Cancer Genetics Referral Criteria

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North East Thames
Regional Genetics Service

Please e-mail all referrals meeting criteria below from an nhs.net account to: gos-tr.clinicalgenetics@nhs.net.
If patients do not meet criteria but there are unusual patterns of cancer or particularly young age of onset please refer for advice. 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, and where they were seen, if possible.
IF YOUR PATIENT HAS NOT DEVELOPED CANCER AND NO GENETIC TESTING HAS TAKEN PLACE IN THE FAMILY, PLEASE INFORM THE PATIENT THAT THEIR LIVING AFFECTED RELATIVE NEEDS TO BE ASSESSED FIRST, AS THIS IS MOST INFORMATIVE.
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. For detailed criteria for each genetic test (designated by an R number) and testing recommendations please see: https://www.england.nhs.uk/publication/national-genomic-test-directories/

Breast, Ovarian and Prostate Cancer

Patient has:

- Breast cancer diagnosed at 40 years or younger
- 2 primary/ bilateral breast cancers, both diagnosed under 50 years
- Triple negative breast cancer diagnosed under 60 years
- Breast cancer under 45 years and a first degree relative with breast cancer under 45 years
- Breast cancer AND Manchester Score of 15 or greater
- Ovarian cancer
- Male breast cancer
- Ashkenazi Jewish ancestry and breast cancer at any age
- For lobular breast cancer see section below
- Patient has Prostate cancer AND Manchester score of 15 or more

Patient is ***unaffected with cancer*** but all affected family members are deceased, AND Manchester Score of 20 or greater.

Manchester Scoring System

Cancer and age at diagnosis	Score
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

*Each side of family to be calculated **separately***

Pancreatic Cancer

Patient has:

- Pancreatic cancer AND Manchester score of 15 or more
- Patient diagnosed with pancreatic cancer younger than 60 years, plus:
  - 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

- Patient diagnosed with medullary thyroid carcinoma at any age
- Two or more cases of endocrine tumours in an individual or family
- Patient diagnosed with paraganglioma / phaeochromocytoma
- Patient has gastrointestinal stromal tumour (GIST) diagnosed under 50 OR at any age with a family history of GIST, phaeochromocytoma or paraganglioma
- Patient diagnosed with hyperparathyroidism under 35 years or parathyroid carcinoma or familial hyperparathyroidism at any age
- Patient diagnosed with pituitary tumour under 20 years or pituitary macroadenoma younger than 30 years

Colorectal and Endometrial Cancer / Lynch syndrome testing / Polyposis

Colorectal

- Patient diagnosed with bowel cancer younger than 40 years
- Patient diagnosed with bowel cancer and family reaches Amsterdam Criteria*
- Other diagnoses of colorectal and endometrial cancer should have tumour MSI / IHC to identify dMMR tumours. Please then refer dMMR tumours to genetics. If loss of MLH1 and PMS2, please first carry out MLH1 promoter hypermethylation studies and refer if not hypermethylated.
- Please review the current version of the test directory regarding testing guidelines for cancers that can be linked to Lynch syndrome** - R210 Inherited MMR deficiency (Lynch syndrome)
- Patient is ***unaffected with cancer*** but all affected family members are deceased AND they have a relative with bowel cancer diagnosed younger than 40 years OR the family history reaches Amsterdam Criteria*

** Amsterdam Criteria: ≥3 closely related family members with Lynch-related cancers over ≥2 generations with ≥1 case diagnosed <50 years*
***Lynch-related cancers comprise: Colorectal cancer, Endometrial cancer, Epithelial ovarian cancer, Ureteric cancer, Transitional cell cancer of renal pelvis, cholangiocarcinoma, Small bowel cancer, Glioblastoma, endocervical cancer, multiple sebaceous tumours*

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
- Unusual types of polyps such as juvenile, Peutz-Jeghers, hamartomatous polyps, or serrated polyps – {please refer to relevant sections in the test directory [R212, R213]}
- Features suggestive of Familial Adenomatous Polyposis [R414]

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 50 years
- Patient diagnosed with diffuse gastric cancer at any age and further family history of diffuse gastric cancer and/or lobular breast cancer under 70
- Patient diagnosed with lobular breast cancer and family history of diffuse gastric cancer (at least one case under 70)
- 2 cases of lobular breast under 50 years

Skin Cancer and associated syndromes

- Patient diagnosed with at least 2 melanomas and or melanoma in situ under 30 years
- Melanoma and /or melanoma in situ AND 2 or more relatives with melanoma in situ
- Patient diagnosed with melanoma with atypical moles and/or family history of melanoma or pancreatic cancer
- Patients with features suggestive of BAP1 associated tumours- BCC or atypical Spitz naevi
- Patients with features suggestive Gorlins - >5 BCC under 50

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, central nervous system tumour, leukaemia, adrenal cortical tumour or choroid plexus tumour
- Patient has features in keeping with Cowden syndrome (PTEN Harmatoma Tumour Syndrome)
- Patient diagnosed with other rare tumours: uveal melanoma, malignant mesothelioma, BAP1-associated tumours
- 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 30 years, or two or more schwannomas at any age, or a schwannoma with a close relative with schwannoma and note related tumours (meningiomas, neurofibromas)