

Service Overview

- This clinic is open to referrals from General Practitioners (GP) and Specialists.
- For patients with identified or probable inherited familial hypercholesterolemia (FH), this Clinic provides the (public) tertiary facility to have expert cardiac genetic advice and investigation.
- Cascade clinical screening of first degree relatives of confirmed proband (including current and planned offspring).
- Entry into genetics clinic database (with consent).
- Ongoing Cardiology clinical care is **not** provided by this service; this will be in the hands of the referring GP/Specialist. (maximum of three clinical visits - the first to assess if testing is of value, and the following to feedback the results and their meaning to the patient, including implications for first degree relatives).
- The clinic will liaise with a patient's referring Physician and GP following initial consultation.

Inclusion Criteria

The patient has:

- Dutch Lipid Clinic Network Score (DLCNS) of ≥ 3 OR
- Highest fasting untreated LDL cholesterol ≥ 5 or highest total untreated cholesterol of ≥ 7 PLUS ANY OF
- Premature coronary/cerebral artery or peripheral vascular disease <60 years old (in self or 1st degree relative)
- Tendon xanthoma and/or arcus cornealis (in self or 1st degree relative)
- Useful link: <https://bit.ly/2ACSrbU>

Exclusion Criteria

- The patient has already been seen by a SA Clinical Genetics Service (unless justification for referral is provided)
- Secondary causes of hypercholesterolaemia have not been excluded (i.e. primary hypothyroidism, proteinuria, cholestasis and medications such as corticosteroids)

Investigations Required *(please provide results with referral)*

Cardiac assessment including:

- Full medical history, including family history
- Examination
- Lipid profile (ideally fasting), where appropriate
- Blood work where appropriate, to exclude diabetes hypothyroidism, hepatic and renal disease

Fax referrals to *(attn. Dr Kathryn Waddell-Smith)*

Flinders Cardiac Clinic Fax (08) 8177 1589

Suggested GP Management

- Manage cardiovascular risk factors aggressively
- Exclude secondary causes of hypercholesterolaemia
- Consider provision of lifestyle advice (i.e. cessation of smoking, weight loss, diet, exercise and alcohol intake)

Clinical Resources

- The diagnosis of Familial Hypercholesterolaemia <https://www.athero.org.au/fh/health-professionals/how-to-diagnose-fh/> <https://www.racgp.org.au/afp/2012/december/familial-hypercholesterolaemia/>
- The Familial Hypercholesterolaemia Australasia Network: <https://www.athero.org.au/fh/>

General Information to assist with referrals is available on the SALHN Outpatient Services website www.sahealth.sa.gov.au/SALHNoutpatients

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