Central Adelaide Local Health Network: Adult Genetics Unit referral form - CLINICAL GENETICS

REFERRAL DATE _____ FAX to 08-8429 6112 or EMAIL to adultgenetics@sa.gov.au | office use ony: received

Referral to: ◯ **Dr Nicola Poplawski** ◯ **Dr Kathryn Waddell-Smith** (cardiac only) ◯ **Dr Sunita De Sousa** (endocrine only)

Patient information

Name	**DOB** **Sex** **F** ◯ **M** ◯ **other** ◯
Address	**phone** **mobile**
Suburb **postcode**	**Interpreter required** **Y** ◯ **N** ◯
Hospital record number	**Language**

Referrer information

Name	**Address/Hospital**
Specialty/role	**Suburb** **Postcode**
provider number	
responsible consultant	**signature**

Alternative contact for patient

Name	**Phone**
Address	**relationship to patient referred**
Suburb **Postcode**	**To attend appointment?** Y ◯ N ◯

Primary Clinical Question or reason for referral

☐ **Request for genetic counselling - known pathogenic variant (mutation) in the family** (include copy of report if available)

☐ diagnostic testing (patient symptomatic) ☐ predictive testing (patient asymptomatic) ☐ carrier testing

gene and variant _____

Name of relative with variant _____ relationship to patient referred _____

☐ **Request for genetic counselling - known genetic condition in the family** (no/unsure if genetic result available)

☐ diagnostic testing (patient symptomatic) ☐ predictive testing (patient asymptomatic) ☐ carrier testing

genetic condition _____

Name of relative with condition _____ relationship to patient referred _____

Please note; if causative variant is not known, usually the best person to refer is an affected relative in whom diagnostic genetic testing can be done

☐ **Request for genetic counselling for patient and family - has abnormal genetic test result** (include copy of report)

gene and variant _____

☐ **Request for diagnostic genetic testing - has suspected genetic disorder** (detail personal and family history below)

suspected genetic condition _____

Please note, the Adult Genetics Unit does not accept referrals for hypermobility type EDS (refer to appropriate management services), familial hypercholesterolaemia (refer to cardiologist/lipid specialist) or MTHFR polymorphisms (see www.racgp.org.au for information about MTHFR testing)

☐ **Other reason for Clinical Genetics assessment** (detail personal and family history below)

Specific question for clinical genetics _____

If the diagnosis is uncertain consider referral to other specialists for assessment and completion of phenotyping, prior to genetic referral

Personal medical history complete for all referred patients; please include copies of relevant key results

Relevant family history complete for all referrals; include relationship to referred patient, diagnosis and age at diagnosis

DNA storage

- **Is the patient palliative** **Y** ◯ **N** ◯ if yes please refer to DNA storage instructions on reverse
- **has DNA been stored** **Y** ◯ **N** ◯

Central Adelaide Local Health Network
Adult Genetics Unit
Specialty Medicine 2
Royal Adelaide Hospital

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Adelaide SA 5000

Tel (08) 7074 2697
Fax (08) 8429 6112
adultgenetics@sa.gov.au

Storing DNA from patients with a suspected genetic disorder

Genetic testing is now a common part of standard medical care for a range of disorders. However, sometimes genetic testing is not yet possible or it is not the right time to arrange genetic testing. It is possible to store DNA so that genetic testing can occur at a later time.

Situations where DNA storage should be considered include;

- A patient has a (suspected) genetic condition but their personal or family history needs to be clarified to determine if genetic testing is indicated.
- A patient has a (suspected) genetic condition but their personal or family history needs to be clarified to determine which gene or genes should be tested.
- A patient has a (suspected) genetic condition but the gene/s associated with the condition has/have not been identified (but may be in the future).
- A patient has a (suspected) genetic condition but the timing is not right for them to consider testing at the present time (e.g. they may be stressed, depressed, or receiving palliative care and likely to die soon).
- A patient has a (suspected) genetic condition, does not want to know their own genetic status but does want their relatives to be able to access genetic information after their death.

WHY bank DNA samples?

Testing may identify a genetic error (mutation) which can then be used to clarify the health risks for relatives, and/or to identify preventative strategies for relatives who have inherited that mutation, and/or to provide information and choices to relatives planning to have children.

- Lymphocyte (blood) DNA from an affected family member is usually the most useful sample for genetic studies in a family with a suspected genetic disorder.
- With appropriate consent, a DNA sample can be used before or after a patient's death.
- Giving a blood sample for future genetic studies is a practical way for a dying patient to provide their family with better medical care.

HOW to bank a DNA sample

Collect 2 x 4ml of blood in EDTA; request form should ask for “DNA storage”

Include a brief note with the sample giving the personal and family history.

Send to **Molecular Pathology, SA Pathology at Frome Road**

If a relative is seeking genetic counselling, refer them to the Adult Genetics Unit.

Call the Adult Genetics Unit at the Royal Adelaide Hospital on 7074 2697 if you have questions.

The DNA sample will be stored, but testing will **not** be done without appropriate genetic counselling and written consent from the patient or their next of kin or their legal representative.