



Cancer Genetics Service for Wales Referral Guidelines

For Individuals with a *Family History* of Cancer please refer:

Any individual with a relative who has a mutation in a cancer predisposition gene

Breast/ Ovarian Cancer

Please note an affected individual through a male (e.g. breast cancer in paternal relatives) may be assessed as a one step-closer relative e.g SDR as FDR. If in doubt please contact us or refer
(Must meet one or more of the following)

1 relative* with:

- Breast cancer age < 40 years
- Male breast cancer (any age)
- Bilateral breast cancer (any age)
- Breast AND ovarian cancer (any age)

***FDR**

2 relatives (male or female; same side of family)* with:

- Breast cancer (any age)
- Ovarian cancer (any age)
- One breast and one ovarian cancer in different relatives (any age)

***2 FDRs or 1 FDR and 1SDR**

3 or more relatives* (same side of family) with:

Breast and/or ovarian cancer at any age

***FDR or SDR**

Colorectal (CRC) / Bowel polyps / Lynch Syndrome Related Cancers (LSRCs)

LSRCs = CRC, endometrial, small bowel, transitional cell carcinoma renal pelvis or ureter

(Must meet one or more of the following)

At least 1 relative* with:

- LSRC age < 45 years
- CRC < 50 years
- 2 or more LSRC (any age)
- CRC and > 5 bowel polyps

***FDR**

2 relatives* with:

- CRC (any age)
- LSRC (one <50 years)

***2 FDRs (including both parents if CRC)
OR 1 FDR and 1 SDR (on same side of family)**

3 or more relatives* (same side of family) with:

- CRC (any age)
- LSRC (one <55 years)

Notes to accompany Referral Guidelines

Other features that may also suggest an underlying hereditary cancer predisposition are:

- Clustering (i.e. 2 or more) of the same cancer in first and second degree relatives (excluding those cancers more likely to have an environmental aetiology e.g. cervical and lung)
- Combinations of cancers occurring at younger than expected ages in multiple closely related family members.

In any families that do not meet the above referral criteria, but in whom you continue to have concerns, please contact the department to discuss further.

First degree relative (FDR) = parent, sibling, child

Second degree relative (SDR) = half sibling, grandparent, grandchild, aunt, uncle, niece, nephew

Death certificates can be obtained at www.gro.gov

If your patient does not meet the referral criteria, another family member may. Consider suggesting they seek their own referral.

If you would like to discuss an individual case please contact us using numbers below.

For up-to-date referral guidelines, please go to <http://www.wales.nhs.uk/sites3/page.cfm?orgid=525&pid=25833>

South East Wales
Cancer Genetics Service
Institute of Medical Genetics
University Hospital of Wales
Heath Park, Cardiff CF14 4XW
Tel: 029 2074 2577

South West Wales
Cancer Genetics Service
Singleton Hospital
Sketty, Swansea, SA2 8QA
Tel: 01792 285347

North Wales
Cancer Genetics Service
Block 12, Glan Clwyd Hospital
Sarn Lane, Rhyl
Denbighshire, LL18 5UJ
Tel: 01745 448788 ext 6181

Cancer Genetics Service for Wales Referral Guidelines

For Individuals with a *Personal History of Cancer* please refer:

Any Individual with a proven cancer predisposition gene mutation in the family

Breast/ Ovarian Cancer (must meet one or more of the following)

Individual with cancer:

- Breast cancer age < 40 years
- Male breast cancer (any age)
- Bilateral breast cancer (any age)
- Breast AND ovarian cancer (any age)

Individual with breast or ovarian at any age and 1 FDR relative* (male or female) with:

- Breast cancer (any age)
- Ovarian cancer (any age)

***SDR (if the intervening relative is male)**

Individual and 2 or more FDR or SDR relatives (same side of family) with:

Breast and/or ovarian cancer at any age

Colorectal (CRC) / Bowel Polyps/ Lynch Syndrome Related Cancers (LSRCs)
LSRCs=CRC, endometrial, small bowel, transitional cell carcinoma renal pelvis or ureter
 (Must meet one or more of the following)

Individual with:

- CRC < 50 years
- LSRC age <45 years
- 2 or more LSRC
- CRC and >5polyps
- >10 polyps
- Proven mismatch repair deficiency

Individual and 1 FDR relative with:

- CRC (any age)
- LSRC (one <50 years)

Individual and 2 or more relatives * (same side of family) with:

- CRC (any age)
- LSRC (one <55 years)

***FDR or SDR**

Notes to accompany Referral Guidelines

Other features that may also suggest an underlying hereditary cancer predisposition are:

- Clustering (i.e. 2 or more) of the same cancer in the individual and at least one first degree relative (excluding those cancers more likely to have an environmental aetiology e.g. cervical and lung)
- An individual with more than one primary cancer or a rare cancer (excluding those cancers more likely to have an environmental aetiology)
- Combinations of cancers occurring at younger than expected ages in multiple closely related family members.

These guidelines are not exhaustive. Any individuals that do not meet the above referral criteria, but in whom you continue to have concerns, please contact the department to discuss further.

First degree relative (FDR) = parent, sibling, child

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