OVERVIEW

1. Role of Familial Cancer Clinic (FCC) genetic counsellors
2. Psychosocial implications of familial cancer genetic counselling/testing
3. FCC referral guidelines
4. GP resources
5. Questions – and please ask any time 😊
ROLE OF FCC GENETIC COUNSELLORS

CANCER CAN BE HEREDITARY, FAMILIAL, OR SPORADIC

SPORADIC CANCER

FAMILIAL CANCER

HEREDITARY CANCER

Understanding which category your cancer falls into will help guide the management of your risk better.
ROLE OF FCC GENETIC COUNSELLORS

• GSWA now has 6 FTE FCC GCs

• Involved in all aspects of FCC patient management
  • Professional shift in recent years

• Triage of all new referrals
  • ~50 new referrals/week
  • Many require additional information before accurate assessment is possible

• Helpful:
  • Histopathology reports
  • Name, DOB and age at cancer dx of affected relatives
  • Details of relatives who have had genetic counselling +/- testing

• Not so helpful:
  • “Please arrange BRCA testing of Mary, she has a strong family history of cancer.”

• Clinic appointments generally reserved for those eligible for genetic testing
ROLE OF FCC GENETIC COUNSELLORS

• Types of patients GCs see at clinic
  • Diagnostic genetic testing
  • Predictive genetic testing
  • Results
  • Review

• Types of patients GCs don’t see at clinic
  • Clinical exam needed
  • Increased risk but unaffected
  • Population risk

• GCs work with CGs
  • Co-counselling (particularly paed onc)
  • Weekly intake meeting
  • Continued professional development
ROLE OF FCC GENETIC COUNSELLORS

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- GCs work with CGs
  - Co-counselling (particularly paed onc)
  - Weekly intake meeting
  - Continued professional development
  - Autographs…
ROLE OF FCC GENETIC COUNSELLORS

- Attend outreach clinics
  - Bunbury, Geraldton, Kalgoorlie, Rockingham
- Contribute to multidisciplinary team meetings
- Education of other healthcare professionals, students, patients, support groups
- Research
- Familial Cancer Registry
- Service development
- Management/leadership/supervision
PSYCHOSOCIAL IMPLICATIONS OF FAMILIAL CANCER GENETIC COUNSELLING/TESTING

Genetic counselling is a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions.

This process integrates the following:

• Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.

• Education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources and research.

• Counselling to promote informed choices in view of risk assessment, family goals, ethical and religious values.

• Support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

PSYCHOSOCIAL IMPLICATIONS OF FAMILIAL CANCER GENETIC COUNSELLING/TESTING
PSYCHOSOCIAL IMPLICATIONS OF FAMILIAL CANCER GENETIC COUNSELLING/TESTING

Information

Support
PSYCHOSOCIAL IMPLICATIONS OF FAMILIAL CANCER GENETIC COUNSELLING/TESTING

• Bit of a misnomer
  • Patient expectations
  • Non-attendance

• But, cancer genetic counselling/testing can have significant non-medical implications for the patient and their relatives
  • Revisiting of past cancer experience
  • Fear/anxiety re cancer development
  • Sense of identity
  • Isolation/belonging
  • Guilt/blame
  • Family dynamics/communication
  • Future reproduction
  • Empowerment
FCC REFERRAL GUIDELINES
Referral Guidelines

More guidelines will be available soon

Sub Categories

- Supervised

Protocols

- Cancer Genetics - Gastric Cancer Referral Guidelines
- General Practitioner Referral Guidelines for Cancer Genetics Assessment
- Referral Guidelines for Breast Cancer Risk Assessment and Consideration of Genetic Testing
- Referral Guidelines for Colorectal Cancer or Polyposis Risk Assessment and Consideration of Genetic Testing
- Referral Guidelines for Endocrine Cancer Risk Assessment and Consideration of Genetic Testing
- Referral Guidelines for Endometrial Cancer Risk Assessment and Consideration of Genetic Testing
- Referral Guidelines for Ovarian Cancer Risk Assessment and Consideration of Genetic Testing
- Referral Guidelines for Renal Cancer Risk Assessment and Consideration of Genetic Testing
FCC Referral Guidelines

Referral Guidelines

Sub categories

General Practitioner Referral Guidelines for Cancer Genetics Assessment

Referral Guidelines for Breast Cancer Risk Assessment and Consideration of Genetic Testing
Referral Guidelines for Colorectal Cancer or Polypoids Risk Assessment and Consideration of Genetic Testing
Referral Guidelines for Endocrine Cancer Risk Assessment and Consideration of Genetic Testing
Referral Guidelines for Endometrial Cancer Risk Assessment and Consideration of Genetic Testing
Referral Guidelines for Ovarian Cancer Risk Assessment and Consideration of Genetic Testing
Referral Guidelines for Renal Cancer Risk Assessment and Consideration of Genetic Testing
• Referral warranted
• But full assessment will determine whether the patient is eligible for appointment/genetic testing

<table>
<thead>
<tr>
<th>Cancer Type</th>
<th>Referral Guidelines and Resources</th>
</tr>
</thead>
<tbody>
<tr>
<td>Breast</td>
<td>Referral Guidelines for Breast Cancer Risk Assessment and Consideration of Genetic Testing</td>
</tr>
<tr>
<td></td>
<td>Breast cancer risk assessment tool (for unaffected women): FRABOC</td>
</tr>
<tr>
<td>Ovarian</td>
<td>Referral Guidelines for Ovarian Cancer Risk Assessment and Consideration of Genetic Testing</td>
</tr>
<tr>
<td>Endometrial</td>
<td>Referral Guidelines for Endometrial Cancer Risk Assessment and Consideration of Genetic Testing</td>
</tr>
<tr>
<td>Colorectal or polyposis</td>
<td>Referral Guidelines for Colorectal Cancer or Polyposis Risk Assessment and Consideration of Genetic Testing</td>
</tr>
<tr>
<td>Other</td>
<td>Referral Guidelines for Renal Cancer Risk Assessment and Consideration of Genetic Testing</td>
</tr>
<tr>
<td></td>
<td>Referral Guidelines for Endocrine Cancer Risk Assessment and Consideration of Genetic Testing</td>
</tr>
<tr>
<td></td>
<td>Referral Guidelines for Gastric Cancer Risk Assessment and Consideration of Genetic Testing</td>
</tr>
</tbody>
</table>
Cancer genetics resources for GPs

It is important that general practitioners (GPs) accurately assess a patient's personal and family history of cancer to identify those who need a referral to a family cancer clinic. Family cancer clinics will estimate an individual's cancer risk, and provide advice on risk-reduction strategies and the relevance of genetic testing.

**eviQ cancer genetics**

eviQ provides current, evidence-based cancer genetics information to assist GPs in the referral process, at the point of care. Free of charge, this includes information about genetic testing and risk management, as well as information for individuals and families with a specific gene mutation, syndrome or condition.

This information is developed by the eviQ Cancer Genetics Reference Committee, which involves clinical geneticists, medical oncologists, genetic counsellors and other health professionals in accordance with the eviQ Governance Framework.

The eviQ Cancer Genetics GP Referral Guidelines, and an outline of other cancer genetics information available on eviQ, is provided in this fact sheet.

**Purpose of the Cancer Genetics GP Referral Guidelines**

The purpose of the guidelines is to define at-risk groups requiring referral to a family cancer clinic that offers genetic counselling and genetic testing, if appropriate.

If needed, there are two stages to genetic testing:

- **Mutation search.** This usually involves testing a blood sample from an affected family member in the first instance. The test determines whether a gene mutation that causes the increased risk of cancer for that family can be identified.
- **Predictive test.** This is only available to family members when a mutation has already been found in a mutation search. This test determines whether or not the patient actually has the family gene mutation.

Access to eviQ is free at www.eviQ.org.au

Username: phc | Password: phc

Working together to lessen the impact of cancer
# FCC Referral Guidelines

**Genetic Services of Western Australia**

**Familial Cancer Program**
King Edward Memorial Hospital for Women
Agnes Vahal Unit
34 Bagot Road, SUBIACO WA 6008
Telephone: 08 9249 9000 (8am - 5pm weekdays)
Fax: 08 9249 9132
Email: fg20@health.wa.gov.au

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## Referral Form

For breast/ovarian cancer, we accept referrals for people at increased risk (high and moderate risk groups). To assess your patient’s risk, please see:

Further risk assessment & referral information is available from our website: http://www.cancerwa.gov.au/services/geneinfo

### Patient Details

<table>
<thead>
<tr>
<th>Patient name</th>
<th>UPHN</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Address</th>
<th>Date of birth</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Suburb</th>
<th>Postcode</th>
<th>Telephone</th>
</tr>
</thead>
</table>

### URGENT / CHEMOTHERAPY / LIMITED LIFE EXPECTANCY / OTHER reason for urgency:

- [ ] URGENT
- [ ] CHEMOTHERAPY
- [ ] LIMITED LIFE EXPECTANCY
- [ ] OTHER reason for urgency:

- [ ] Yes / [ ] No

### This person has had:
- [ ] Breast cancer
- [ ] Ovarian cancer
- [ ] Bowel cancer
- [ ] Bowel polyps

### Other cancer type:

- [ ] Other cancer type:

**PLEASE ATTACH ALL MORTUARY REPORTS**

### Treatment

<table>
<thead>
<tr>
<th>Treatment</th>
<th>Start date</th>
<th>End date</th>
</tr>
</thead>
</table>
- [ ] Chemotherapy
- [ ] Radiotherapy
- [ ] Surgery

### Family Information

- [ ] Yes / [ ] No

<table>
<thead>
<tr>
<th>Full name</th>
<th>Genetic Service centre</th>
<th>City/State</th>
</tr>
</thead>
</table>

| Genetic test results (if any) | | |

| There is a family history of: | | |
- [ ] Breast cancer
- [ ] Ovarian cancer
- [ ] Bowel cancer
- [ ] Bowel polyps

### Additional referral information:

- [ ] Additional referral information:

### Referring Doctor Details:

<table>
<thead>
<tr>
<th>Name</th>
<th>Practice name</th>
<th>Practice address</th>
<th>Suburb/Postcode</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Contact Phone/Fax</th>
</tr>
</thead>
</table>
FCC REFERRAL GUIDELINES

GENETIC SERVICES OF WESTERN AUSTRALIA

RECOMMENDED CANCER PROGRAMS
King Edward Memorial Hospital for Women
AGCAT (WA) Inc.
45 Bagot Road, SUBIACO WA 6008
Telephone: 6227 2999; 6240 1859 (Fax: 6240 1858)
Facsimile: 6240 1933
Email: fcc@health.wa.gov.au

REFERRAL FORM

For breast/ovarian cancer, we accept referrals for people at increased risk (high and moderate risk groups). To assess your patient’s risk, please see:

Further risk assessment & referral information is available from our website:

Patient Details

Patient name: [ ]
URN: [ ]
Address: [ ]
Date of birth: [ ]
Suburb: [ ]
Postcode: [ ]
Telephone: [ ]

URGENT CHEMOTHERAPY LIMITED LIFE EXPECTANCY OTHER reason for urgency: [ ]

Does this person have any Jewish ancestry? [ ] Yes / [ ] No
This person has had: [ ] Breast cancer [ ] Ovarian cancer [ ] Bowel cancer [ ] Bowel polyps
Other cancer types: [ ]

"PLEASE ATTACH ALL METASTASIS REPORTS (and all colonoscopy reports for bowel cancer/polyp referrals)

TREATMENT

Chemotherapy: [ ] Start date: [ ]
Radiotherapy: [ ] End date: [ ]
Surgery: [ ]

Family Information

Has anyone in the family had genetic testing or attended a genetics clinic in Australia or overseas? [ ] Yes / [ ] No
Full name: [ ]
Genetics Service name: [ ]
City/State: [ ]
Gene tested: [ ]

There is a family history of: [ ] Breast cancer [ ] Ovarian cancer [ ] Bowel cancer [ ] Bowel polyps
Other cancer type: [ ]

Additional referral information:

Referral Doctor Details

Name: [ ]
Practice name: [ ]
Practice address: [ ]
Suburb/Postcode: [ ]
Contact Phone/Fax: [ ]

If yes for any of these questions, please complete details in the table (page over)
GP RESOURCES

- Cancer Australia
  - [https://canceraustralia.gov.au/](https://canceraustralia.gov.au/)
- FRA-BOC

What FRA-BOC assesses

Assessing a family history of cancer with FRA-BOC

FRA-BOC uses a maximum of eight questions to provide an estimation of risk.

It captures information about a woman’s 1st (parents, siblings, children) and 2nd (aunts, uncles, nieces, nephews, grandparents) relatives on both sides of the family. However, FRA-BOC assesses family history of breast and ovarian cancer on each side of the family separately.

FRA-BOC begins the assessment with questions about family history of invasive epithelial ovarian cancers.

As ovarian cancer is less common than breast cancer in the population, a family history of ovarian cancer can sometimes be a stronger predictor of risk than family history of breast cancer. Where there is no history of ovarian cancer, a series of questions are then asked about breast cancer family history.

FRA-BOC asks about Ashkenazi Jewish ancestry.

Although genetic factors that may influence the risk of developing breast cancer are found in people of all nationalities, some are more common in people of Ashkenazi Jewish ancestry (Jews whose origins can be traced back to Eastern Europe) than in members of the general population. In Australia, most Jewish families are of Ashkenazi ancestry, so FRA-BOC simply uses the term ‘Jewish ancestry’. As many as one in 40 individuals (men and women) of Ashkenazi Jewish descent has one of the three founder mutations (traits) in the breast/ovarian cancer susceptibility genes BRCA1 and BRCA2. Women who carry such a gene fault have a high lifetime risk of breast cancer, estimated to be in the range of 20-66%, and a lifetime ovarian cancer risk of about 20%.
GP RESOURCES

- Cancer Australia
- FRA-BOC
- Advice about familial aspects of breast cancer and ovarian cancer

Assessing family history

Family history of breast or ovarian cancer can be used to estimate:
- a woman’s risk of developing these cancers
- the probability of having an inherited mutation in a known cancer-predisposing gene

Key factors associated with increased risk include:
- multiple relatives affected by breast (male or female) or ovarian cancer
- young age at cancer diagnosis in relatives
- relatives affected by both breast and ovarian cancer
- relatives affected with bilateral breast cancer
- Ashkenazi Jewish ancestry

Taking a family history

Consider relatives on each side of the family separately:
- asking the woman about any primary cancer in all 1° relatives (parents, siblings, children) and 2° relatives (aunts, uncles, nieces, nephews, grandparents) on both sides of the family
- establishing the site and age at diagnosis of the cancer(s)
- confirming, if possible, reports of cancer in relatives — a person’s knowledge of their family history may be inaccurate
- updating the family history regularly — it may change with time.

Which genes are associated with a predisposition to breast or ovarian cancer?

Violeuve both with a mutation in one of several known genes (see Table 2) have an increased risk of breast and/or ovarian cancer. There may be other genes, as yet undiagnosed, in which mutations are also associated with a risk of breast or ovarian cancer. The women most likely to have inherited a mutation are those with a strong family history of breast or ovarian cancer.

Breast cancer

One in 8 Australian women develops breast cancer before the age of 85. Breast cancer is the second most common cause of cancer death in Australian women.

Ovarian cancer

One in 87 Australian women develops ovarian cancer before the age of 85. Ovarian cancer is the leading cause of death from gynecological malignancy.

RD. The average life expectancy of Australian women is 80 years. For key statistics about breast and ovarian cancer see www.canceraustralia.gov.au

Family cancer clinics

Family cancer clinics provide a service for people with a family history of cancer and their health professionals. The service is offered to any family members, whether or not they have been diagnosed with cancer. After assessing detailed information about a woman’s family history of cancer, these clinics provide genetic counselling including:
- information about a person’s risk of developing cancer based on family history and other relevant factors
- advice about strategies that reduce the risk of cancer
- information about early detection of cancer
- an estimate of the likelihood of carrying an inherited mutation in a cancer-predisposing gene
- if appropriate, the offer of genetic testing.

Genetic testing

It is possible to detect mutations in some cancer-predisposing genes. Some mutations may not be detected using current technology. Testing involves first searching for a gene mutation, usually in a blood sample from an affected family member. Should a mutation be found, testing may then be offered to other adult relatives who may carry the same mutation. Genetic testing is offered only with pre-and post-test counselling to discuss the limitations, potential benefits, and possible consequences.

For a list of family cancer clinics see canceraustralia.gov.au/familial

Table 1: Approximate risk of developing breast or ovarian cancer in the next 10 years.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Mutation Frequency</th>
<th>Major sites of risk</th>
<th>Risk to age 75 in mutation carriers</th>
<th>Other possible sites with up to 10% lifetime risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>BRCA1</td>
<td>1/3000*</td>
<td>Breast, Ovary</td>
<td>50% - 70%, 10% - 15%</td>
<td>Breast, colon, prostate, pancreas, liver, stomach, bone, breast, ovary, uterus, testes, skin, brain, kidney, salivary gland, T-cell lymphoma, synovial sarcoma, synchronous bilateral breast cancers</td>
</tr>
<tr>
<td>BRCA2</td>
<td>1/1500*</td>
<td>Breast, Ovary</td>
<td>40% - 60%, 10% - 20%</td>
<td>Male breast, prostate, pancreas, liver, stomach, bone, breast, ovary, uterus, testes, skin, lymphoma, leukemia, other</td>
</tr>
<tr>
<td>TP53</td>
<td>1/100,000</td>
<td>Breast, Bone or soft tissue</td>
<td>50%, 10% - 15%</td>
<td>Skin, breast, adrenal gland, hematological, and other</td>
</tr>
<tr>
<td>NUTmid repair genes (BRIP1)</td>
<td>1/3000</td>
<td>Large bowel, ovaries, liver</td>
<td>50%, 40% - 60%</td>
<td>Ovary, other gastro-intestinal, renal tract</td>
</tr>
</tbody>
</table>

* <1/1000 is a rare cause of ovarian cancer. **BRCA1 and BRCA2 are the most common causes of inherited breast and ovarian cancer. |
GP RESOURCES

- Cancer Australia
- FRA-BOC
  - Advice about familial aspects of breast cancer and ovarian cancer
- Centre for Genetics Education
  - Fact sheets
GENETIC AND GENOMIC TESTING

CHROMOSOME MICROARRAY (CMA) TESTING IN CHILDREN & ADULTS
Testing Guide: Chromosome Microarray (CMA) – Children and Adults

FORENSIC, PATERNITY AND ANCESTRY DNA TESTING

WHEN PARENTS ARE RELATED – CONSANGUINITY

ETHICAL ISSUES IN HUMAN GENETICS AND GENOMICS

LIFE INSURANCE PRODUCTS AND GENETIC TESTING IN AUSTRALIA

PHARMACOGENETICS / PHARMACOGENOMICS

CLONING AND STEM CELLS

GENE THERAPY

PRENATAL TESTING OVERVIEW
Booklet: Prenatal Testing: Special tests for your baby during pregnancy

SCREENING TESTS DURING PREGNANCY
Brochure: Screening Tests for Your Baby in Early Pregnancy

DIAGNOSTIC TESTS DURING PREGNANCY

NON-INVASIVE PREGNATAL TESTING (NIPT)

CHROMOSOME MICROARRAY (CMA) TESTING DURING PREGNANCY

PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

FOLATE BEFORE AND DURING EARLY PREGNANCY

CANCER AND GENETICS OVERVIEW

BREAST AND OVARIAN CANCER AND INHERITED PREDISPOSITION

BOWEL CANCER AND INHERITED PREDISPOSITION

GENETICS AND MELANOMA

GENETICS AND PROSTATE CANCER

TRISOMY 21 - DOWN SYNDROME

TRISOMY 13 - PATAU SYNDROME

TRISOMY 18 - EDWARDS SYNDROME

KLINEFELTER SYNDROME - XY SYNDROME

TURNER SYNDROME - XO SYNDROME

CYSTIC FIBROSIS

TAY-SACHS DISEASE AND OTHER CONDITIONS MORE COMMON IN THE ASHKENAZI JEWISH COMMUNITY

THALASSAEMIA

SICKLE CELL DISEASE

NEUROFIBROMATOSIS TYPE 1

AUTISM SPECTRUM DISORDERS

HEREDITARY HAEMOCHROMATOSIS

DIABETES TYPES 1 AND 2 AND INHERITED PREDISPOSITION

BLOOD CLOTTING CONDITIONS (HEREDITARY THROMBOPIALIAS)

ALZHEIMER DISEASE

PARKINSON DISEASE

HAEMOPHILIA

DUCHENNE AND BECKER TYPES OF MUSCULAR DYSTROPHY

FRAGILE X SYNDROME

HUNTINGTON DISEASE
Booklet - Huntington Disease and Genetic Testing

FAMILIAL HYPERCHOLESTEROLAEMIA

CARDIOMYOPATHIES

PRIMARY ARRYTHMOGENIC DISORDERS
GP RESOURCES

• Cancer Australia
  • https://canceraustralia.gov.au/
  • FRA-BOC
  • Advice about familial aspects of breast cancer and ovarian cancer

• Centre for Genetics Education
  • https://canceraustralia.gov.au/
  • Fact sheets

• WOMEN Centre
THANK YOU 😊

• Questions?
• sarah.osullivan@health.wa.gov.au
• sarah@womencentre.com.au