GENETICS AND PALLIATIVE CARE

Play, Eat, Love - Palliative Care Conference
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OVERVIEW

- What do Clinical Genetics Services offer?
- When is it appropriate to refer?
- Which patients are offered genetic testing?
- Case examples from neurology, cardiac and cancer
- Genetic Counselling in palliative care patients
  - What does this involve?
  - Referrals, Logistics and Timing

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CLINICAL GENETICS SERVICES

Patients referred to Genetics:

- All stages of life
- All kinds of medical conditions
- Assess the likelihood a condition is genetic:
  - Review personal medical history
  - Clinical examination
  - Collect family Hx information,
- Discuss the availability and utility of genetic testing
- Referrals to specialist services
- Provide general risk advice for relatives

Regional Genetics clinics - Ballarat:

Familial Cancer Clinic (FCC): Austin Health

General Genetics: Victorian Clinical Genetics Service (VCGS)

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WHO AND WHEN TO REFER?

Who to refer:

- A patient affected by a condition with suspected genetic aetiology - based on the condition or the family history

When to refer:

- If the patient is interested – as early as possible – to enable an outpatient appointment or ward visit (metropolitan only)
- If the family is interested – as early as possible – even if the timing is not suitable immediately, we can liaise with the family and the referrer regarding timing of next steps

In the meantime:

- Consider DNA storage – particularly if the patient is deteriorating

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WHO IS OFFERED GENETIC TESTING?

Genetic testing is most informative when performed in an affected person, not an unaffected relative

The likelihood of a genetic test being offered is associated with the likelihood that a genetic mutation will be identified. If this is considered "high" (generally >10%) then genetic testing is likely to be offered. The chance of identifying a VOUS is 5%. A negative genetic test result does not exclude a genetic aetiology

Not all patients will be offered genetic testing. Not all genetic testing is funded. Patients/families may elect to self-fund genetic testing.

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WHO IS OFFERED GENETIC TESTING?

Assessment based on:

- Phenotyping – previous investigations, natural history of condition, clinical examination
- Family history, inheritance pattern, penetrance
- Mutation detection rate for condition – how much could other factors have contributed?
- Impact of +ve genetic result – for patient and their family

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CASE EXAMPLE 1

- 69yo man referred urgently for genetic assessment
- Dx 65 MND/FTD, rapidly deteriorating, inpatient on ward
- No fam Hx but wife wanted to clarify genetic risk for their adult children. DNA stored
- Geneticist visited on the ward for examination. Genetic testing was not indicated as low chance of detecting a mutation, however wife (NOK) elected to fund panel testing
- No mutation identified

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CASE EXAMPLE 2

- 55yo healthy man with a fam hx of pancreatic cancer:
  - mother dx 76y (d.77)
  - maternal uncle dx 54 (d.54)
  - No further fam hx cancer known/reported
- Based on this information the family did not meet criteria for a funded genetic test. We raised the possibility of storing a DNA sample on his mother before she passed
- Later on, the family elected to self-fund genetic testing in the stored DNA sample – a *BRCA2* mutation was identified
- Funded PT was available to the family

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CASE EXAMPLE 3

- 28yo man died unexpectedly while exercising. Post mortem suggestive of ARVC and other differentials. Coroner stored a DNA sample from spleen
- Family referred to genetics
- Family consented to genetic testing for inherited cardiac conditions in deceased, including genes associated with ARVC
- Mutation in *DSG2* identified – ARVC confirmed
- PT offered to family members

Nb: sometimes stored DNA samples are of insufficient quality

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REFERRALS, LOGISTICS AND TIMING

- Possibility of genetic aetiology raised to/by the patient/family
- Arrange DNA storage in the affected patient:
  - 20ml EDTA blood
  - send to Austin Molecular Pathology lab for DNA storage only
  - Collect consent for DNA storage: from patient/NOK
- Offer patient/family referral to Clinical Genetics
  - Include details of the appropriate contact person in lieu of the patient – usually a family member or the NOK
  - No genetic testing will be performed without involvement of the patient/NOK AND a Clinical Genetics Service
- Any questions – Call the Clinical Genetics Service and discuss your case with the on call Genetic Counsellor

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