Familial Cancer Syndromes

Disclaimer

Contents

Background

About familial cancer syndromes

- *Most cancer is not due to an inherited mutation e.g., in 2017 only 5% of all breast cancer and 5% of all colon cancer is attributable to identified inherited mutations.*
- *When searching for a mutation it is best to start in an affected family member.*
- *Predictive testing is only possible in families where the mutation has been identified as there are hundreds to thousands of different mutations in each gene.*
- *Publicly funded genetic testing is only available through selected Victorian genetic services. Medicare rebates now apply to women affected by breast or ovarian cancer who meet strict criteria when ordered by a specialist.*
- *Patients with a potentially high risk of a familial cancer syndrome usually have:*
  - *multiple relatives on the same side of the family with cancers of the same type or related type.*
  - *cancers occurring at a younger age.*
  - *≥ 2 primary cancers of the same or different type.*

Assessment

Practice Point

Family history is most important
In general practice, the most important initial investigation is obtaining the family history. Genetic testing is arranged, if appropriate, only after genetic counselling and referral to a genetics service.

1. Clarify whether the patient has any current symptoms. If so, manage these symptoms appropriately before fully assessing the family history.

2. Take a ***family history*** and sketch a ***3-generation family tree.*** Understand ***cultural considerations*** for Aboriginal and Torres Strait Islander patients.

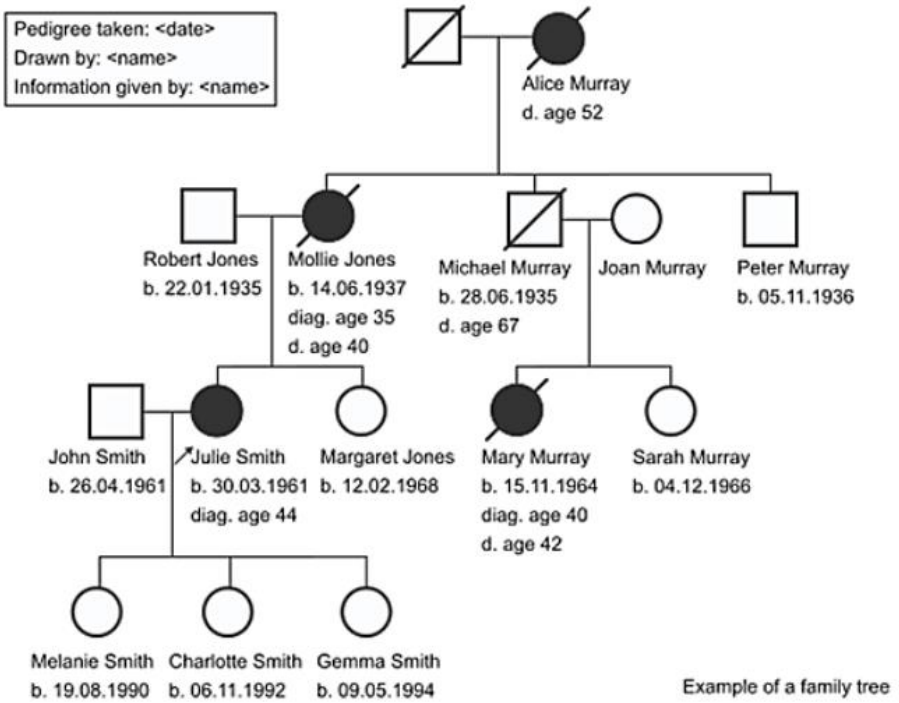
   - ***Family history***
     Note in particular:
     - *Type of primary cancers present in the family*
     - *Number of relatives affected*
     - *Age of onset in the family members*
     - *Multiple primary cancers*
     - *Any known mutation carrier*
     - *Ancestral background (particularly of Jewish decent)*

- **3-*generation* family tree**
  - Genetic pedigrees are drawn using standardised symbols:

	Male	Female	Sex Unknown
Individual			
Affected individual (symbol coloured in)			
Multiple individuals	5	5	5
Deceased			
Pregnancy	P	P	P
Miscarriage	male	female	
Person providing pedigree information			

Marriage/partnership	
Divorce/separation	
Where the partners are blood relatives (consanguineous relationship)	
Children/siblings	sibship line, line of descent, individual line
Identical twins (monozygotic)	
Non-identical twins (dizygotic)	

  - *See the following example of a family tree:*



Example of a family tree

Both reproduced with permission from Health Education England's Genomics Education Programme – [Taking and Drawing a Family History]

- *Start with the patient in the centre (note the arrow)*
- *Include first degree relatives (children, siblings, parents), and second-degree relatives (aunts, uncles, grandparents) on both sides of the family.*
- *Include age at diagnosis, other conditions as well as presenting condition, miscarriages, stillbirths, infertility and birth defects.*

- ***Cultural Considerations***
  - *The concept of family is broader that being genetically related for many Aboriginal and Torres Strait Islander patients.*
  - *Be sensitive when talking about cultural taboos such as Sorry Business (people who have died), Men's Business, and Women's Business, and references to members of the Stolen Generation.*

3. Determine any **specific risk factors** for uncommon familial cancer.

   Specific risk factors
   - *Blood relative of a known carrier of a cancer predisposing mutation*
   - *Three 1st or 2nd degree relatives with same or related cancers*
   - *Two 1st or 2nd degree relatives with the same or related cancers, one being diagnosed aged < 50 years*
   - *Rare tumour diagnosed aged < 45 years e.g:*
     - *Adrenal cancer*
     - *Choroid plexus carcinoma*
     - *Glioblastoma*
     - *Paraganglioma*
     - *Phaeochromocytoma*
     - *Retinoblastoma*
     - *Sarcoma*
   - *Medullary thyroid cancer at any age*
   - *Ashkenazi Jewish ancestry*
   - *Diffuse gastric cancer with any of:*
     - *Age < 40 years*
     - *Personal or first-degree relative with lobular breast cancer (one with diagnosis aged < 50 years)*

Management

1. If the patient is terminally ill, seek genetic advice for consideration of DNA banking.
2. Manage according to the type of cancer:
   - Familial breast or ovarian cancer syndromes
   - Familial colorectal or endometrial cancer syndromes
3. Request genetic assessment if a potentially high risk of familial cancer, for example:
   - *specific risk factors* for uncommon familial cancer syndromes.
   - cancers occurring at a younger age.
4. Advise:
   - that genetic testing may be available if genetic counselling assessment recommends it is appropriate.
   - any preventive strategies e.g., healthy diet, healthy lifestyle, screening programmes.

Referral

- If the patient is terminally ill, seek genetic advice for consideration of DNA banking.
- Request genetic assessment if:
  - any ***specific risk factors*** for uncommon familial cancer syndromes.
  - there is a potentially high risk of familial cancer.

In all referrals, include a copy of the 3-generation family tree.

Information

For health professionals

- Cancer Council – Optimal Care Pathways for Aboriginal and Torres Strait Islander People with Cancer
- Centre for Genetics Education – Cancer Genetics Overview
- eviQ – Cancer Genetics: Adult
- Royal Australian College of General Practitioners (RACGP) – Genomics in General Practice

For patients

- Cancer Australia
- Cancer Council – Checking for Cancer: What to Expect
- Cancer Council Victoria – Aboriginal Communities: Information
- Centre for Genetics Education:
    - Cancer in the Family
    - Your Family Health History
- Genetic Alliance – A Guide to Family Health History
- National Indigenous Cancer Network – About Cancer

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