

ORGANISATIONAL MODEL OF CARE

# Clinical genomics model of care

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The information is not a substitute for healthcare providers' professional judgement.

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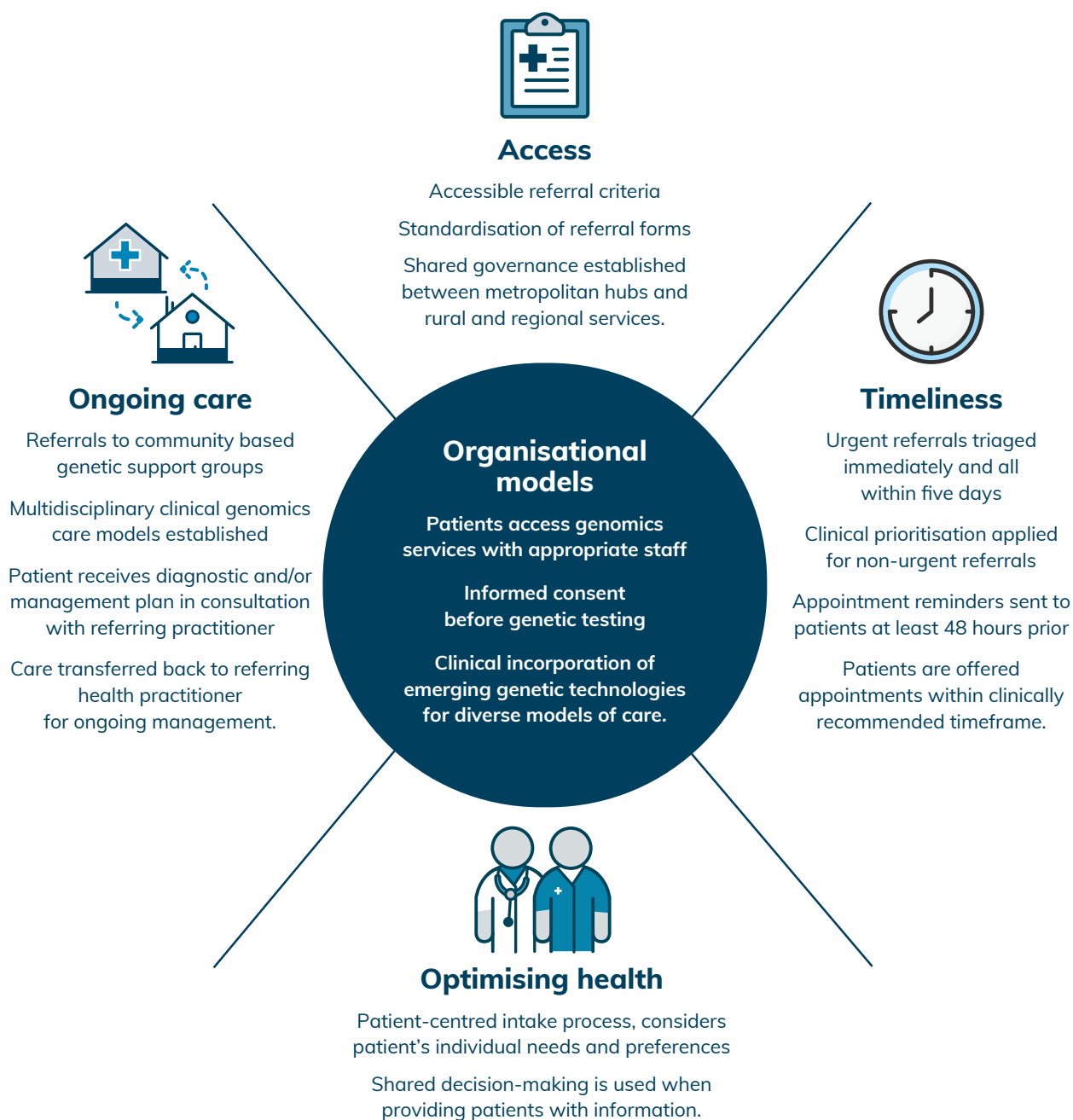
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# Clinical genomics model of care – at a glance

The clinical genomics model of care outlines best practice to support equitable access by patients to NSW genomics services. It identifies the four clinical priority areas to ensure that patients receive the right services at the right time.



# Clinical genomics model of care

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## Introduction

The Clinical genomics model of care outlines best practice to support equitable access to clinical genomics services by patients in NSW. It identifies clinical priority areas to ensure that patients receive the right service at the right time.

NSW clinical genomics services are generally delivered via an outpatient setting in NSW public hospitals. Services are provided to paediatric and adult patients, predominantly delivered across three areas; general genetics, cancer genetics and metabolic genetics. Clinical genomics specialty services are an emerging and growing field and are predicted to be one of the drivers of demand for clinical genomics services into the future.

Over the last two decades, NSW Health clinical genomics services have grown from a boutique sub-specialty into a mainstream clinical specialty, with matching increases in patient demand. The application of genetic and genomic medicine during this time has seen major advances in the understanding of the human genome and the association between genetic variation and disease.

Rare diseases are a condition, syndrome or disorder that affects less than 5-7 individuals in 10,000.<sup>1,2,3</sup> Rare diseases affect over 640,000 people in NSW. There are around 7,000 different types of rare diseases, with approximately five new rare diseases being described in the medical literature each week and 80% of these rare diseases are considered genetic.<sup>4,5</sup>

In addition, with advances in cancer genetics, metabolic genetics and other sub-specialty areas, there is the possibility of over 510,000 people in NSW with a rare genetic condition requiring access to NSW clinical genomic services at some point in their life.<sup>6</sup>

In NSW, patients may wait to enter some services for up to, and over, 12 months. This model of care aims to address these access issues to improve service delivery across the four identified clinical priority areas.

At least **7,000** rare diseases and **5** new ones described in the medical literature each week

**70** million people world wide are collectively affected by **rare diseases**



Over **500,000** people in NSW are living with **rare genetic conditions**



Approximately **2** million people in Australia are living with **a rare disease**



**80%** of rare diseases are **considered genetic**

## Background

This model of care has been developed using the Agency for Clinical Innovation's (ACI) redesign methodology.<sup>7</sup> This methodology has been used to get an understanding of the issues impacting on delivery of clinical genomics in NSW from differing perspectives; agree on solutions to improve patient care; and find new ways of working that are scalable across NSW.

The diagnostic phase was conducted over a six-month period from January to June 2019. In NSW, 83% of clinical genomics services (29 of 35 services) were visited to understand the current state of service provision. Three questions were asked of each service:

1. What does the patient journey look like through your service? (the current state)
2. What are the main issues you are currently facing within your service? (issues identification)
3. What would your service look like in the perfect world? (the future state).

The current state for clinical genomics services is a fragmented delivery of services, compounded by increasing patient demand and an increase in patients waiting more than 365 days to access a service.

The identified future state is cohesive delivery of clinical genomics services where patients have equitable access and receive the right service at the right time. This would be through delivery of a best practice clinical genomics model of care.

A summary of issues identified in the diagnostic phase is provided below:

- **Patients with multiple problems of a rare and complex nature** – one hour per patient appointment equates to a minimum of four hours of work up and follow up i.e. researching diagnosis, utility of diagnostic pathways, options for genetic testing, chasing results
- **Service capacity not meeting patient demand** – availability of clinic appointments and services is not fulfilling the number of patients entering some services, resulting in long waitlists to access services
- **Competing priorities** – not all data is captured due to increased complex patient demand; clinicians' focus on clinical workload; administrative tasks completed when able via intermittent administration workforce
- **Genetic testing** – large amounts of clinical time spent determining which laboratories tests should be sent to; factors to be considered include turnaround time, interpretation ability and cost
- **Limited physical space** – excessive amounts of satellite files are stored within genetics departments mainly due to conditions set out in the NSW Health Privacy Manual for Health Information<sup>8</sup>
- **Volume of regulation** – large and varied amounts of legislation, policies and guidelines guiding genetics services.

This model of care will address the first three points. The remaining issues are being addressed by the recommendations of the NSW Health Genomics Strategy (the Strategy)<sup>9</sup>. The Strategy articulates a shared vision for NSW genomics promoting collaboration and the development and use of appropriate genomic technologies in healthcare and public health for the benefit of the NSW population.

# Clinical priority: Access

## Patient has equitable access to NSW clinical genomics services

There is often an imbalance between the available capacity of a service and the ability to meet demand within the community.

Referrers should understand the clinical genomics referral criteria and what options are available when a patient does not meet the criteria. Patients should be advised of and supported through alternative pathways when their needs cannot be met through the clinical genomics service.

Referral pathways that support primary care providers to make referrals for patients to access clinical genomics services are recommended. A clinical genomics services referral criteria and information template is available in the Clinical genomics model of care toolkit.

Clinical genomics services should have a process to capture the referral information and ensure that service providers can easily access the referral form.

A clinical genomics services referral form template is available in the Clinical genomics model of care toolkit.

Where clinical genomics services are provided through a cooperative arrangement between facilities, or local health districts (LHDs) and specialty health networks (SHNs), an agreement should be in place to support the governance of clinical and administrative functions. Rural and regional clinical genomics services are predominantly delivered in a cooperative arrangement with metropolitan hubs and need agreements that support shared governance and oversight.

### Key components

Clinical genomics services should aim to achieve the following key components.

#### Referral criteria

Primary care, medical specialists and patients can easily access referral criteria via the hospital websites and the NSW Health Centre for Genetics Education website<sup>10</sup>. This will support patients being referred to the right service, at the right time.

#### Standard referral form

A standardised referral form is used and is available on the hospital's website to support rapid access for urgent patients and provide timely access for non-urgent patients.

#### Governance

Shared governance and oversight is established between metropolitan hubs and their satellite services and is reviewed on a yearly basis. This will support patients having equitable access to NSW clinical genomics services regardless of location.



## Clinician and patient stories (experiential evidence)

### Royal North Shore Clinical Genetics Service – referral criteria

#### Situation

Royal North Shore Clinical Genetics Service was experiencing a steady increase in patients and their families being referred, that did not necessarily require genetics input. These patients and families were experiencing diagnostic fatigue, the expectation was a genetic test and diagnosis could be the answer to their concerns. Management of these expectations formed a large portion of the consultation with further referrals to appropriate services to manage their condition.

#### Action

In 2019, Royal North Shore Clinical Genetics Service sought to communicate their core business and alternative pathways by targeting their major referrers (GPs and subspecialists). They did this through establishment of a referral criteria document that was also translated into a HealthPathway (for GPs) and used as the basis for information about their clinical genomics services on their website.

#### Outcome

A steady decline in referrals for patients that did not necessarily require genetics input. GPs and subspecialists now have the information to be able to refer patients and their families to the right service the first time.

### Prince of Wales Familial Cancer Centre – cooperative arrangement governance

#### Situation

Familial Cancer Service across NSW are presently providing patient care to 12 satellite services from six metropolitan hubs. The hubs and their satellite services provide care in a cooperative arrangement between facilities, districts and networks. Not all have agreements in place to support governance.

#### Action

Prince of Wales (POW) Familial Cancer Centre is the hub for satellite services provided in hereditary cancer centres at St George, Wollongong, Sydney Children's Hospital, ACT and Hunter New England Paediatric Service, and the Royal Hospital for Women High Risk Breast Clinic and Chemotherapy Suite Clinics. These clinical services are provided to other organisations and are included within 2019/20 South Eastern Sydney Local Health District Service Agreement. Memorandums of understanding were also established to support governance of clinical and administrative functions.

#### Outcome

Patients can access clinical genomics services regardless of their location; and also access the specialised expertise of the clinicians from POW Familial Cancer Centre that is not able to be provided elsewhere.

## Tools

- Service directory ([Appendix 1](#))
- Clinical genomics services referral criteria and information template
- Clinical genomics services referral form template
- Family history questionnaire template.



# Clinical priority: Timeliness

## Patient has timely access to NSW genomics services

Clinical genomics services should ensure patients are prioritised and seen within clinically appropriate timeframes. It is recommended that clinical genomics services have mechanisms in place to manage service availability and capacity to see the patient within clinically appropriate timeframes.

Managing service availability and capacity includes:

- supporting non-genetic health professionals to order genetic tests
- recommending alternative pathways to clinical genomics services to a referrer, if this is more appropriate.

Patients' needs and circumstances should be considered when allocating appointments to increase patient satisfaction and decrease cancellation and no-show rates. Prioritisation and appointment allocation should be consistent with the NSW Outpatient Services Framework<sup>11</sup>.

### Key components

Clinical genomics services should aim to achieve the following key components.

#### On receipt of referral:

##### Referral triage

Urgent referrals immediately triaged to a health practitioner.

Non-urgent referrals triaged within five days by delegated staff member either for registration; redirection; or return to referring clinician and entry into a referral register.

##### Capacity and demand management

Acceptance of non-urgent referrals aligns to the service availability and capacity to see the patient within the clinically appropriate timeframe.

Clinical referral criteria reflects core business and may change depending on service capacity.

Clinical governance records maintained (i.e. waitlist timeframes) and reporting is consistent with local frameworks.

##### Appointment timeframe notification

Non-urgent referrals are booked an appointment or added to an appointment list (waitlist). Patient and referrer notified of expected timeframes for appointment.

#### Appointment allocation:

##### Urgent appointment allocation

Urgent appointments are booked immediately in consultation with patient.

##### Non-urgent appointment allocation

Non-urgent appointments are allocated in consultation with the patient, based on clinical priority and offered  $\geq 10$  working days prior to the appointment.

##### Appointment reminder

Appointment reminders sent to patients at least 48 hours prior to clinic attendance.

##### Attendance at appointment

Patients attend appointments within clinically recommended timeframes.

## Clinician and patient stories (experiential evidence)

### Nepean Genetics Service – incomplete non-urgent referrals

#### Situation

Nepean Genetics Service was receiving large volumes of non-urgent referrals that contained minimal information and required clinical staff to phone the referring doctors to complete the information required to prioritise and manage the referral.

#### Action

Nepean Genetics Service implemented a simple solution of issuing an incomplete referral letter outlining the required information which was sent to the referrer so the patient could be prioritised and assessed appropriately by the clinical team.

#### Outcome

This simple process has resulted in improved efficiency on multiple levels along with improved patient care. There has been a significant decrease in inappropriate referrals. Both referrers and patients have been engaged in the process resulting in referrers (GPs and subspecialists) being upskilled, with electronic material and links to websites, and patients being empowered to follow-up with their own healthcare. Subsequently, the genetic counsellors are spending less time chasing information on inappropriate referrals.

### Westmead Familial Cancer Service – family history questionnaire

#### Situation

Westmead Familial Cancer Service developed a family history questionnaire (FHQ) and patient registration form that is mailed to non-urgent patients who do not need treatment focussed genetic testing and meet certain criteria that deems them a low risk referral. The prior process was all conducted via time consuming phone calls.

#### Action

The FHQ information is recorded by the clinical team in TrakGene and reviewed by a genetic counsellor prior to scheduling an appointment. If the FHQ is not returned within two months a reminder letter is sent. If there is still no response after an additional month, a final follow up phone call is made to the patient. Patients who fail to respond are sent a standard discharge letter with a copy sent to referring doctor.

#### Outcome

The FHQ and patient registration process has reduced the amount of time clinicians spend on phone calls and has resulted in increased efficiencies in triaging and seeing urgent patients. Overall this has resulted in 80% of referrals successfully processed and 20% of non-contactable patients discharged from the service without an appointment.

## Tools

- Clinical genomics prioritisation categories ([Appendix 2](#))
- Incomplete referral – request for further information sample template
- Acceptance or non-acceptance of non-urgent referral sample template
- Appointment confirmation sample template
- Patient appointment list audit letter sample template



# Clinical priority: Optimising health

## Patient, family and carers have the right information to inform decision-making

NSW clinical genomics services should support a patient and family-centred approach to deliver care to patients. Research demonstrates that patient-centred care improves the patient experience and creates public value for services.<sup>12, 13</sup> Family-centred care is an integral component of clinical genomics services, as care is delivered to not only the patient but also their family (or genetic relatives).

NSW clinical genomics services should use a shared decision-making model when providing patients with information, including patient consent to genetic testing. 60% of people in Australia have low health literacy, meaning that they do not have the knowledge to find, understand and use information about their health and healthcare.<sup>14</sup> The health literacy of patients should be considered to ensure that they can understand and use information provided to make informed decisions.<sup>15, 16, 17</sup>

### Key components

Clinical genomics services should aim to achieve the following key components.

#### Patient and family-centred care

NSW clinical genomics services use a patient and family-centred approach to delivering care. The clinical genomics team has conversations with the patient that considers the individual needs and values of that patient and their family.

Options include:

- patient intake process (clinician to patient consultation, optimally via phone or telehealth prior to appointment)
- clinician to clinician intake process (genetic counsellor to geneticist consultation, optimally in-person or via phone or telehealth)
- team intake process (genetic services team consultation, optimally in-person or via telehealth).

#### Shared decision-making

NSW clinical genomics services use a shared decision-making model when providing patients with information to make a decision.

#### Patient consent and genetic testing

Patient consent is received if genetic testing is to be undertaken. The patient is appropriately consented using the NSW Health Genetic Testing Consent Form, where required.

## Clinician and patient stories (experiential evidence)

### Cancer Institute NSW – telehealth project

#### Situation

Two familial cancer genetics services were experiencing an increase in demand to access their services. With telehealth being a key enabler to improving access to care, the teams developed telehealth workflows to improve access, reduce travel and associated costs with accessing the service.

#### Action

The familial cancer genetics teams with the support of the LHD telehealth managers or leads and Cancer Institute NSW developed and implemented clear telehealth workflows that improved access to specialist cancer genetics clinicians.

#### Outcome

Over the project period (nine months), the familial cancer genetics teams developed and implemented workflows using telehealth as the modality of service delivery. As a result, patients avoided 5,248km in travel (approximately 66.4 hours) during the first three months of implementation. These workflows will be made available to clinicians and managers of cancer services to further enhance and develop their processes.

### Tools

**Clinical Genomics Resources:** An overview of genomic information for consumers and non-genomic health professionals to direct NSW Health clinicians to appropriate high-quality clinical genomic information for patients, consumers, general practitioners and non-genomic health practitioners.



## Clinical priority: Ongoing care

### Patient has ongoing, supportive care

To support the ongoing care of patients and their families, communication between the clinical genomics service and the referring health practitioner and GP is integral.

#### Key components

Clinical genomics services should aim to achieve the following key components.

#### Support services

Referral to community based genetic support groups for peer support and extended information for individuals and families affected by genetic conditions.

#### Management plan and discharge

Patient receives diagnostic and/or management plan in consultation with referring health practitioner. Care is transferred from clinical genomics services back to referring health practitioner for ongoing management.

# Organisational models: Clinical genomics care delivery models

## Patients have the right care delivered at the right place by the right person

Clinical genomics services have many different care models available to deliver patient and family-centred care; either in-person or via telehealth.

Models available include:

- traditional models where the patient navigates individual specialty clinics i.e. patient attends a clinical genomics outpatient clinic appointment
- multidisciplinary models where individual specialties are navigated for the patient i.e. patient attends an outpatient clinic and the clinical genomics consult is one component of that service

- innovative models that incorporate emerging genetics technologies e.g. specialty services such as neurofibromatosis service at Westmead Children's Hospital and Royal North Shore.

Clinical genomics specialty services are predicted to be one of the drivers of demand for clinical genomics services into the future. Capacity and demand management is essential for long term sustainability and maintaining patient access.

## Key components

Clinical genomics services should aim to achieve the following key components.

### Clinical genomics care delivery models

A range of options are available to ensure a patient attends an appropriate service with the appropriate staff. These include:

- rapid access clinics and services for urgent patients
- genetic counsellor clinics
- combined genetic counsellor and genetic specialist clinics
- genetic specialist diagnostic clinics
- telehealth and virtual services
- outreach services.

### Clinical genomics care in the community

Clinical genomics care in the community provides options for patients and improves service capacity (e.g. NSW BRCA Screening Program and Jewish Screening Program).

### Multidisciplinary clinical genomics care models

Multidisciplinary clinical genomics care models established to optimise patient care:

- clinical genomics services integrated into subspecialty clinics
- subspecialty services integrated into clinical genomics clinics
- consultative input by clinical genomics into subspecialty services.

### Innovation to achieve better patient care outcomes

Clinical incorporation of emerging genetic technologies for diverse models of care i.e. clinical genomics specialty services.

## Clinician and patient stories (experiential evidence)

### Royal North Shore Genetics Service and Westmead Children's Genetics Service – Neurofibromatosis (NF) Service

#### Situation

Royal North Shore Hospital (RNSH) and The Children's Hospital at Westmead (CHW) provide Specialist Hub Services (the Hub) for NF. Depending on where the patient lives, staff at the Hub may liaise with local hospitals or doctors for ongoing care such as imaging, surgery or chemotherapy, to enable patients to be cared for as close to home as possible, but under the guidance of the Hub.

#### Action

Doctors refer patients to RNSH or CHW Specialist Hub services, including consultation and imaging. This provides a flexible, virtual spoke model.

#### Outcome

Patients accessing services for NF via the Hub have gained access to life saving and disease modifying treatments delivered as a comprehensive service across NSW. Improvements have been identified in coordination of care; patient and clinician experience; consistency in delivery of care for NF patients across NSW; improved treatment of disfigurement in NF; and improved patient access to new clinical therapeutic and testing approaches.

### The model of care project for genomics of intellectual disability

#### Situation

In May 2020, Medicare provided new item numbers to allow specialist paediatricians to order a diagnostic exome in children under ten with a moderate to severe intellectual disability or a likely monogenic syndrome. The intention was to allow equitable and timely access to testing that may provide families with a diagnosis, improve patient care and influence future reproductive decision-making in these families. A model of care project started concurrently with the intention to understand weaknesses in the existing system and strategies to allow optimal utilisation of this item number.

#### Action

The model of care project has:

1. Explored the preparedness of paediatricians to deliver this service, their confidence and knowledge gaps.
2. Created support and information for clinicians to facilitate access to testing.
3. Carried out online education webinars and in person training for paediatricians within target sites.
4. Trialled critical infrastructure and system improvements to support paediatrician ordering.
5. Observed the success of local strategies to support education and paediatric ordering.

#### Outcome

A final report will be delivered with a model of care and recommendations at the end of October 2021.



## **St Vincent's Clinical Genomics – Neurogenomics model of care**

### **Situation**

St Vincent's Clinical Genomics (SVCG) is a tertiary referral centre within St Vincent's Health Network. SVCG has been receiving complex neurological referrals for patients from across Australia including regional and remote areas. It was recognised that many of these patients, who often had to travel long distances, would benefit from seeing neurologists, geneticists and genetic counsellors in the same clinic setting.

### **Action**

A Neurogenomics Model was established in 2017. This service aims to provide patient-centred care, whereby each patient is assessed by neurologist(s) with different subspecialty interests, including movement disorders, dystonia, and ataxia, as well as a geneticist, jointly in the same clinic room, supported by a genetic counsellor.

Decisions regarding genetic and genomic testing are made jointly at point of care, and management advice given by neurologists. Patients are then seen, counselled and consented by a genetic counsellor.

### **Outcome**

Currently three neurologists, each with different subspecialty interests, attend the clinic on a regular basis. This has provided high-level specialised patient-centred care and expertise that regional and remote patients would otherwise not be able to access; has achieved high diagnostic yield of 36.7% and provided genomics teaching and training opportunities to neurology trainees.

## Appendix 1: Service directory

The **NSW Health Centre for Genetics Education website** houses the clinical genomics service directory for general and specialist clinical services.

Search if the home address is within local health district (LHD) with the **NSW Health LHD map**.

Prenatal specialised services focused on providing information and support regarding testing and screening during pregnancy are provided by most local health districts through maternal fetal medicine units and genetic services as appropriate.

Patients from Albury are referred to Victoria services, patients from Broken Hill are referred to South Australia services. Patients from the Tweed area for prenatal and fetal medicine services are referred to Queensland services.

The following table displays general and cancer genetic services by LHD and specialty health network (SHN). It includes differentiation of metropolitan centres and outreach services, rural and regional genetic counsellor led services.

LHDs and SHNs							
Hunter New England LHD (HNELHD)	Nepean Blue Mountain LHD (NBMLHD)	Northern Sydney LHD (NSLHD)	South East Sydney LHD (SESLHD) and Sydney Children's Hospital Network (SCHN) – Randwick	Western Sydney LHD (WSLHD) and SCHN - Westmead	South West Sydney LHD (SWSLHD)	Sydney LHD (SYDLHD)	St Vincent's Hospital Network (SVHN)
Outreach within LHD and to Mid North Coast LHD (MNCLHD) and Northern NSW LHD (NNSWLHD)	Nil Outreach	Outreach to Central Coast LHD (CCLHD)	Outreach to Illawarra Shoalhaven Local Health District (ISLHD)	Outreach to Western NSW LHD (WNSWLHD) and Murrumbidgee LHD	Outreach within LHD and to Murrumbidgee LHD and Southern NSW LHD (SNSWLHD)	Outreach within LHD	Cancer Outreach to CCLHD (Gosford)
General							
Hunter Genetics	Nepean Genetics Services	Royal North Shore Hospital (RNSH) Genetics Service	SES Centre for Clinical Genetics SCHN Randwick - Paediatric St George Genetics Service and Royal Hospital for Women (RHW)	Western Sydney Genetics Program SCHN Westmead Paediatric Westmead Hospital Department Genetic Medicine Adults and Neonatal intensive care unit (NICU)	Liverpool Genetics Services	Royal Prince Alfred Hospital Clinical Genetics Service	St Vincent's Clinical Genomics Services
General outreach							
Tamworth Taree/Forster Coffs Harbour Port Macquarie/ Kempsey Kingscliff	Gosford	Wollongong	Dubbo Bathurst/Orange	Wagga Goulburn, Moruya, Merimbula and Queanbeyan			

LHDs and SHNs							
Hunter New England LHD (HNELHD)	Nepean Blue Mountain LHD (NBMLHD)	Northern Sydney LHD (NSLHD)	South East Sydney LHD (SESLHD) and Sydney Children's Hospital Network (SCHN) – Randwick	Western Sydney LHD (WSLHD) and SCHN - Westmead	South West Sydney LHD (SWSLHD)	Sydney LHD (SYDLHD)	St Vincent's Hospital Network (SVHN)
Outreach within LHD and to Mid North Coast LHD (MNCLHD) and Northern NSW LHD (NNSWLHD)	Nil outreach	Outreach to Central Coast LHD (CCLHD)	Outreach to Illawarra Shoalhaven Local Health District (ISLHD)	Outreach to Western NSW LHD (WNSWLHD) and Murrumbidgee LHD	Outreach within LHD and to Murrumbidgee LHD and Southern NSW LHD (SNSWLHD)	Outreach within LHD	Cancer outreach to CCLHD (Gosford)
Cancer services							
Hunter Family Cancer Service	Patient referrals to Familial Cancer Service Westmead	RNSH Familial Cancer Service	Prince of Wales Hereditary Cancer Centre	Familial Cancer Service Westmead Hospital	SWSLHD Cancer Genetics Service	SYDLHD Cancer Genetics Service	St Vincent's Hereditary Cancer Clinic
Cancer outreach							
Tamworth Taree/Forster Coffs Harbour Port Macquarie/ Kempsey Kingscliff			Kogarah Wollongong ACT Outreach Service	Wagga Dubbo			Gosford Goulburn, Queanbeyan, Pambula and Moruya
Speciality							
Genetics of Learning Disability (GOLD) – Hunter Genetics and RNSH Neurofibromatosis - RNSH and SCHN-Westmead Maternal Fetal Mothersafe – RHW Metabolic Services – WSLHD Westmead Hospital Adults and SCHN Paediatrics							

## Appendix 2: Clinical genomics prioritisation category

Category	Minimum referral criteria	Classification
<b>Urgent</b>	<p>Actioned within one week optimal for:</p> <ul style="list-style-type: none"> <li>pregnant patients</li> <li>cancer patients scheduled for surgery or therapies where genetic testing will be required for immediate surgical or treatment options</li> <li>patients where results of the genetic consultation are required for urgent medical management decisions.</li> </ul>	Urgent
<b>Category A</b>	<p>Appointment scheduled within 30 days for conditions that may deteriorate if assessment is delayed, optimal for patients with:</p> <ul style="list-style-type: none"> <li>a personal history of a genetic diagnosis and is currently on or about to go onto a palliative care pathway</li> <li>any child under 12 months of age.</li> </ul>	Non-urgent and ready for appointment
<b>Category B</b>	<p>Appointment scheduled within 90 days optimal for patients with conditions that may require more complex care if assessment is delayed:</p> <ul style="list-style-type: none"> <li>a personal or family history (genetic relatives) of a confirmed genetic diagnosis</li> <li>a personal or family history (genetic relatives) of a genetic diagnosis, where a specific gene mutation has been identified on a genetic test</li> <li>deteriorating with a genetic disorder</li> <li>child affected by a neurodegenerative genetic condition</li> <li>preconception counselling of couples who are at an increased risk of having children with a genetic condition because of a personal and or any family history (genetic relatives) of a known or suspected genetic condition and or consanguinity</li> <li>any child under 6 years of age.</li> </ul>	Non-urgent and ready for appointment
<b>Category C</b>	<p>Appointment scheduled within 365 days for conditions unlikely to deteriorate quickly, optimal for patients with:</p> <ul style="list-style-type: none"> <li>A personal and or any family history (genetic relatives) of a genetic diagnosis, where a specific gene mutation has not been identified on a genetic test.</li> </ul>	Non-urgent and ready for appointment

Source: Created by Agency for Clinical Innovation, Clinical Genetics Network, Clinical Genetic Executive Committee and adapted from GL2019\_011 Outpatients Services Framework and PD2012\_011 Waiting Time and Elective Surgery Policy.

## References

1. Therapeutic Goods Administration. Orphan drugs program discussion paper. [Internet]. Canberra: TGA; 2015 [cited 26 November 2020]. Available from: <https://www.tga.gov.au/sites/default/files/consultation-orphan-drugs-program.pdf>
2. Orphanet. Search for a rare disease. [Internet]. Paris: Orphanet; 2021 [updated 15 May 2021; cited 3 June 2021]. Available from: <https://www.orpha.net/consor/cgi-bin/Disease.php?lng=EN>
3. The Orphan Drug Act 1983 (USA) 21CFR Part 316.
4. Auvin S, Irwin J, Abi-Aad P; et al. The problem of rarity: Estimation of prevalence in rare disease. *Value Health*. 2018;21(5):501–507.
5. Rare Voices Australia. What is a rare disease? [Internet]. Mentone (VIC): Rare Voices Australia; 2021 [cited 7 December 2021]. Available from: <https://rarevoices.org.au/what-is-a-rare-disease/>
6. Australian Bureau of Statistics. National, state and territory population [Internet]. Canberra (ACT): Australian Bureau of Statistic; 18 March 2021 [cited 3 June 2021]. Available from: <https://www.abs.gov.au/statistics/people/population/national-state-and-territory-population/latest-release>
7. NSW Agency for Clinical Innovation. Redesign: A methodology for improvement and innovation [Internet]. Sydney: ACI; 2021 [cited 19 May 2021]. Available at: <https://aci.health.nsw.gov.au/resources/redesign/redesign-factsheets/redesign-methodology-for-improvement-and-innovation>
8. NSW Ministry for Health. Privacy manual for health information [Internet]. Sydney: NSW Health; 2015 [cited 3 December 2020]. Available from: <https://www.health.nsw.gov.au/policies/manuals/Documents/privacy-manual-for-health-information.pdf>
9. NSW Ministry of Health. NSW Health genomics strategy [Internet]. Sydney; NSW Health: 2017 [cited 3 June 2021]. Available from: <https://www.health.nsw.gov.au/services/Publications/nsw-health-genomics-strategy.pdf>
10. Centre for Genetics Education. Sydney: NSW Health; 2021 [cited 7 June 2021]. Available from: <https://www.genetics.edu.au/>
11. NSW Ministry of Health. Outpatient services framework: Guideline. GL2019\_011 [Internet]. Sydney: NSW Health; 2019 [cited 3 June 2021]. Available from: [https://www1.health.nsw.gov.au/PDS/ACTIVEPDSDOCUMENTS/GL2019\\_011.PDF](https://www1.health.nsw.gov.au/PDS/ACTIVEPDSDOCUMENTS/GL2019_011.PDF)
12. Australian Commission on Safety and Quality in Healthcare. Patient-centred care: Improving quality and safety through partnerships with patients and consumers. Sydney: ACSQHC; 2011.
13. Clinical Excellence Commission. Person-centred care. [Internet]. Sydney: CEC; 2021 [cited 9 May 2021]. Available from: <https://www.cec.health.nsw.gov.au/improve-quality/teamwork-culture-pcc/person-centred-care>
14. Australian Commission on Safety and Quality in Healthcare. Health literacy [Internet]. Sydney: ACSQHC; 2019 [cited 3 June 2021]. Available from: <https://www.safetyandquality.gov.au/our-work/patient-and-consumer-centred-care/health-literacy>
15. Department of Health, NSW Health, HealthDirect. NSW health literacy hub [Internet]. 2021 [cited 9 May 2021]. Available from: <https://www.healthliteracyhub.org.au/>
16. Clinical Excellence Commission. Health literacy [Internet]. Sydney: CEC; 2021 [cited 9 December 2021]. Available from: <https://www.cec.health.nsw.gov.au/improve-quality/teamwork-culture-pcc/person-centred-care/health-literacy>
17. NSW Agency for Clinical Innovation. Consumer enablement: A clinician's guide [Internet]. Sydney: ACI; 2021 [cited 9 December 2020]. Available from: <https://aci.health.nsw.gov.au/resources/primary-health/consumer-enablement/guide>
18. NSW Agency for Clinical Innovation. Genetic and genomic testing consent forms [Internet]. Sydney: ACI; 2021 [cited 3 June 2021]. Available from: [https://aci.health.nsw.gov.au/networks/clinical\\_genetics/genetic-and-genomic-testing-consent-forms](https://aci.health.nsw.gov.au/networks/clinical_genetics/genetic-and-genomic-testing-consent-forms)

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## For more information

The [Clinical genomics model of care toolkit](#) includes links to a number of resources, tools and templates to assist with implementing this model of care.

The Agency for Clinical Innovation (ACI) is the lead agency for innovation in clinical care.

We bring consumers, clinicians and healthcare managers together to support the design, assessment and implementation of clinical innovations across the NSW public health system to change the way that care is delivered.

The ACI's clinical networks, institutes and taskforces are chaired by senior clinicians and consumers who have a keen interest and track record in innovative clinical care.

We also work closely with the Ministry of Health and the four other pillars of NSW Health to pilot, scale and spread solutions to healthcare system wide challenges. We seek to improve the care and outcomes for patients by re-designing and transforming the NSW public health system.

Our innovations are:

- person centred
- clinically led
- evidence based
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