Monash Health Referral Guidelines
GENERAL GENETICS

EXCLUSIONS

Services not offered by Monash General Genetics

For more information on exclusions, click on the link in the text

- **Paternity testing** - refer to private paternity testing laboratory (locate via internet search)
- **Prenatal testing for advanced maternal age** – refer directly to Fetal Diagnostic Unit
- **Increased aneuploidy risk –** on First trimester combine screening (FTCS) and Second Trimester Maternal Serum Screening
- Increased risk results on **Non-Invasive Prenatal Testing (NIPT)** will not be accepted as this is a private test and genetic counselling is provided by the test provider (lab) as part of the test cost.
- **Recurrent pregnancy loss–** if normal banded karyotype and no FHx of recurrent miscarriage (see note below; also see Prompt procedure on Monash Health intranet)
- **Recurrence risk counselling for previous child/pregnancy with aneuploidy (eg. Trisomy 21) –** if non-disjunction aneuploidy confirmed with banded karyotype
- **Reproductive Genetic Carrier Screening (RGCS)** for couples with no family history or family history of an autosomal recessive condition where the gene is on a carrier screening panel – consider referral to private RGCS provider
- Consanguineous couples with no other indication for a genetics appointment - consider referral to private RGCS provider
- **Generalised hypermobility or hypermobile EDS**, in the absence of evidence of a more serious connective tissue disorder. We cannot offer genetic testing to patients in this group and do not provide surveillance and management (see note below)
- **Pre-implantation Genetic Testing (PGT)-** refer to Royal Women's Hospital Fertility or to a private IVF provider or Fertility Specialist
- **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
  - **Factor V Leiden Thrombophilia** – consider referral to Haematologist

REFERRALS ACCEPTED FOR:

- Diagnosis of a genetic condition
- Family history of a genetic condition, excluding autosomal recessive genetic condition where the gene is on a carrier screening panel
- Recurrence risk counselling (eg. history of a genetic condition in a child, risk of recurrence in future pregnancy
- Pregnancy counselling (including pre-conception, fetal abnormality, family history of a genetic condition)
- Prenatal screening and testing counselling (e.g. increased risk results, excluding NIPT)
- 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

Last updated:
30/01/2026

Monash Health Referral Guidelines
GENERAL GENETICS

CONDITIONS

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

Referrals will not be accepted if required work-up is not complete prior to referral.

CONDITIONS/CLINICS REQUIRING ADDITIONAL WORKUP:

Medicare Funded Exome Sequencing

Connective Tissue Disorders:

- Ehlers-Danlos Syndrome
- Hypermobility
- Marfan Syndrome

Developmental delay or intellectual disability

Short stature with a suspected bone dysplasia

Autism - Non syndromic & Syndromic

Dysmorphic Child or Adult

Hearing Impairment & Deafness

Cardiac Genetic Referrals

Chromosome Microarray Variant of Uncertain Significance (VUS)

Familial Hypercholesterolaemia / Hyperlipidemia

MODY

Hereditary Haemorhagic Telangiectasia (HHT)

SPECIALTY CLINICS:

- Neurogenetics
- Inherited Cardiac Disease
- Skeletal Dysplasia
- Cleft Lip/Palate
- Skin Conditions
- Renal
- Thalassemia, Haemophilia, Blood disorders
- Immunogenetics

PRIORITY

All referrals received are triaged by **Monash Health 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 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

Last updated:
30/01/2026

Monash Health Referral Guidelines
GENERAL GENETICS

REFERRAL

How to refer to Monash Health

Find up-to-date information about how to send a referral to Monash Health on the eReferrals page on our website.

IMPORTANT NOTES:

- Referral of relatives: For almost all paediatric patients we will also need to assess the parents and affected siblings, and for those children found to have a genetic diagnosis we will need to test the parents and siblings; also for almost all pregnant women we will need to assess and potentially test their partner. Therefore, please provide referral of other relatives at the tie of referring your patient as this will help us provide comprehensive genetic services and testing to the family.
- There is a very high demand for genetics appointments and there can be a long wait for appointments. Other Victorian Genetic Services may have shorter wait times.
- If your patients condition changes, please contact us so that we can re-triage the referral and determine if a more urgent appointment is required.
- If your patient or a close relative is family planning please state this on the referral.
- 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.

Last updated:
30/01/2026

Monash Health Referral Guidelines
GENERAL GENETICS

CONTACT US

Medical practitioners

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

General enquiries

P: (03) 9594 2026

CONTACT US

Austin Hospital – Genetics Service
(Patients seen: Paediatrics, Adults)

Address:145 Studley Rd, Heidelberg, 3084
Email: genetics@austin.org.au
Phone: 9496 3027
Fax: 9496 4385

Royal Women's Hospital – Clinical Genetics Service
(Patients seen: Prenatal, Reproductive, Neonates)

Address: 20 Flemington Rd, Parkville, 3052
Phone: 8345 2180
Fax: 8345 2179

Royal Melbourne Hospital – Genomic Medicine Department
(Patients seen: Adults)

Address: 300 Grattan St, C-PO RMH Parkville, 3050
Email: genetics@mh.org.au
Phone: 9342 7151
Fax: 9342 4267

Sunshine Hospital – Genetics Service
(Patients seen: Prenatal, Reproductive, Neonates)

Address: 176 Furlong Road, St Albans, 3021
Phone: 8458 4346
Fax: 8458 4254

Victorian Clinical Genetics Service (VCGS)
(Patients seen: Paediatrics, Adults regional Vic only)

Address: Royal Children's Hospital, Flemington Road, Parkville, 3052
Email: vcgs.clinical@vcgs.org.au
Phone: 8341 6201
Fax: 8341 6390

For VCGS quarterly visiting clinics to **Warragul** and **Sale** use contact details above.

MEDICARE FUNDED EXOME SEQUENCING IN CHILDREN 0-10 YEARS

MEDICARE FUNDED EXOME SEQUENCING

New Medicare item numbers allow paediatricians to request exome sequencing for their patients, after consultation with, and approval from, a Clinical Geneticist. Melbourne Genomics have developed a procedure for Paediatricians (internal and external to Monash Health) to request exome sequencing in tandem with local laboratories.

Medicare Eligibility Criteria

To be eligible for a Medicare funded exome, the child must have:

A. Intellectual disability (at least moderate severity)

OR

B. Dysmorphic AND at least 1 congenital anomaly

AND

The test must be requested prior to the child's 11th birthday (i.e. 10 years and 364 days).

The child must have had a non-diagnostic microarray.

They must be an outpatient (inpatients not eligible).

HOW TO ORGANISE?

Visit the website for all information and to order the test:

www.paediatricgenomics.org.au

WHEN TO REFER?

In most circumstances it will be significantly quicker for a paediatrician to order testing themselves, however they can refer to Genetics to organise if required, or under the following circumstances:

- Assessment by a Clinical Geneticist preferred
- Patient does not meet eligibility criteria for Medicare Funded Exome Sequencing

BACK

CONNECTIVE TISSUE DISORDERS

EHLERS-DANLOS SYNDROME (with concerning features)

- Refer if concerning features such as personal or family history of: spontaneous internal organ rupture/collapse, unexplained prolapse, aortic/arterial aneurysm or dissection, arterial tortuosity, cleft palate, craniosynostosis, ectopia lentis, or dysmorphic features that may indicate conditions such as Marfan syndrome, vascular EDS, Loeys-Dietz syndrome, TAAD, or arterial tortuosity syndrome
- We can no longer accept patients with generalised hypermobility, or hypermobile EDS due to limited service capacity, lack of genetic testing for hypermobility spectrum disorder, and we do not offer a surveillance and management service.
- In many circumstances, allied health input can significantly improve symptoms and quality of life for individuals with generalised hypermobility and hypermobile EDS.
- Referral to organisations such as Zebras Australia (www.zebrasaus.com.au), may be helpful. Patients may be eligible to Medicare rebates, however there are out of pocket costs.

Initial GP/Specialist Work Up

- Beighton Score
- Ehlers-Danlos Syndrome assessment
- Include inflammatory markers
- echocardiogram or other specific assessments for connective tissue disorders listed above.

WHEN TO REFER?

Urgent

Patients with a personal or family history of high risk features of serious connective tissue disorder.

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

Routine

Patients with a personal/ family history of high risk features of a serious connective tissue disorder under appropriate management, with no immediate or short-term management implications.

BACK

MARFAN SYNDROME

Initial GP/Specialist Work Up

- Diagnostic criteria here
- Marfan systemic score **essential**
- Echocardiogram
- Optometry/Ophthalmology assessment for ectopia lentis

WHEN TO REFER?

Urgent

Patients with a personal or family history of high risk features of serious connective tissue disorder.

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

Routine

Patients with a personal/ family history of high risk features of a serious connective tissue disorder under appropriate management, with no immediate or short-term management implications.

BACK

GENETIC CONDITION

AUTISM - Non syndromic (i.e. no additional phenotypic features)

WHEN TO REFER?

Initial GP/Specialist Workup

- Most non-syndromic level 1 or level 2 autism is multifactorial and not well assessed on genetic testing. Referrals will be rejected unless there is an acceptable reason for referral.
- Perform a microarray and Fragile X testing, ideally on a saliva sample.
- If Fragile X and microarray tests are normal, there is little more to be gained from a genetic assessment unless the parents or individual want to discuss recurrence risk in a pregnancy.
- If microarray identifies a copy number variant, please complete targeted parental microarrays prior to referral to Monash Genetics.
- If there is a history of significantly delayed motor milestones (e.g. >18mth to start walking independently) but not moderate-profound ID, consider referral to Monash Genetics.
- If a microarray only identifies LCSH, this suggests a consanguineous parental relationship and referral to Monash Genetics is not indicated. Please consider advising parents planning on further reproduction to seek personal GP review regarding reproductive genetic carrier screening options.

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/ family history of Autism, under appropriate management, with no immediate or short-term management implications.

BACK

AUTISM – Syndromic

WHEN TO REFER?

Initial GP/Specialist Workup

- If there are dysmorphic features (see dysmorphism guidelines), neurocutaneous features, other medical problems and/or internal organ malformations, then first perform microarray and Fragile X testing, ideally on a saliva sample.
- In this scenario there is utility in a genetics consultation.
- If aged between birth and 10 years 364 days, and syndromic, consider eligibility for a **Medicare Funded Exome**

Urgent

Patients whose conditions is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/ family history of Autism, under appropriate management, with no immediate or short-term management implications.

BACK

GENETIC CONDITION

CARDIAC GENETIC REFERRAL

Initial GP/Specialist Workup

- Must have seen a cardiologist
- When providing ECG - Please provide **12 lead ECG print out and full report**
- Always provide echocardiogram report
- **Aortopathy** – must have transthoracic echocardiogram OR CT aortogram or MRI aortogram report documenting aortopathy
- **Cardiomyopathy** – must have transthoracic echocardiogram report and ECG; Holter monitor ideal but not necessary
- **Arrhythmia** – must have ECG and transthoracic echocardiogram report; Holter monitor report ideal but not necessary
- **Congenital heart disease** – must have transthoracic echocardiogram or cardiac MRI report; ECG and Holter monitor report ideal but not necessary

WHEN TO REFER?

Urgent

Patients with abnormal cardiac assessment, family history of cardiac condition, with immediate or short-term management implications.

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

Routine

Patients with a personal/family history of cardiac condition, under appropriate management, with no immediate or short-term management implications.

BACK

CHROMOSOME MICROARRAY VARIANT OF UNCERTAIN SIGNIFICANCE (VUS)

Initial GP/Specialist Workup

- Please arrange targeted testing of both parents for the identified chromosome abnormality **before referral to Genetics** (and include copies of report with referral)
- If parental results are not included, the referral will not be accepted (please indicate if there are circumstances which mean testing of both parents cannot be arranged prior to referral)

Patient information Resources:

Unique group: www.rarechromo.org

VCGS microarray information sheet

WHEN TO REFER?

Urgent

Patients with a personal/family history of a chromosome microarray variant, with immediate or short term management implications. Pregnant patients should be referred as early as possible (<10 weeks gestation if possible).

Routine

Patients with a personal/family history of chromosome abnormality, under appropriate management, with no immediate or short-term management implications.

BACK

GENETIC CONDITION

CLEFT PALATE & CRANIOFACIAL

WHEN TO REFER?

Initial GP/Specialist Workup

- Patients must have had a chromosomal microarray (molecular karyotype) prior to referral
- If a **submucosal cleft palate** is a possible diagnosis, referral to the Cleft Clinic prior to referral to Monash Genetics through Tania Green: https://monashchildrenshospital.org/cleft-and-craniofacial-anomalies-service/
- If dysmorphic – see dysmorphology guideline above

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of a cleft lip/palate, under appropriate management, with no immediate or short-term management implications

BACK

DEVELOPMENTAL DELAY OR INTELLECTUAL DISABILITY

WHEN TO REFER?

Initial GP/Specialist Work Up

- Clarification about the degree of GDD or ID is essential for us to accept referrals and appropriately triage patients. Please clarify if the degree is mild, moderate, severe or profound based on DSM V criteria.
- Ideally provide a WISC, WIPPSI, or other formal developmental assessment.
- Microarray and Fragile X testing, ideally on a saliva sample
- If dysmorphism (see dysmorphism guideline) or major congenital anomaly, consider ordering: Renal USS, Echocardiogram, Cranial USS or MRI brain
- If seizures – EEG
- If focal neurology – must see neurology first
- If under 10 years 364 days with at least moderate ID – see Medicare exome guideline.

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/ family history of developmental delay or intellectual disability, under appropriate management, with no immediate or short-term management implications.

BACK

GENETIC CONDITION

FAMILIAL HYPERCHOLESTEROLAEMIA / HYPERLIPIDEMIA

WHEN TO REFER?

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal history of hypercholesterolaemia, under appropriate management, will not be seen

Initial GP/Specialist Workup

Patients must first be referred to the Advanced Lipid Disorder Clinic for workup. Please refer to the following link https://www.monashheart.org.au/index.php/for-medical-professionals/referrals/

Genetics does not accept referrals for lipid disorders that have not come through the Advanced Lipid Disorder Clinic.

- Must have lipid profile
- Must fulfil Dutch lipid criteria (score greater ≥4)
- Dutch lipid criteria does not apply for children

BACK

DYSMORPHIC CHILD or ADULT

WHEN TO REFER?

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients who are dysmorphic, under appropriate management, with no immediate or short-term management implications.

Initial GP/Specialist Workup

- Microarray essential, ideally on a saliva sample
- Unless already performed, perform Renal USS, Echocardiogram, Cranial USS (or consider brain MRI) and ideally audiology
- Consider optometry/ophthalmology assessment if eyes involved
- Consider skeletal survey if short or disproportionate stature
- If has intellectual disability then perform formal psychometric testing as per referral guideline for ID.
- If under 10 years 364 days with at least moderate ID – see Medicare exome guideline.

BACK

GENETIC CONDITION

HEARING IMPAIRMENT / DEAFNESS

WHEN TO REFER?

Initial GP/Specialist Workup

Children with hearing loss/deafness: Please refer to Dr Talia Maayan and Dr Saniya Kazi, PHLIC (Paediatric Hearing Loss Investigation Clinic), Monash Children's Hospital, or a PHLIC equivalent at another hospital, for medical work up prior to referring to genetics.

Adults with hearing loss/deafness: Please refer to ENT first for medical workup prior to referring to genetics.

Patients will not be accepted in genetics until assessed in PHLIC/PHLIC equivalent at another hospital (children) or by an ENT (adults) and all relevant reports (as below) are provided.

Information to be included in the referral

(Referral will be rejected without reports marked as 'required'):

- Formal audiometry – for patients (required) and first degree relatives (recommended)
- Inner ear MRI (child/adult) or CT (adults) for structural anomalies (recommended)
- Urine – blood (required), protein, electrolytes (recommended)
- TSH, T3, T4 (required)
- Chromosome microarray (required if learning difficulties, dysmorphism or bilateral sensorineural hearing loss)
- Cx26/30 gene testing (if available)
- Ophthalmology review (required if vision impaired, balance issues or delayed motor milestones in infancy)
- ECG (recommended if bilateral severe/profound hearing loss with a family history of sudden death)
- If under 11 yr with dysmorphism and a congenital anomaly, they may be eligible for Medicare funded exome sequencing – see Medicare exome guideline.
- If hearing loss presented before 18 years of age, is permanent bilateral moderate, severe, or profound (>40 dB in the worst ear over 3 frequencies) and classified as sensorineural, auditory neuropathy or mixed,

Note PN.7.13 | Medicare Benefits Schedule (health.gov.au)

Adult reproductive partner or biological relative of a person with genetic hearing loss/deafness: Please ensure the laboratory report for the partner/relative with genetic hearing loss/deafness including the pathogenic/causative gene variant is provided when referring to genetics.

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of hearing loss, under appropriate management, with no immediate or short-term management implications

BACK

GENETIC CONDITION

HEREDITARY HEAMORRHAGIC TELANGIECTASIA (HHT)

WHEN TO REFER?

Initial GP/Specialist Workup

- Please include all medical correspondence and imaging reports with referral. This includes: oxygen saturation check, echocardiographic “bubble study”, abdominal (liver) ultrasound and MRI brain results.
- Please include copies of all investigations if referring for a family history of HHT.
- Genetics does not provide ongoing medical surveillance. If requesting surveillance, patients can be referred to: A/Prof Joanne Rimmer, Complex Rhinology Clinic, Monash Health.

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of HHT, under appropriate management, with no immediate or short-term management implications

BACK

IMMUNOGENETICS

WHEN TO REFER?

Initial GP/Specialist Workup

- Initial referral to child or adult Immunology Service at Monash Health

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of immunological condition, under appropriate management, with no immediate or short-term management implications

BACK

MODY

WHEN TO REFER?

Initial GP/Specialist Workup

- Must have seen an endocrinologist
- Must provide: HbA1c, C-peptide > 0.2nmol/L, b-cell autoantibody negative
- If patient has renal cysts, please indicate and include renal ultrasound report (not mandatory)

- Calculator for patients of Caucasian descent (refer if >25% probability)

https://www.diabetesgenes.org/exeter-diabetesapp/

- For non-Caucasian patients with diabetes, the calculated probability is likely to be much lower due to the higher prevalence of young-onset Type 2 diabetes. Low BMI and age of diagnosis are the most important discriminators for MODY vs Type 2 diabetes in non-Caucasian groups.

Urgent

Patients with renal cysts or poor control, on insulin. Pregnant patients should be referred as early as possible (<10 weeks gestation if possible).

Routine

Diagnosis <30 years of age without insulin resistance and family history of DM in at least 1 individual < 25 in at least two generations.

BACK

GENETIC CONDITION

NEUROGENETICS

Initial GP/Specialist Workup

- Only refer patients/families with a likely genetic basis to their condition and refer parents/siblings if relevant
- Enclose any relevant correspondence and investigations performed (i.e., EEG, MRI, CT, nerve conduction studies, muscle biopsy, genetic testing, microarray etc.)
- If patient is symptomatic, referral to neurologist should also be considered for management of their condition. Share correspondence and results
- For brain abnormalities, referrals will not be accepted without imaging reports (unless performed at Monash Imaging)
- Referrals will not be accepted for APOE testing
- Please indicate if patient may be eligible for therapies following genetic testing
- For pre-symptomatic testing, please provide genetic test results where possible

WHEN TO REFER?

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of a neurological disorder, under appropriate management, with no immediate or short-term management implications

PAEDIATRIC RENAL GENETICS

BACK

Initial GP/Specialist Workup

- Must have seen a nephrologist and provide correspondence.
- Please include all imaging and investigation reports. Referrals will be rejected if Ix are not included in the referral.
- If a patient is syndromic (additional features to the renal condition) then they will be seen in the general genetics clinic before being seen in renal genetics clinic

WHEN TO REFER?

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of renal disease with a likely genetic basis, under appropriate management, with no immediate or short-term management implications

ADULT RENAL GENETICS

BACK

- Referrals for suspected isolated renal genetic disorders in adults will be redirected to the Monash Health Nephrology Service who run a dedicated monthly renal genetics clinic
- Syndromic patients can still be seen through our service – please follow the same process as per Paediatric patients

WHEN TO REFER?

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of renal disease with a likely genetic basis, under appropriate management, with no immediate or short-term management implications

BACK

GENETIC CONDITION

SKIN CONDITIONS

Initial GP/Specialist Workup

- Patients must have had a specialist dermatology assessment prior to referral (please include correspondence)
- If dysmorphic – see dysmorphology guideline above
- If multiple café au lait macules – consider ophthalmology/optometry assessment, and brain MRI
- If features of tuberous sclerosis complex – consider renal USS, MRI-brain and ophthalmology assessment +/- echocardiogram (if <3yo)
- If features of Incontinentia Pigmenti in a newborn/infant, strongly consider urgent referral to Ophthalmology due to risk of retinal detachment

WHEN TO REFER?

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of a skin disorder, under appropriate management, with no immediate or short-term management implications.

BACK

THALASSAEMIA, HAEMOPHILIA & BLOOD DISORDERS

Initial GP/Specialist Workup

- Patients and their partner should have a FBE, Haemoglobin electrophoresis and iron study performed
- If abnormalities detected on FBE/ Hb electrophoresis, DNA studies for thalassemia should be ordered and a referral to genetics made.

Information to be included in the referral

Referral MUST include:

- Partner details must be included on the referral
- All test results for both patient and partner

Arrange testing and referral prior to pregnancy or as early in pregnancy as possible

WHEN TO REFER?

Urgent

Patients whose condition is deteriorating, or have immediate or short-term management implications.

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

Routine

Patients with a personal/family history of a thalassaemia condition, under appropriate management, with no immediate or short-term management implications and no reproductive implications.

BACK

SHORT STATURE WITH A SUSPECTED BONE DYSPLASIA

If short stature with no disproportion, then refer to endocrinology first.

Initial GP/Specialist Work Up

- Full genetic skeletal survey, ideally through Monash (please provide access to visual/physical copies of X-rays if not performed at Monash Imaging)
- Microarray (see dysmorphism guidelines)
- Optional other investigations if indicated: Echocardiogram, abdominal ultrasound, developmental assessment, vision & hearing assessments
- **If osteogenesis imperfecta is suspected, a skeletal survey is not necessary**

WHEN TO REFER?

Urgent

Patients whose conditions is deteriorating, or have immediate or short-term management implications

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

Routine

Patients with a personal/ family history of short stature or bone dysplasia, under appropriate management, with no immediate or short-term management implications.

BACK

EXCLUSIONS (Additional Information)

MTHFR

Background

- We are unable to accept referrals for this indication.
- MTHFR variants/ polymorphisms are very common in the population.
- The majority of people who have one or two variants in the MTHFR gene do not develop health problems

ADDITIONAL INFORMATION?

There is minimal/conflicting evidence to support testing or treating for MTHFR polymorphisms.

Click here for a useful review article about MTHFR polymorphisms.

Some helpful patient fact sheets are:
- https://www.genetics.edu.au/health-professionals/FS64-MTHFR-GENE-TESTING-FOR-PATIENTS.pdf
- VCGS MTHFR fact sheet

BACK

NON-INVASIVE PRENATAL TESTING (NIPT)

Background

- We are not able to accept referrals for this indication.
- NIPT is a private test with out of pocket costs to the patient.
- NIPT providers offer private telephone genetic counselling as part of the cost of the test.
- If your patient has an increased risk on their NIPT test, please refer your patient to the NIPT service provider (lab) that performed their test, for genetic counselling.
- If your patient is interested in having NIPT, please order testing yourself, or refer to an NIPT service provider to arrange testing.

WHATS NEXT?

If your patient decides to have diagnostic testing following genetic counselling for an increased risk result, they can be referred to one of the following:
- A private obstetric ultrasound clinic.
- The public hospital where they are booked to deliver their baby (if diagnostic testing is available).
- The Fetal Diagnostic Unit at Monash Health.

If a chromosome abnormality is confirmed after amniocentesis/CVS testing, and your patient requires genetic counselling to assist with decision making, you are welcome to refer to us.

BACK

Increased Aneuploidy Risk – First Trimester Combined Screening (FTCS) and Second Trimester Maternal Serum Screening (MSS)

Background:
We are unable to accept referrals for increased aneuploidy risk based on FTCS or MMS results.

Referral Guidance:
If the patient opts for Non-Invasive Prenatal Testing (NIPT), please arrange this testing as the initial step.
If counselling is required prior to NIPT or diagnostic testing:
- For patients within the MH Maternity catchment, refer to the local maternity unit for counselling with a consultant or senior registrar within the Antenatal Care (ANC) service.
- For patients outside the MH Maternity catchment, refer to the local maternity department for ongoing counselling and care.

Counselling should include:
- An increased risk result is NOT a diagnosis
- A discussion of care options with the couple, including NIPT.
- If NIPT is declined, offer an ultrasound or diagnostic procedure (CVS/amniocentesis).
- If diagnostic testing is requested, refer directly to a Fetal Diagnostic Unit/Service or private obstetric clinic.

EXCLUSIONS (Additional Information)

RECURRENT PREGNANCY LOSS

Background

- We are unable to accept referrals for this indication.
- If 3 or more losses then perform standard (G-Banded) karyotype in both partners looking for balanced chromosomal rearrangements (e.g. translocations).
- If karyotype normal, then no indication to refer to genetics.
- If strong multigenerational family history of recurrent pregnancy loss (3 losses in 3 relatives), then genetics can consider accepting referral.

BACK

REPRODUCTIVE GENETIC CARRIER SCREENING (RGCS)

Background

- We are not able to accept referrals for this indication.
- There is MBS funding for RGCS for 3 genes (*CFTR*, *FMR1* and *SMN1*) available through multiple local laboratories but there is currently an out-of-pocket cost for expanded RGCS.
- RGCS providers offer private genetic counselling as part of the cost of the test.
- If your patient requests RGCS, in the absence of a family history of genetic disease, please refer to a RGCS provider to arrange testing.
- If your patient has had RGCS and obtained a carrier result, please refer to the RGCS provider that performed their test, for genetic counselling.

OTHER SUGGESTIONS?

There are many RGCS providers available. If you are looking for a suggestion, some providers that we are aware of who provide a comprehensive service are:

- Victorian Clinical Genetics Service (VCGS)
  - https://www.vcgs.org.au/expandedcarrier
  - https://www.vcgs.org.au/prepair
- Eugene
  - https://eugenelabs.com/
- Lumi Health
  - www.lumihealth.com.au
- Virtus Genetics
  - Genetic Carrier Screening - Virtus Genetics
- Genomic Diagnostics
  - www.genomicdiagnostics.com.au/individuals/genetic-carrier-screening/

BACK

RECURRENCE RISK COUNSELLING FOR PREVIOUS CHILD/PREGNANCY WITH ANUEPLOIDY (EG. Trisomy 21)

Background

- We are unable to accept referrals for this indication.
- If prenatal/child's banded karyotype confirms non-disjunction trisomy, then relatively low recurrence risk and recommend prenatal aneuploidy screening for future pregnancy.
- If prenatal banded karyotype is not available (ie. Chromosome microarray only) then organise parental banded karyotypes
- If parental karyotype normal, then no indication to refer to genetics.

EXCLUSIONS (Additional Information)

PRE-IMPLANTATION GENETIC TESTING (PGT)

Background

- We are unable to accept referrals for this indication.
- PGT is offered through the Monash Fertility Clinic
- PGT is also offered by private IVF providers.
- If your patient has questions regarding the option of PGT, please refer to an IVF provider, or Fertility Specialist.

ADDITIONAL INFORMATION?

https://monashhealthfertility.org/

A list of IVF providers can be found at:

- https://www.varta.org.au/fertility-treatment/getting-started-find-fertility-clinic#Accredited-fertility-clinics

Check the providers website to determine if PGT is offered.

BACK