Services for Children and Young People

Paediatric and Adult Metabolic Services

Tier 2 Service Specification

September 2024

Health New Zealand
Te Whatu Ora

Contents

1. Status 2
2. Review History 2
3. Introduction 3
   3.1 Background 3
4. Service Definition 4
5. Service objectives 4
   5.1 General 4
   5.2 Māori Health 5
6. Service Users 5
7. Access 5
   7.1 Entry Criteria 5
   7.2 Exit Criteria 5
   7.3 Time 5
8. Service Components 6
   8.1 Assessments and Treatment – Metabolic Management 6
   8.2 Pacific Health 6
   8.3 Other Ethnicities 6
   8.4 Settings 6
   8.5 Service Levels 6
   8.6 Equipment 7
   8.7 Key Inputs 7
9. Service Linkages 7
10. Exclusions 8
11. Quality Requirements 8
   11.1 General 8
   11.2 Safety and Efficiency 8
12. Purchase Units and Reporting Requirements 8
   12.1 Service Planning 9

1. Status

Approved to be used for mandatory nationwide description of services to be provided.

MANDATORY ☑ RECOMMENDED ☒

It is compulsory to use this Specification when purchasing services. No Districts should use a local service specification instead of this mandatory specification.

2. Review History

Review History	Date
Published on NSFL	June 2011
Review: Metabolic Services service specification (2003). Amendments: updated the service specification to meet requirements for national services. PU code definition updated.	May 2011
Amendment to purchase units to align with current purchase unit data dictionary	February 2016
Moved to Health NZ template. Updated links for PUDD and NSFL only. Amended DHB to become District/Region where appropriate. No other changes to content made.	September 2024
Consideration for next Service Specification Review	Within five years

Note: In September 2024 a small programme of work moved all Service Specifications to Health New Zealand branded templates. No amendments were made to the body text or content of the Service Specification, so references to DHB, Ministry of Health or other pre-2022 reforms vocabulary will still exist. A larger programme of work to review and revise all Service Specifications is planned for late 2024 to early 2025.

Note: Contact the NSF Team, Te Whatu Ora | Health New Zealand to discuss proposed amendments to the service specifications and guidance in developing new or updating and revising existing service specifications. NSF@tewhatuora.govt.nz

Nationwide Service Framework Library web site here

3. Introduction

This tier two Paediatric and Adult Metabolic Services service specification (the Service) must be used in conjunction with the overarching tier one Specialist Medical and Surgical Services and the tier one Services for Children and Young People service specifications.

Refer to the overarching tier one service specifications for generic details on:

- Service Objectives
- Service Users
- Access
- Service Components
- Service Linkages
- Exclusions
- Quality Requirements

The above heading sections are applicable to **all** service delivery.

3.1 Background

From 1 July 2011, planning and funding decisions for the Service will be made at a national level because it requires a highly specialised workforce and has a low volume patient group.

The desired outcome of this national approach is for the service to be sustainable, delivered consistently and accessed equitably. The Service was implemented over a three year period by 1 July 2014.

Inborn errors of metabolism (IEM) are genetic diseases caused by a defect in the function of a cellular protein (for example an enzyme or transporter) involved in the metabolism of various chemicals in the body.

Metabolic diseases can be complex to diagnose, can affect multiple organ systems and can present at any age. The work-up, investigation and management of a child with suspected or proven IEM requires a highly specialised physician (a metabolic physician) who remains up to date with current literature and research in this field and works in close co-operation with a team of dietitians, neuro-psychologists, nurse specialists and biochemical genetics and molecular laboratories. It requires close collaboration with the wider community of laboratories and physicians specialising in the diagnosis, management and research of IEMs.

Many disorders are treatable. For example, by reducing the accumulation of toxic substances by adhering to a special diet, by providing high concentrations of a cofactor required to improve or correct enzyme function, or by providing the dysfunctional enzyme itself. Some early diagnoses, sometimes even in-utero or before symptoms begin, can lead to treatment that would be more effective than if it was begun when the condition presented symptomatically.

4. Service Definition

The Service is delivered by the multidisciplinary team that provides tertiary level diagnosis and management, including crisis management of individuals with known or possible complex metabolic diseases.

The Service will provide care for patients, and their families and whānau, with metabolic diseases including a comprehensive consultative service, encompassing the investigation, diagnosis, treatment (including crisis treatment) and referral to genetic counselling for patients with metabolic diseases.

The Service will provide an equitable outreach component, working with teams of local physicians (usually paediatricians), dietitians and other health care professionals in secondary and primary health care who will assist with delivery of ongoing case management.

The Service will include:

- specialist tertiary care and support of metabolic patients
- consultation using a range of modes including tele / video communication technologies, such as: face to face, written, or by telephone
- referral to another specialty for an opinion, and / or management, or shared care
- referral to genetic counselling (including pedigree analysis, risk assessment and testing)
- specialist testing including biochemical genetics; diagnostic and monitoring) requested as part of the consultation and investigation.
- compliance with national protocols and guidelines developed in collaboration with national and international colleagues
- continuing education and training for other health care professionals and students that are engaged in the care and management of a metabolic patient and their family and whānau
- providing information to individuals and families about sources of community support and support groups
- audit and relevant review processes of the service and complex metabolic disease management in New Zealand.

5. Service objectives

5.1 General

The objectives of the Service are to provide:

- timely consultation, investigation, diagnosis and interventions to identify and meet the clinical requirements of the metabolic patient
- linkages and liaison with community services and supports, resulting in metabolic patients and their families being more informed and accessing the services and supports that they need.

5.2 Māori Health

An overarching aim of the health and disability sector is the improvement of health outcomes and reduction of health inequalities for Māori. Health providers are expected to provide health services that will contribute to realising this aim. This may be achieved through mechanisms that facilitate Māori access to services, provision of appropriate pathways of care which might include, but are not limited to:

- referrals and discharge planning
- ensuring that the services are culturally competent
- providing services that meet the health needs of Māori.

Additionally, to improve access to the Service by Māori and their whānau by the identification of barriers and the deployment of culturally specific approaches to engage Māori thereby improving Service outcomes.

6. Service Users

Service users will be patients and their family and whānau with symptoms and or signs of metabolic disease or those with confirmed metabolic disorders, or positive metabolic newborn screening tests.

7. Access

Access to the Service will be via a referral from a health care professional, usually a secondary health care provider. The Service is a week day service.

7.1 Entry Criteria

Patients and their family and whānau will enter the Service following receipt of and the clinical triage of a referral. The referral will indicate that the patient and their family and whānau require a greater level of specialist metabolic expertise than can be provided by generalists in primary or secondary health care.

7.2 Exit Criteria

Discharge planning will assist exit from the Service including handover to secondary or primary health care providers for ongoing management.

However most patients with a metabolic disease will remain linked to the Service during their lifetime.

7.3 Time

A clinical triage process will determine level of priority.

- Urgent referral (acute): immediate telephone consultation within one working day
- Semi-urgent: video/teleconference consultation within 5 working days
- Routine (Chronic): consultation within 6 months of referral.

8. Service Components

The Service components to be provided by the multi-disciplinary team are:

8.1 Assessments and Treatment – Metabolic Management

The metabolic management offered by the Service will include the following:

- specialist assessment (including history, clinical investigations and diagnosis), in person or by consultation using a family / whānau centred approach
- treatment for patients and their family and whānau with metabolic conditions requiring inpatient care at Auckland District and in local Districts in partnership with the medical team in the local District
- health promotion and education - teaching sessions for special interest groups, support groups, health professionals and interested clinical groups
- tertiary outpatient service for children and adults, including an outreach programme
- follow up, re-admission and treatment of all patients in whom complications arise in the course of treatment by the Service
- long term follow-up as needed including liaison with primary and secondary health care providers
- specialist input into clinical genetics services when required
- referral for Genetic counselling and request for Specialist biochemical genetics testing.

8.2 Pacific Health

The Service will recognise the unique metabolic profile present in the Pacific peoples and the responsibility to prevent, treat and report this accordingly. The Service will also consider the cultural requirements of Pacific people individuals and their families, and support their access to service and consent procedures.

8.3 Other Ethnicities

The Service will consider the ethnicities of the local populations and their specific cultural requirements including access to interpreter services and support to promote access to services.

8.4 Settings

The Service should be mainly conducted in outpatient and / or as a consultative service and, as and where appropriate, may be provided to patients and their family and whānau while the patient is an in-patient.

8.5 Service Levels

The Service will be provided at a tertiary level and there will be an expectation that shared care occurs with generalist paediatricians in secondary health services and primary health care providers.

8.6 Equipment

The Service requires access to telecommunications / telemedicine to facilitate non contact / virtual and distant consultations.

8.7 Key Inputs

The major input to the Service is the multi-disciplinary workforce which will include the following:

- Metabolic Physician (Senior Medical Officer)
- Registered Nurse with metabolic nursing training and experience
- Dietitian with metabolic training and experience
- Clinical Psychologist.

9. Service Linkages

Generic service linkages are described in the tier one Specialist Medical and Surgical Services and Services for Children and Young People service specifications.

Service specific linkages include, but are not limited to the following.

Other Service Provider	Nature of Linkage	Accountabilities Associated with Linkages
Primary Care Organisations/ General Practitioners Nurse Practitioners Lead Maternity Carers Well Child/Tamariki Ora Service providers	Referral or liaison	Refer or liaise re individual patients as appropriate. Service access criteria are communicated. Work with other relevant professionals whenever there are concerns relating to an individual.
Public Health Service – public health nurses, health protection officers	Referral, provide information or expert opinion.	Provide education, information or expert opinion. Work with other relevant professionals whenever there are concerns relating to an individual
Hospital services: • Clinical Genetic Services • Tertiary paediatric services • Newborn Services • General paediatric services (in or outpatient) • Allied Health workers, nursing staff, junior medical staff • Home Care for Kids services, Occupational therapists, Speech language therapist, Neuro-psychologist	Liaison on referral Referral and counselling	Provide education, information or expert opinion. Work with other relevant professionals whenever there are concerns relating to an individual Provide clinical assessment, treatment and intervention in shared care arrangements and as required.

Other Service Provider	Nature of Linkage	Accountabilities Associated with Linkages
• General Medicine / Surgical services (adults in-outpatient) • Physiotherapy • Social services/social worker		
Newborn metabolic screening programme	Referral or liaison	Responding to referrals.
Specialist biochemical and molecular genetics laboratories	Liaison	Respond to laboratory results.
Community support groups	Liaison	Provide information.

10. Exclusions

This service specification does not include the following:

- laboratory tests ordered by clinicians not related to the Service
- special foods, dietary supplements or other medications for metabolic patients supplied by PHARMAC.

Also refer to the tier one Specialist Medical and Surgical Services service specification.

11. Quality Requirements

11.1 General

The Service must comply with the Provider Quality Standards described in the Operational Policy Framework or, as applicable, Crown Funding Agreement Variations, contracts or service level agreements. All clinical services will meet all applicable service guidelines.

11.2 Safety and Efficiency

All laboratories undertaking genetic testing (biochemical, molecular, metabolic and cytogenetic) as part of, or on behalf of, the Service must have current IANZ[1] or equivalent accreditation and should participate in recognised quality assurance programmes.

A programme of routine audit will be developed and undertaken for the Service.

12. Purchase Units and Reporting Requirements

Purchase Unit Codes are defined in Health New Zealand's Nationwide Service Framework Purchase Unit Data Dictionary. The following codes apply to this Service.

The Service must comply with the requirements of national data collections where available.

[1] International Accreditation New Zealand is the national authority for the accreditation of testing and calibration laboratories, inspection bodies and radiology services.

PU Code	PU Description	PU Definition	PU Unit of Measure	National Collections or Payment Systems
NS10031	National Services Pediatric and Adult Metabolic 1st Attendance	First attendance to a medical officer at registrar level or above or nurse practitioner for specialist assessment. (Excludes M45010)	Attendance	NNPAC
NS10032	National Services Paediatric and Adult Metabolic Subsequent Attendance	Follow-up attendance to a medical officer at registrar level or above or nurse practitioner for specialist assessment. (Excludes M45010)	Attendance	NNPAC
M00010	Medical non contact First Specialist Assessment - Any health specialty	A review is undertaken by a Registered Medical Practitioner of Registrar level or above, or a Registered Nurse Practitioner, of patient records and any diagnostic test results. The original referral should only be generated after a face to face contact by the referrer. A written plan of care is developed for the patient and provision of that plan and other necessary advice is sent to the referring clinician and the patient. The patient should not be present during the assessment.	Written plan of care	NNPAC
M00011	Medical non contact Follow Up - Any health specialty	A review is undertaken by a Registered Medical Practitioner of Registrar level or above, or a Registered Nurse Practitioner, of patient records and any relevant diagnostic test results. The patient is not present during this follow up that should only be undertaken after a face to face contact by the same service. A written plan of care is developed for the patient and that plan and other necessary advice is sent to patient and if applicable to referrer. Diagnostics are only to be included if ordered by the District providing the non-contact follow up.	Written plan of care	NNPAC

Explanation of the unit of measure for the purchase unit.

Unit of Measure	Definition
Attendance	Number of attendances to a clinic/department/acute assessment unit or domiciliary
Written plan of care	Written plan of care provided by the specialist to the referring GP

12.1 Service Planning

The Service will be cognisant of future workforce needs and participate in succession planning.