

ROYAL CHILDREN'S HOSPITAL

POSITION DESCRIPTION

POSITION TITLE: Fellow in Genetic Metabolic Diseases (Metabolic Medicine)

DEPARTMENT: Metabolic Service, Genetic Health Services, Victoria; RCH

ROSTERED HOURS OF WORK: Full time (9-5) + on call

REPORTS TO: A/Prof Avihu Boneh

SUPERVISOR (Person responsible for feedback / training): A/Prof Avihu Boneh

Department's purpose or mission statement:

The metabolic service provides diagnostic, therapeutic and genetic-counselling services to children and adults with metabolic disorders, and their families, from all over Victoria and Tasmania. This includes the provision of results and the management of newborn babies diagnosed through the expanded newborn screening program. We focus on the integration of this area of paediatric medicine with general paediatrics as well as other paediatric sub-specialties. We emphasise joined care management with Paediatricians in the community (mainly country Victoria and Tasmania).

There are over 500 'active' patients and over 1000 patients listed in our books. The number of new patients has risen in the past few years. As well, the number of consults, either from within RCH or from paediatricians, neurologists, endocrinologists and GPs from the community has risen.

Exposure to as many patients as possible, and dealing with as many clinical dilemmas as possible, all under appropriate supervision, will ensure proper use of the training time for learning the fundamentals of Metabolic Medicine: diagnosis, therapy and genetic counselling.

Clinical and 'basic' research is encouraged.

Individual training or development objectives:

Full training in metabolic medicine is currently under the SAC for Genetics. The curriculum for such training includes 2 years Metabolic Medicine and one year compulsory clinical Genetics.

Alternatively, training in metabolic medicine can be complementary to a fellowship in another discipline in Paediatrics or Adult Medicine such as: General Paediatrics, Endocrinology, Neurology, Child Development, Gastroenterology/Hepatology, Clinical Genetics, Intensive Care etc. You will need approval of the appropriate SAC.

It is hoped that by the end of your training period you will have a solid knowledge of the clinical approach to patients with suspected metabolic disorders, and be able to interpret results of metabolic tests and administer therapy.

It is also hoped that you will produce a scientific paper at the end of your training period. Support and guidance in research and presentation of research are provided.

Qualifications required

The position would be appropriate for an advanced trainee in Paediatric Medicine.

Experience required

Advanced training in Paediatric Medicine. Experience in neonatology, intensive care, neurology, nephrology or endocrinology is advantageous.

Position summary:

As a trainee in Metabolic Medicine you will participate in the management of patients with metabolic disorders who are admitted to the Royal Children's Hospital as well as outpatient clinics. This includes patients admitted for investigations, patients admitted acutely for decompensation and patients admitted for enzyme therapy. You will also participate in laboratory reporting sessions for a variety of metabolic tests (urine organic acids and amino acids, 'urine metabolic screen', plasma amino acids, Tandem Mass Spectrometry, mitochondrial enzymology).

You will work in a multidisciplinary team comprising physicians, metabolic nurse/genetic counsellor, dietitians and a social worker.

Exposure to as many patients as possible, and dealing with as many clinical dilemmas as possible, all under appropriate supervision, will ensure proper use of time for learning the fundamentals of Metabolic Medicine: diagnosis, therapy as well as some genetic counselling.

Key areas of responsibility within the department:

Inpatients:

- Ward Rounds: Daily, if there are patients admitted.
- Multidisciplinary management of metabolic patients in liaison with other medical services at RCH, as needed.
- Consults and follow up of investigations for in-patients referred from other services.

Outpatients:

- Weekly follow-up clinic.
- Weekly new consults clinics.
- Phone consults.
- Newborn screening – results and management.

Administrative tasks:

Discharge summaries: For patients electively admitted for metabolic investigations.

Other: Clinic letters, other letters of support to child/family, as needed.

Departmental meetings

- Clinical metabolic meeting (weekly)
- Biochemical Genetics Laboratory reporting meeting (weekly)
- Plasma amino acids reporting meeting (weekly)
- Mitochondrial reporting session (weekly)
- Newborn Screening: reporting and liaison

Departmental training activities

- Journal club (Genetic Health)
- Clinical seminar (Genetic Health)

Communication / lines of responsibility:

Working with Neurology/Metabolic registrar and resident
Working in a multi-disciplinary team
Consultants: A/Prof A. Boneh, Dr H. Peters and Dr J. Lee
Director of Metabolic Service: A/Prof A. Boneh

People management: N/A

General hospital responsibilities: N/A

General or Specialities Afterhours, weekend roster etc:

A metabolic fellow is on call at all times (shared roster).

A metabolic consultant is always available as back-up.