

**Any Individual with a proven cancer predisposition gene variant 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**

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

*Death certificates can be obtained at [www.gro.gov](http://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, Rhyd  
Denbighshire, LL18 5UJ  
Tel: 01745 448788 ext 6181