Clinical Genetics and Genomics Service Referral Guidelines

AlfredHealth

Medical practitioners are encouraged to refer eligible patients to the Alfred Hospital Clinical Genetics and Genomics Service (CGGS) for evaluation of suspected genetic or inherited predispositions. These guidelines aim to streamline the referral process and ensure comprehensive assessment and advice.

To refer a patient, please submit a referral electronically through one of the following mechanisms:

- Direct to CGGS at genetics@alfred.org.au or via **fax to (03) 9076 8546.**
- Via Alfred Health Outpatient referral in team op.referrals@alfred.org.au or via fax (03) 9076 6938.

Please include in the referral:

Demographic details: • Date of birth • Patient's contact details including mobile phone number • Referring GP details • If an interpreter is required • Medicare number	**Clinical information:** • Reason for referral • Relevant investigations and correspondence to facilitate a thorough evaluation • Where a pathogenic variant is known to be present in a family, please provide relation to the family member and service where family member tested

Exclusion criteria

The following services are not offered by Alfred Hospital Clinical Genetics and Genomics Service (CGGS):

- Children or adolescents <18yrs of age (Please refer to paediatric genetic services)
- Individuals with a personal or family history of a health condition where a monogenic cause has not been established
- Individuals with a personal or family history of a health condition explained by confirmed/likely somatically acquired pathogenic variants
- Individuals who are pregnant or require prenatal genetic counselling and care
- Individuals seeking or considering 'direct to consumer' genetic testing, pharmacogenetic or paternity testing
- Common genetic conditions (e.g., hemochromatosis, MTFHR mutation carriers or Factor V Leiden Thrombophilia) not within the scope of CGGS
- Referrals for individuals with Ehlers-Danlos syndrome, type 3 / hypermobility / joint laxity will not be accepted unless there are additional and proven clinical features suggestive of a specific connective tissue disorder. Additional features may include; hernias, spontaneous internal organ rupture/collapse, aortic/arterial aneurysm or dissection, arterial tortuosity, cleft palate, craniosynostosis, ectopia lentis, easy fractures, dysmorphic features that may indicate conditions such as Marfan syndrome, vascular EDS, Loeys Dietz syndrome, TAAD, or arterial tortuosity syndrome. Please see further information below.

Referral Indications

Referral to the CGGS may be indicated for individuals with a relevant personal and/or family history of conditions including, but not limited to:

- Familial cancer syndromes (e.g., hereditary breast and ovarian cancer, Lynch syndrome)
- Neurological and muscle disorders (e.g., Huntington's disease, muscular dystrophy, epilepsies)
- Cardiac conditions with suspected genetic basis (e.g., long QT syndrome, cardiomyopathies)
- Familial aortopathies and connective tissue disorders (e.g., Marfan's syndrome, Loeys-Dietz syndrome)
- Renal conditions suggestion of genetic aetiology (e.g., polycystic kidney disease)
- Immune or autoinflammatory disorders (e.g. Familial Mediterranean fever, primary immunodeficiencies)
- Inherited pulmonary conditions (e.g., Cystic Fibrosis)
- Intellectual disability with suspected genetic cause
- Dysmorphic features or congenital anomalies suggestive of a genetic syndrome
- Patients with an established genetic or chromosomal diagnosis requiring further evaluation
- Predictive testing for individuals with a known pathogenic variant in a family member

For a more detailed explanation please see RACGP Genomics in general practice

Please note there are DH statewide referral criteria for advice on inherited breast cancer (high risk patients)

Inquiries about referral criteria or specific cases, please contact CGGS on phone number (03) 9076 8554 and ask to speak to the genetic counsellor on duty.

Hypermobile Ehlers-Danlos Syndrome

Joint hypermobility is common in the general population and often familial. The diagnosis of hypermobile Ehlers-Danlos syndrome (hEDS) is made based on clinical features, as the genetic basis is not yet understood. Because of this, we do not offer genetic testing to patients with hEDS. A clinical diagnosis can be made by a GP or other medical specialists involved in the patients care and does not require attending a genetics service for this purpose. The 2017 International Diagnostic Criteria provides guidance regarding making a clinical diagnosis. The checklist can be found at The Ehlers-Danlos Society website (https://www.ehlers-danlos.com/heds-diagnostic-checklist/).

In general, treatment for hEDS should support patients to manage and alleviate symptoms. Patients may benefit from specific medical and allied health services such as, physiotherapy, psychology. Low impact exercise is advisable. Management of symptoms should be through referral to relevant medical and allied health specialists. Guidelines for managing patients with hEDS, as well as the clinical features and natural history, can be found at The Ehlers-Danlos Society website (https://www.ehlers-danlos.com/).

Other types of Ehlers-Danlos syndrome

There are at least 12 other types of EDS, some of which are associated with life-threatening complications (such as arterial or organ rupture). Clinical genetics assessment and testing is available for other types of EDS, as well as other connective tissue disorders associated with arterial dissection/aneurysm (such as Marfan Syndrome or Loeys-Dietz syndrome) where there is proven clinical features present and genetic testing is clinically indicated.

Where a patient has proven clinical features such as those listed below, referral to a Clinical Genetics can be made:

- Extensive widened atrophic scars
- Significant sagging skin
- Recurrent large hernias
- Significant kyphoscoliosis
- Personal or family history of spontaneous internal organ rupture/collapse
- Aortic/arterial aneurysm or dissection
- Arterial tortuosity
- Cleft palate
- Craniosynostosis
- Ectopia lentis
- Easy fractures
- Dysmorphic features that may indicate conditions such as Marfan syndrome
- Vascular EDS
- Loeys Dietz syndrome
- Recurrent pneumothoraces
- Hand and foot deformities
- TAAD
- arterial tortuosity syndrome.