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
Monash Health Referral Guidelines

GENERAL GENETICS

PRIORITIZE
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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Last updated: 02/04/2019
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
**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.

**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.
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.

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.
GENETIC CONDITION

AUTISM - Non syndromic

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.

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.

AUTISM – Syndromic

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.

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.

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

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**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)
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.

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.