Nepean Hospital Clinical Genetics Referral Criteria

Nepean Hospital provides a diagnostic and genetic counselling service for patients in the Nepean Blue Mountains Local Health District. We are not a management service for genetic conditions.

Mandatory referral information

Please **complete our referral form** to ensure all relevant information is included. **Referrals with insufficient information will be returned to the referring doctor until further information is provided to the clinic.**

Urgent referrals:
Contact Clinical Genetics (02) 4734 3362 or via Nepean Hospital Switchboard (02) 4734 2000 and ask for the on-call Geneticist or Genetic Counsellor.

Urgent referrals are accepted for;

1. Patients who are currently pregnant or undergoing IVF with a genetic concern related to the current pregnancy - clearly indicate EDD/LMP
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 **completed referral form and ALL relevant information** to:
Dr Ingrid Sinnerbrink and Associates
Department of Clinical Genetics
Nepean Hospital
NBMLHD-Genetics@health.nsw.gov.au

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.

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 47343362

NSW GOVERNMENT Health
Nepean Blue Mountains Local Health District

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Indications for referral

Reason for referral	When to Refer
Developmental Delay/ Congenital anomalies **Paediatric Whole Exome Testing**	Anyone with a rare genetic or chromosomal diagnosis, congenital anomalies and/or significant developmental delay (mild, moderate, severe ID). Please attach **copies of ALL completed baseline investigations, including where indicated:** • chromosome microarray (CMA) **including completed parental CMAs where indicated** • Fragile X (FraX) • Urine Metabolic Screen (UMS) • CK, lactate, thyroid function tests • transferrin isoforms, long chain fatty acids • Other relevant investigations as per clinical phenotype eg X-ray, ultrasound, MRI etc. • Please send pathology investigations to the Children's Hospital at Westmead for testing where possible - this will assist with potential future genetic investigations if indicated **Referrals for children will only be accepted from Paediatric Specialists, and requires referral of BOTH PARENTS (where possible).** **Paediatric Whole Exome Testing** Follow the NSW Health education resource for paediatricians regarding genomic sequencing in childhood for guidance before contacting us - https://www.genetics.edu.au/health-professionals/genomics-1/intellectual-disability-genomic-testing
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. **Please refer both partners where possible with all relevant information/investigations.**
Thalassaemia/ Haemoglobinopathy	Haemoglobinopathy screening in at risk populations (refer to RACGP guidelines) should be first investigated with FBC, HbEPG, Iron studies in both partners, then discuss/refer to Haematology, and referral to Genetics if indicated.
Pregnancy Planning	Anyone with a personal and/or family history of a genetic condition seeking updated information and options including prenatal diagnosis and Preimplantation Genetic Diagnosis (PGD)/IVF for identified genetic risk. **Attach all relevant information and refer both partners where possible.**

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Indications for referral

Reason for referral	When to Refer
Predictive genetic testing	Predictive testing for inherited disorders in unaffected individuals with a family history. Please enclose all available family history information.
Connective Tissue Disorder / Hypermobility	Anyone with a personal history of the following **red flag complications,** or a family history of these in the presence of hypermobility (Beighton score >4 in adults, >6 in children) should be referred for genetics. **Attach all relevant investigations/correspondence/documentation confirming these red flag complications**: • Young onset of thoracic aortic enlargement, vascular dissection or extensive varicosities • Ectopia lentis • Extensive widened atrophic scars and poor wound healing/ recurrent large hernias • Severe scoliosis • Personal/family history of organ rupture • Recurrent pneumothoraces **Referrals will not be accepted without completed cardiology assessment and echocardiogram attached to referral, plus Ophthalmology assessment if indicated.** **Hypermobility alone without the above is not an indication for genetics referral:** Further details including management see our exclusion criteria
Suspected Marfan syndrome	**Referrals will not be accepted without completed investigations** attached to referral: • cardiology assessment and echocardiogram • Ophthalmology review • chromosome microarray, UMS Please include description of all relevant clinical features on referral, including any additional medical imaging completed.

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The Nepean Hospital Genetics Clinic is unable to provide a service for the following:

Concern	Exclusion criteria/Alternative options
Ehlers Danlos syndrome (EDS) type 3, hypermobility, joint laxity	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. **Nepean Clinical Genetics service is not able to provide treatment or ongoing management or surveillance** for individuals with a personal and/or family history of Ehlers Danlos syndrome type 3/hypermobility/joint laxity WITHOUT red flags – **see indications for genetics referral** 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, **therefore genetics appointment for hypermobility, without significant red flags, is not indicated**. https://ehlers-danlos.com/wp-content/uploads/hEDS-Dx-Criteria-checklist-1.pdf Referral is recommended to relevant medical specialists- paediatrician for children, rheumatologist, rehabilitation physician, pain physician and allied health professionals physiotherapist and occupational therapists. Families might find this useful: https://www.schn.health.nsw.gov.au/fact-sheets/joint-hypermobility Further information for management: www.rcgp.org.uk/clinical-and-research/resources/toolkits/ehlers-danlos-syndromes-toolkit.aspx
Autism Spectrum Disorder (ASD) – without Intellectual Disability (ID)	Children/adults with autism WITHOUT intellectual disability, family history of a genetic condition/ID or unusual facial features do not meet our referral criteria. Baseline investigations should be performed by the managing doctor, including **chromosome microarray, fragile X and urine metabolic screen**. **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/autism-spectrum-disorder

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Diagnosis	Exclusion criteria/Alternative options
Fetal Alcohol syndrome (FAS)	Please refer directly to CICADA Centre NSW – Care and Intervention for Children and Adolescents affected by Drugs and Alcohol. **Phone:** 02 9845 2246 **Fax:** 02 9845 2517 **SCHN-CICADA@health.nsw.gov.au**
Variants of uncertain significance (VOUS) on CMA	Variants of uncertain significance on chromosomal microarray (CMA) that have no gene content, or isolated loss of heterozygosity
Carriers of rare genetic conditions	Individuals who have a population risk of lower than 1 in 50 of being a carrier for a rare autosomal recessive disorder, where their partner is a known carrier of a rare autosomal recessive disorder. Consider reproductive carrier screening – see Centre for Genetic Education fact sheet
Increased risk NIPT or nuchal translucency screening	Pregnant women with a high risk due to advanced maternal age or first trimester screening investigations, who have not yet had a diagnostic test. For increased risk NIPT, please arrange genetic counselling through NIPT provider – instructions usually provided with high risk results to referring doctor.
Recurrent miscarriages for unknown/obstetric reasons	Couples who have had recurrent miscarriages where the cause is NOT due to a chromosomal anomaly. Conventional karyotype should be performed for **both parents** by managing doctor, and referral to genetics with results if indicated. Consider reproductive carrier screening – see CGE fact sheet.
Common genetic changes	Individuals who have had or are considering genetic testing of the MTHFR gene, hemochromatosis, Alpha-1-antitrypsin testing. Management for these conditions is via a GP, with fact sheets available: Centre for Genetic Education, RACGP Genomics in General Practice
Consanguinity	Partners who are related, in absence of family or personal history of genetic condition do not require genetic consultation. See Centre for Genetic Education (CGE) Fact Sheet on Consanguinity. Also consider: haemoglobinopathy screen of both partners for increased risk populations and reproductive carrier screening – see CGE fact sheet.

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The Nepean Hospital Genetics Clinic is unable to provide a service for the following:

Diagnosis	Exclusion criteria/Alternative options
Direct to consumer testing	Individuals who have had or are considering 'direct to consumer' genetic testing with no identified significant family or personal history.
Teratogen exposure, medications in pregnancy	Contact Mothersafe for advice, Phone: 9382 6539
Paternity/Ancestry testing	Not offered by Clinical Genetics, private testing available.
Cancer genetic testing	Please refer NBMLHD patients requiring genetic counselling for familial cancer to: **Familial Cancer Service** **The Crown Princess Mary Cancer Centre Westmead** Phone: 8890 6947, Fax: 8890 9217, Email: WestmeadFCS@health.nsw.gov.au https://www.sydneywestcancer.org/patient-care/genetic-risk/
Out of area referrals	Referrals are accepted for residents of NBMLHD. Check if the home address is within NBMLHD here: https://www.health.nsw.gov.au/lhd/Pages/lhd-maps.aspx Out of area referrals may be accepted in the following circumstances, if documented explicitly on referral: • Resident of other Local Health District that does not provide the clinical service e.g. rural, outer metro • Continuing care of a patient previously seen by NBMLHD • Demonstrated complexity requiring services of Nepean Hospital • Compassionate circumstances (e.g. family proximity, staff)