Clinical Genetics: Referral Criteria

Royal North Shore Hospital Ambulatory Care Centre

Royal North Shore Hospital

At RNSH we provide the following General Genetics clinics:

- **Diagnostic Genetics Clinic**
- **Genetic Counsellor Clinic**
- **Prenatal Genetics Clinic**
- **Neurofibromatosis Clinics**

In addition, we provide a number of specialty clinics for those with specific types of Neurofibromatosis: including the:

- **NF1 skin clinic**
- **NF1 breast screening clinic**

For information on how to refer to the Neurofibromatosis genetics and speciality clinics, please see referral criteria for RNSH NF clinics.

If you are a health professional and you are looking to refer a patient to our service, please see the information provided below.
Referral criteria information is divided into the following sections:

1. **Mandatory referral information**
2. **Indications for referral**
3. **When the genetic clinic is unable to provide a service**
4. **Out of area referrals**
5. **How to refer to the diagnostic, genetic counsellor and prenatal genetics clinics**
6. **RNSH out of area referrals**

1. Mandatory referral information

All Genetics referrals must include a consultant being referred to in RNSH outpatients. If unsure, please address to **A/Prof Yemima Berman and Associates.**

All referrals must include patients details, provision diagnosis and reason for referral, finding/treatment to date, how this affects the patient, significant medical history, list of medications and relevant social information. Please indicate preferred language if an interpreter is required.

Please clearly indicate if this referral relates to an ongoing pregnancy.

Please attach results of any investigations to the referral and ensure the patient brings hard copies to their appointment. This will help avoid unnecessary duplication of tests, additional appointments and delay.

Referrals with insufficient information will be returned to the referring doctor until further information is provided to the clinic.

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2. Indications for referral

Referrals for children will only be accepted from Paediatricians

Presenting complaint	When to Refer	RNSH Clinic
Developmental Delay/ Congenital anomalies	Anyone with a rare genetic or chromosomal diagnosis, congenital anomalies and/or significant developmental delay (e.g., mild ID).	Diagnostic Genetic Clinic
Pregnancy concern	Pregnant women or their partners who are affected, or who have a family history of an inherited condition or foetal abnormality suggestive of an underlying genetic disorder.	Prenatal Clinic
Preconception concerns	• Anyone with a personal and/or family history of a genetic or chromosomal condition who is seeking updated information (e.g., before starting their family). • Thalassaemia testing where FBE/HbEP has been performed in both partners and is not reassuring in at risk populations. Appendix 1 • Couples who have had reproductive carrier screening and have been identified as carriers of the same genetic condition. Appendix 2	Genetic Counsellor Clinic
Predictive genetic testing	Predictive testing for inherited disorders in unaffected individuals with a family history.	Genetic Counsellor Clinic
Hypermobility/ Connective Tissue Disorder	Anyone with a personal history of the following red flag complications, or a family history of these in the presence of hypermobility (Beighton >4 in adults, >6 in children) • Young onset of thoracic aortic enlargement, vascular dissection or extensive varicosities • Ectopia lentis • Extensive widened atrophic scars and poor would healing/ recurrent large hernias • Severe scoliosis • Personal/family history of organ rupture • Recurrent pneumothoraces Further details including management. Appendix 3	Diagnostic Genetic Clinic
Neurofibromatosis Last update: March 2021	Adults and Children with confirmed or suspected Neurofibromatosis type 1, type 2, or schwannomatosis for diagnosis, management plan or complex management.	Neurofibromatosis Clinic (suspected NF1 may be seen in General clinic)

3. The RNSH Genetics Clinic is unable to provide a service for the following:

- Individuals without a family history of a genetic condition who are seeking reproductive carrier screening. Appendix 2
- Individuals with a personal and/or family history of Ehlers Danlos syndrome type 3 / hypermobility / joint laxity without red flags. Appendix 3
- Children/ adults with autism without intellectual disability, family history or unusual facial features- baseline investigations should be performed by the managing doctor. Appendix 4
- Variants of uncertain significance on chromosomal microarray that have no gene content, or isolated loss of heterozygosity. Appendix 5
- Pregnant women with a high risk due to advanced maternal age or first trimester screening investigations, who have not yet had a diagnostic test.
- Couples who have had recurrent miscarriages where the cause is NOT due to a chromosomal anomaly. Conventional karyotype should be performed by the managing doctor.
- Individuals who have had or are considering genetic testing of the MTHFR gene. Appendix 6
- Individuals who have had or are considering 'direct to consumer' genetic testing.
- Individuals who have had or are requesting genetic testing relating to paternity
- Consanguinity. Appendix 7
- Teratogen exposure. Contact Mothersafe 93826539 for advice. Appendix 8
- Individuals who are not residents of NSLHD. See Out of Area Referral page.

Please note: Patients can also be referred to a Genetics Specialist in private rooms, which usually have a significantly shorter waiting period. There will likely be an out of pocket cost for the consultation. See page 7 for contact details.

If your patient does not meet our referral criteria but you would like advice, or think they would benefit from a Genetics consultation please contact us on 94631727.

4. Out of Area Referral

Resident of Northern Sydney Local Health District Catchment	Yes - referral accepted at RNSH
Referral from other specialist, for specialist opinion	Yes - referral accepted at RNSH
Resident of other Local Health District that DOES NOT provide the clinical service e.g. rural, outer metro	Yes, but service or problem needs to be documented on referral
Continuing care of existing condition we already manage	Yes, provided existing or related condition documented on referral
Demonstrated complexity requiring services of Royal North Shore Hospital Ambulatory Care Centre	Yes but must be explicitly documented on referral
Compassionate circumstances (e.g. family proximity, staff)	Yes but must be explicitly documented on referral
Resident of other Local Health District that offers the service	Refer to your Local Health District

Check if the home address is within Northern Sydney Local Health District here:
https://www.health.nsw.gov.au/lhd/Pages/lhd-maps.aspx

5. To refer

Urgent referrals: Contact the RNSH Switchboard (9926 7111) and ask to page the on-call Genetics Fellow, Geneticist or Genetic Counsellor.

Urgent referrals are accepted for;

1. Patients who are currently pregnant or currently undergoing IVF with a genetic concern related to the current pregnancy- please ensure an EDD/LMP are clearly indicated.
2. Inpatient consultations
3. Children under the age of 6 months
4. Where the results of the consultation are expected to be relevant for urgent medical management decisions

Routine referrals:

Send a referral letter to:
Department of Clinical Genetics: NSLHD-ClinicalGenetics@health.nsw.gov.au or fax to (02) 9463 1057.

Please note: genetic testing may or may not be offered to your patient. The decision to offer genetic testing is made on a case-by-case assessment.

Contact us:

Clinical Genetics Department: (02) 9463 1727

Clinic Location:
Ambulatory Care Centre
Check-In B, Level 3
Royal North Shore Hospital
Reserve Road
St Leonards 2065

Postal Address:
Clinical Genetics Department
Clinical Admin 3E, Acute Services Building
Royal North Shore Hospital
Reserve Rd
St Leonards 2065

6. RNSH consultant referral options

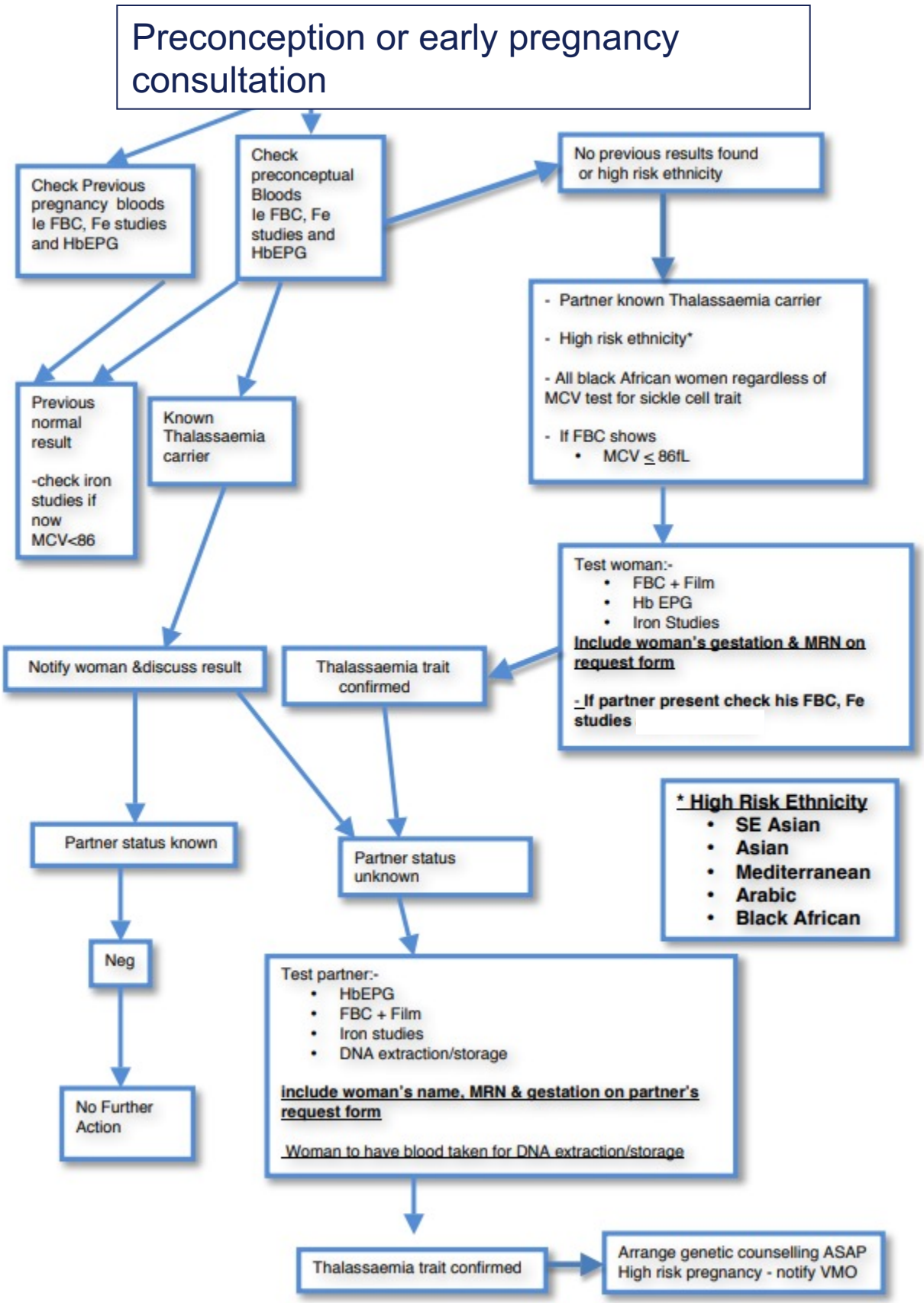
The following Clinical Geneticists work at RNSH. Some have private rooms, which patients can be referred to and usually have a significantly shorter waiting period. From the rooms patients may still have access to the public system for testing if they fulfil relevant clinical / proritisation criteria. There will likely be an out of pocket cost for the consultation at the private rooms.

Consultant	RNSH Clinic	Location of private services
Dr Sondhya Ghedia	Clinical Genetics	**North Shore Private Hospital** Suite 6, Level 3 North Shore Private Hospital, 1 Westbourne Street St Leonards NSW 2065 Ph: 94395594 Fax: 9475 0640
Dr Mary Louise Freckmann	Clinical Genetics	**North Shore Private Hospital** Suite 6, Level 3 North Shore Private Hospital, 1 Westbourne Street St Leonards NSW 2065 Ph: 94395594 Fax: 9475 0640
Dr Katrina Morris	Neurofibromatosis	**The Brain and Mind Centre** Level 4/94 Mallett St, Camperdown NSW 2050 Ph: 9351 0730

This document has been developed in consultation with:

1. A/Prof Yemima Berman, HOD Clinical Genetics
2. The RNSH Clinical Genetics Team

Appendix 1- **Thalassaemia testing pathway**



Appendix 2- **Reproductive carrier screening**

Reproductive carrier screening is a genetic test that can determine whether couples are carriers of genetic conditions to inform their family planning. Reproductive carrier screening should be offered to all individuals and couples planning a pregnancy regardless of family history or ethnicity[1].

Reproductive carrier screening for individuals and couples at population risk can be arranged by general practitioners, obstetricians, ultrasound and IVF clinics and private genetics services. The RNSH Department of Clinical Genetics is unable to offer genetic testing to individuals or couples who do not have a family history of a genetic condition. If you are unsure whether your patient or their partner should be referred RNSH Department of Clinical Genetics, please call the department (02 9463 1727) and ask to speak with the genetic counsellor on-call.

Resources for health professionals:
RACGP Genomics in General Practice Reproductive Carrier Screening Guidelines
https://www.racgp.org.au/clinical-resources/clinical-guidelines/key-racgp-guidelines/view-all-racgp-guidelines/genomics/reproductive-carrier-screening

RACGP Beware the Rare Carrier Screening website
(NB: includes factsheet, guidelines and list of providers/labs through link to McKenzie's mission website)
https://bewaretherare.com.au/carrier-screening/

Resources for consumers:
Centre for Genetics Education factsheet
https://www.genetics.edu.au/publications-and-resources/facts-sheets/FS65REPRODUCTIVECARRIERSCREENING.pdf

References:
[1]RANZCOG (2019). Genetic carrier screening.
https://ranzcog.edu.au/RANZCOG_SITE/media/RANZCOG-MEDIA/Women%27s%20Health/Statement%20and%20guidelines/Clinical-Obstetrics/Genetic-carrier-screening(C-Obs-63)New-March-2019_1.pdf?ext=.pdf (accessed 12th January 2021)

Appendix 3- **Hypermobility Spectrum Disorder Information**

Joint hypermobility is common in the general population and often familial. Only a small proportion of people with joint hypermobility will require medical surveillance and genetic advice and they will usually have additional distinctive clinical features. The relatively common hypermobility spectrum disorder; (HSD) which may include individuals who meet criteria for hypermobile EDS (hEDS;) can be a multisystem disorder and may have associated pain, autonomic dysfunction and psychological impact with altered quality of life. There is no known underlying genetic change for this condition and no genetic testing is available.
https://ehlers-danlos.com/wp-content/uploads/hEDS-Dx-Criteria-checklist-1.pdf

The clinical genetics service is not able to provide treatment or ongoing management or surveillance.

Referral - is recommended to a relevant medical specialists- paediatrician for children, rheumatologist, rehabilitation physician, pain physician and allied health professionals physiotherapist and occupational therapists.

Further information for management
https://www.schn.health.nsw.gov.au/fact-sheets/joint-hypermobility
www.rcgp.org.uk/clinical-and-research/resources/toolkits/ehlers-danlos-syndromes-toolkit.aspx

Appendix 4- **Autism Information**

Further information regarding autism;

Great Ormond Street Hospital fact sheet on genetics of autistic spectrum disorders
https://www.gosh.nhs.uk/file/11716/download?token=4HHz6kc-

RACGP Genomics in General Practice – Autism Spectrum Disorder
https://www.racgp.org.au/clinical-resources/clinical-guidelines/key-racgp-guidelines/view-all-racgp-guidelines/genomics-in-general-practice/autism-spectrum-disorder

Appendix 5- **Chromosome Microarray**

Centre for Genetics Education Factsheet: CHROMOSOME MICROARRAY (CMA) TESTING IN CHILDREN & ADULTS
https://www.genetics.edu.au/publications-and-resources/facts-sheets/fact-sheet-16-chromosome-microarray-cma-testing-in-children-and-adults

Unique: Understanding Rare Chromosome and Gene Disorders
https://www.rarechromo.org/disorder-guides/

Appendix 6- **MTHFR Information**

Centre for Genetics Education Fact Sheet on MTHFR
https://www.genetics.edu.au/health-professionals/FS64-MTHFR-GENE-TESTING-FOR-PATIENTS.pdf

Appendix 7- **Consanguinity Information**

Centre for Genetics Education Fact Sheet on Consanguinity
https://www.genetics.edu.au/publications-and-resources/facts-sheets/fact-sheet-18-when-parents-are-relatives-consanguinity/view

Appendix 8- **Teratogen exposure information**

Mothersafe Statewide Service
https://www.seslhd.health.nsw.gov.au/royal-hospital-for-women/services-clinics/directory/mothersafe