Monash Health Referral Guidelines
GENERAL GENETICS

EXCLUSIONS

Services not offered by Monash General genetics

- Paternity testing - refer to private paternity testing laboratory
- Prenatal testing for advanced maternal age – refer directly to Fetal Diagnostic Unit
- Reproductive carrier screening for couples with no family history – consider referral to private genetic counselling service
- Women with increased risk results on First Trimester Screening for whom Non-Invasive Prenatal Testing (NIPT) has already been requested (please wait until NIPT result available before referring to Genetics - only if high risk)
- Cancer, lumps or tumours – Refer to Dr Marion Harris, Familial Cancer Clinic, Monash
- Management for common genetic conditions such as:
  - Haemochromatosis - consider referral to Haematologist.
  - MTFHR mutation carriers - consider referral to relevant specialist
  - Familial Hypercholesterolemia - consider referral to Advanced Lipid Disorder Clinic

REFERRALS ACCEPTED FOR:

- Diagnosis of a genetic condition
- Family history of a genetic condition
- Recurrence risk counselling (eg. history of a genetic condition in a child, risk of recurrence in future pregnancy)
- Pregnancy counselling (including pre-conception, consanguinity, fetal abnormality, family history of a genetic condition)
- Prenatal screening and testing counselling (e.g. increased risk results)
- Support after fetal loss or genetic diagnosis
- Pre-symptomatic and predictive testing counselling for adult onset conditions
- Discussions surrounding genetic testing
- Arranging genetic testing, where indicated

COMMON CONDITIONS AND CLINICS

Please complete work-up prior to referral to assist with patient care

CONDITIONS REQUIRING ADDITIONAL WORKUP:

Marfan Syndrome

Hypermobility or Ehlers-Danlos Syndrome

Developmental delay or intellectual disability

Short stature with a suspected bone dysplasia

Autism - Non syndromic & Syndromic

Dysmorphic Child

Childhood Hearing Loss

Cardiac Genetic Referrals

Chromosome Microarray Variant of Uncertain Significance (VUS)

SPECIALTY CLINICS:

- Neurogenetics
- Inherited Cardiac Disease
- Skeletal Dysplasia
- Childhood Hearing Loss
- Cleft Lip/Palate
- Skin Conditions
- Renal
- Haemoglobinopathies (thalassemia)

Head of unit:
Dr Matthew Hunter,
Consultant Clinical Geneticist

Program Director:
Professor William Sievert

Last updated:
02/04/2019

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GENERAL GENETICS

PRIORITY

All referrals received are triaged by **Monash Genetics clinicians** to determine **urgency of referral.**

EMERGENCY

For emergency cases please do any of the following:
- send the patient to the Emergency department OR
- Contact the on call registrar OR
- Phone 000 to arrange immediate transfer to ED

URGENT

The patient is pregnant.
The patient has a condition that has the potential to deteriorate quickly with significant consequences for health and quality of life if not managed promptly.

ROUTINE

The patient's condition is unlikely to deteriorate quickly or have significant consequences for the person's health and quality of life if the specialist assessment is delayed beyond one month

REFERRAL

How to refer to Monash Health

Mandatory referral content

Demographic:
Full name
Date of birth
Postal address
Contact number(s)
Email address
Referring GP details
including **provider number**
Usual GP (if different)
Interpreter requirements

Clinical:
Level of urgency
Reason for referral
Duration of symptoms
Management to date and response to treatment
Relevant test results, especially genetic test results
Past medical history
Current medications and medication history if relevant
Functional status
Psychosocial history
Family history
Letters confirming diagnosis
Diagnostics as per referral guidelines

Click here to download the outpatient referral form

PLEASE NOTE

- There is a very high demand for genetics appointments and there can be a long wait for routine appointments.
- In some situations genetic testing may be offered. Genetic testing may not be available or appropriate for all situations.
- Most genetic tests have no Medicare rebates. Some genetic tests may incur out of pocket costs, which will be discussed with your patient.
- Some genetic test results can take a very long time (sometimes 6 months or longer).
- It is important to plan a referral to genetics well in advance of future family planning, if possible.

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GENERAL GENETICS

CONTACT US

Medical practitioners

To discuss complex & urgent referrals or for general enquires contact the genetics team directly on the number below

P: (03) 9594 2026
F: (03) 9594 6022
E: monashgenetics@monashhealth.org

Submit a fax referral

Fax/post/email **ALL referral forms** to:

Dr Matthew Hunter
Monash Genetics
Special Medicine Centre
Monash Medical Centre
246 Clayton Road, Clayton VIC 3168

P: 9554 6022, **F:** 95946022
Email: monashgenetics@monashhealth.org

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GENETIC CONDITION

MARFAN SYNDROME

Initial GP/Specialist Work Up

- Echocardiogram
- Optometry/Ophthalmology assessment for ectopia lentis

WHEN TO REFER?

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK

HYPERMOBILITY OR EHLERS-DANLOS SYNDROME

Initial GP/Specialist Work Up

- Beighton Score
- Ehlers-Danlos Syndrome assessment
- Refer if concerning features such as: personal history of hernias, spontaneous internal organ rupture/collapse, aneurysm, cleft palate, craniosynostosis, ectopia lentis, easy fractures, dysmorphic features.
- Or a family history of above concerning features.

WHEN TO REFER?

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK

GENETIC CONDITION

DEVELOPMENTAL DELAY OR INTELLECTUAL DISABILITY

Initial GP/Specialist Work Up

WISC, WIPPSI, or other formal developmental assessment and:

- Tier 1: Fragile X, microarray and urine metabolic screen tests
- If tier 1 normal: Renal USS, Echocardiogram, Cranial USS or MRI brain (if dysmorphism or additional feature/s e.g. seizures)
- If seizures – EEG

WHEN TO REFER?

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK

SHORT STATURE WITH A SUSPECTED BONE DYSPLASIA

Initial GP/Specialist Work Up

Full genetic skeletal survey (please provide copies of Xrays if not performed at Monash Imaging)

WHEN TO REFER?

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK

GENETIC CONDITION

AUTISM - Non syndromic

WHEN TO REFER?

Initial GP/Specialist Workup

- If there are no other associated dysmorphic features or organ malformations, perform a Fragile X, microarray and metabolic screen.
- After these tests, there is little more to be gained from a genetic assessment unless the parents want to discuss recurrence risk in a subsequent pregnancy or the individual is planning to start a family and wants reproductive advice.

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK

AUTISM – Syndromic

WHEN TO REFER?

Initial GP/Specialist Workup

- If there are dysmorphic features, other medical problems and/or internal organ malformations, then again perform a Fragile X, microarray and metabolic screen.
- In this scenario there is good utility in a genetics consultation. Please refer.

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK

GENETIC CONDITION

DYSMORPHIC CHILD or ADULT

Initial GP/Specialist Workup

- Unless already done, perform Renal USS, Echocardiogram, Microarray, Cranial USS (or Consider brain MRI)
- Consider ophthalmology assessment if eyes involved
- Consider skeletal survey if short or disproportionate

WHEN TO REFER?

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK

CHILDHOOD HEARING LOSS

Initial GP/Specialist Workup

- Please refer to PHLIC clinic (Dr Kerryn Saunders) for medical work up for hearing loss, or perform the following assessments:
- Formal audiometry – for patients and first degree relatives
- CMV PCR on saliva (or urine) in neonate
- CMV PCR on guthrie card if not a neonate
- CMV IgG and IgM
- Cx26/30
- Inner ear MRI scan for structural anomalies
- Urine – for protein, blood, electrolytes
- TSH, T3, T4
- ECG if syncopal episodes , seizures or black outs
- Microarray

WHEN TO REFER?

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

BACK

GENETIC CONDITION

CARDIAC GENETIC REFERRAL

Initial GP/Specialist Workup

- Must have seen cardiology
- Echo/ECG/holter monitor as appropriate
- Rhythm disturbances – treadmill ECG, holter monitor

WHEN TO REFER?

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK

CHROMOSOME MICROARRAY VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)

Initial GP/Specialist Workup

Please arrange testing in both parents before referral to Genetics

WHEN TO REFER?

Urgent

Pregnant patients should be referred as early as possible (<10 weeks gestation)

Routine

Please provide all relevant information, testing results and family history with the referral.

BACK