast cancers, both diagnosed under 50 yearsTriple negative breast cancer diagnosed under 60 yearsBreast cancer under 45 years and a first degree relative with breast cancer under 45 yearsBreast cancer AND Manchester Score of 15 or greaterOvarian cancerMale breast cancerAshkenazi Jewish ancestry and breast cancer at any agePatient is unaffected with cancer but all affected family members are deceased, AND Manchester Score of 17 or greater	Female breast cancer <30	11
	Female breast cancer 30-39	8
	Female breast cancer 40-49	6
	Female breast cancer 50-59	4
	Female breast cancer >59	2
	Male breast cancer <60	13
	Male breast cancer >59	10
	Ovarian cancer <60	13
	Ovarian cancer >59	10
	Pancreatic cancer	1
	Prostate cancer <60	2
	Prostate cancer >59	1
	<i>Each side of the family to be calculated separately</i>	

Pancreatic Cancer
<ul style="list-style-type: none">Patient diagnosed with pancreatic cancer younger than 50 yearsPatient diagnosed with pancreatic cancer younger than 60 years, plus:<ul style="list-style-type: none">- patient also diagnosed with breast cancer or melanoma younger than 60 years, or- patient also diagnosed with ovarian cancer at any age, or- two relatives diagnosed with pancreatic, breast, melanoma or ovarian cancer

Endocrine Cancer
<ul style="list-style-type: none">Patient diagnosed with medullary thyroid carcinoma at any ageTwo or more cases of endocrine tumours in an individual or familyPatient diagnosed with paraganglioma / pheochromocytomaPatient has gastrointestinal stromal tumour (GIST) diagnosed under 50 OR at any age with a family history of GIST, pheochromocytoma or paragangliomaPatient diagnosed with hyperparathyroidism under 35 years or parathyroid carcinoma or familial hyperparathyroidism at any agePatient diagnosed with pituitary tumour under 20 years or pituitary macroadenoma younger than 30 years

Colorectal Cancer / Polyposis

Colorectal cancer:

- Loss of proteins on mismatch repair immunohistochemistry (MMR IHC). If loss of MLH1 and PMS2, please first carry out MLH1 promoter hypermethylation studies and refer if not hypermethylated.
- Patient diagnosed with bowel cancer younger than 50 years
- Patient diagnosed with two Lynch syndrome-related cancers* where one is either colorectal or endometrial cancer
- Patient diagnosed with bowel cancer and family reaches Amsterdam Criteria**
- Patient diagnosed with a non-colorectal Lynch syndrome-related cancer* younger than 50 years e.g. endometrial or ovarian cancer (MMR IHC testing to first be performed locally, if possible)
- Patient is **unaffected with cancer** but all affected family members are deceased, AND they have a first degree relative with bowel cancer younger than 30 years, OR they have two first degree relatives diagnosed with a Lynch syndrome-related cancer* younger than 60 years, OR family reaches Amsterdam Criteria**

**Lynch-related cancers comprise: colorectal, endometrial, endocervical, ovarian, pancreatic, ureteric, transitional cell cancer of renal pelvis, gastric, hepatobiliary tract, small bowel, glioblastoma, pancreatic, prostate, multiple sebaceous adenomata, multiple sebaceous epitheliomas, multiple keratoacanthomas, and sebaceous carcinoma.*

*** Amsterdam Criteria: ≥ 3 closely related family members diagnosed with Lynch-related cancers over ≥ 2 generations with ≥ 1 case diagnosed < 50 years*

Polyposis:

- Patient diagnosed under 40 years: 5 or more adenomas
- Patient diagnosed under 60 years: 10 or more adenomas, or 5 or more adenomas with first degree relative with 5 or more adenomas younger than 60
- Patient diagnosed at any age: 20 or more adenomas, or bowel cancer at any age and 5 or more adenomas
- Patient has unusual types of polyps such as juvenile, Peutz-Jeghers or hamartomatous polyps
- Patient has clinical signs indicating potential diagnosis of Familial Adenomatous Polyposis e.g. FAP-related CHRPE

Renal Cancer

- Patient diagnosed with any type of renal cancer at 40 years or younger
- Patient diagnosed with type 2 papillary renal cancer at 50 years or younger
- Patient diagnosed with multifocal or bilateral renal cancers
- Patient diagnosed with renal cancer at any age, with family history of renal cancer

Diffuse Gastric Cancer and Lobular Breast Cancer

- Patient diagnosed with diffuse gastric cancer under 40 years
- Patient diagnosed with diffuse gastric cancer at any age and further family history of diffuse gastric cancer and/or lobular breast cancer
- Patient diagnosed with lobular breast cancer and family history of diffuse gastric cancer

Skin Cancer

- Patient diagnosed with at least 2 melanomas under 30 years
- Patient diagnosed with melanoma with atypical moles and/or family history of melanoma, pancreatic cancer, mesothelioma or uveal melanoma
- Patient diagnosed with BAP-oma (atypical spitz naevus with loss of BAP1 on IHC)
- Patient diagnosed with basal cell carcinoma under 30 years
- Patient diagnosed with greater than 5 basal cell carcinomas at any age
- Patient diagnosed with basal cell carcinoma in presence of syndromic features suggestive of Gorlin Syndrome

Other Criteria

- Patient has two or more primary cancers diagnosed under 60 years, or three or more primary cancers under 70 years
- Patient and first degree relative with any two of: sarcoma, breast cancer, brain tumour, leukaemia, adrenal cortical tumour or choroid plexus tumour
- Patient has features in keeping with Cowden syndrome (PTEN Hamartoma Tumour Syndrome)
- Patient diagnosed with uveal melanoma
- Patient diagnosed with malignant mesothelioma and a family history of either malignant mesothelioma or uveal melanoma or BAP-oma (atypical spitz naevus)
- Patient has features in keeping with Von Hippel Lindau (VHL) syndrome, including retinal angioma, spinal or endolymphatic sac tumour or cerebellar haemangioblastoma, or other VHL criteria
- Patient has one or more schwannoma under 25 years, or two or more schwannomas at any age, or a schwannoma with a close relative with schwannoma

Please also send us an informal query if unusual patterns of cancer, especially if young onset.