Rare Disease

Supporting Murdoch Children's Research Institute's drive towards Precision Child Health

Key metrics

75 per cent of rare diseases affect children

Rare disease is estimated to cause **35 per cent** of deaths in children younger than 1 year

More than **5000** patients are seen for rare disease diagnosis each year on the Melbourne Children's campus

Rare disease is responsible for around **20 per cent** of paediatric hospital admissions

Around **80 per cent** of rare diseases have a genetic origin

MCRI has **177 rare disease publications** in top 10 per cent of journals

MCRI has made more than **100** genetic discoveries

Rare disease and children

At least one in 12 babies is born with a rare disease.

There are more than 6000 known rare diseases and 75 per cent affect children. The causes of many of these diseases remain unexplained — 30 per cent of affected children will die before they are 5.

Despite the progress in diagnosis of rare diseases, and in genetic and genomic research, many Australian families still do not have access to genomic testing. When families do get a diagnosis, many rare diseases have no treatment options.

Interventions that improve outcomes for children with genetic rare diseases require early, rapid and accurate genetic diagnosis.

Rapid genetic diagnosis provides benefits to patients and families, and economic benefits to the health care system.

In Australia, current delays in obtaining accurate genomic diagnosis prevent early interventions, personalised care and future family planning.

Vision

For all people living with a rare, indeterminate disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention.

Current mission

An MCRI-led rare diseases program that facilitates:

discovery of novel disease genes, mechanisms and treatments
engagement with clinicians, researchers and consumers
translation of research via a streamlined system and infrastructure
equitable access to services, treatment and care for all those with rare disease.

MCRI's Rare Disease flagship

MCRI has made over 100 genetic discoveries, led the development of national and international genomic alliances, houses a purpose-built genetics service and is home to Australia's leading paediatric trials centre.

Our Campus partner The Royal Children's Hospital and MCRI's Victorian Clinical Genetics Services see over 5000 patients and make more than 250 genomic diagnoses of rare diseases every year.

MCRI can now further understand the mechanisms that cause rare disease. Critically, MCRI has the capacity to develop novel treatment options and personalised care for those with rare disease.

MCRI's Rare Disease flagship is a strategic, collaborative program uniting clinicians, biologists and bioinformaticians to tackle unanswered and important issues for those affected by rare disease.

The MCRI Rare Disease flagship can accelerate our understanding of the causes of each and every rare disease and develop new interventions. Ultimately, it can find the prevention strategies that are so desperately needed.

Understanding causes and developing new interventions will not only improve children's lives, but provide the evidence needed for policy change so all Australian families can have access to rapid and accurate genomic testing.

Leadership

Prof David Thorburn

A/Prof Sue White

Dr Cas Simons

A/Prof Tiong Tan

The Murdoch Children's Research Institute

The Murdoch Children's Research Institute (MCRI) is Australia's leading child health research organisation.

Our researchers work to translate knowledge into effective prevention, intervention and treatment to address a range of disorders affecting infants, children and adolescents.

MCRI advocates for quality equitable care for all children and works closely with its partners, The Royal Children's Hospital and the University of Melbourne's Department of Paediatrics, within a single, purpose-built facility. The Campus partnership is known as Melbourne Children's, with many of the key campus research initiatives supported by The Royal Children's Hospital Foundation.