



Pioneering a Pathway to Curing Metabolic Disorders for Children

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The Westmead Institute
FOR MEDICAL RESEARCH



"As a senior research scientist with an extensive diagnostic background, I'm involved in translational research and personalised medicine to find treatment for amino acid metabolism disorders in children, including newborns, as well as young adults at The Storr Liver Centre, The Westmead Institute for Medical Research and the Children Hospital at Westmead.

Metabolic conditions are not the same so it can take a while to find the best treatment option. Treatment strategies are usually focused on special diets that are typically complex, costly, and inconvenient, plus hard to maintain, particularly later, through adolescence and adulthood.

I believe there is a better way and am very excited to be breaking new ground by generating a biobank of induced pluripotent stem cell from blood or skin fibroblast of patients with amino acid metabolism disorders to be able to perform a large-scale drug screening, taking a vital step towards personalised medicine to find targeted treatments for each child. Ultimately, my aim is to provide not just a treatment, but a cure.

I am very thankful to the community of supporters who have contributed to help make my work possible so far. If you too are in a position to provide your support, I would be truly grateful."

Dr Michael Nafisinia
Storr Liver Centre

We're using affected patient's cells to find targeted drugs for amino acid metabolism disorders.

Metabolic disorders, such as PKU (phenylketonuria), are genetic and mean that a person can't process and digest a certain protein because of a deficiency of enzymes, converting one amino acid to another or to final product in the liver. Different amino acid metabolism disorders are caused by specific genetic variations.

Dr Nafisinia and his team are pioneering a radically new way of understanding each disorder and how it might be treated – using variety of expertise at the Storr Liver Centre, we're taking patient's blood or skin samples, reprogramming them backwards to become induced pluripotent stem cells, then differentiate those induced pluripotent stem cells into cell of interest such as liver cells, neuronal cells, and bone cells (two dimensional cell culture (2D)) for the purpose of large scale drug screening. Further, we're able to generate organoids which are three-dimensional (3D) cell cultures that incorporate some of the key features of the represented organ. These in vitro (in laboratory) culture systems are characterized by the self-organization of multiple, organ-specific cell types into a spatial organization similar to what is observed in vivo (within living organism) and are capable of recapitulating some functions of the represented organ. Testing drugs on organoids is giving better indication of how the effected organ will respond to the treatment.

Using both 2D and 3D cell cultures, we're able test these to properly understand the faulty enzyme activity characteristic of each genetic disorder and trial them against different drug candidates to see what impact is achieved.

Most excitingly, this technique can be used to test existing drugs already approved and proven to work in the treatment of other diseases with similar genetic variations. This means that rather than trying to develop new drugs from scratch, we'll be able to start identifying the most effective targeted treatment options for each metabolic disorder even sooner.

There is much more work to be done but progress to date is extremely promising. Ultimately, it is set to dramatically improve the way metabolic disorders are treated, or even cured, improving quality of life and bringing relief to all those impacted.



We welcome your support

Your support will ensure that the research work being undertaken by Dr Michael Nafisinia and his team can proceed without delay.

Please consider a tax-deductible gift at the level that is right for you:

\$50 – cover costs of essential lab consumables used every day (e.g. microscope slides, disposable gloves)

\$100 – cover costs of test tubes and pipettes for one experiment

\$500 – cover costs of specialised cell culture media

\$2000 – cover costs of producing a partial batch of organoids from stem cells

\$5000 – cover costs of collecting and analysing data from one six-week experiment

Medical research is time intensive and exacting. Community support is essential to ensuring continuity and sustainability.

Outstanding research requires exceptional people

You don't have to be a medical researcher to make a difference.

Donate today to help improve treatments, prevent, and cure some of the most serious health issues affecting Australians and people around the world.



Visit our website to donate now. Simply hover your phone over the QR code or visit westmeadinstitute.org.au

For Dr Michael Nafisinia's research, please direct your donation to Metabolic Disorders for Children.

To find out how your support can make a difference, contact The Westmead Institute for Medical Research Foundation:

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Thank you for your support and generosity. Together we are giving hope and saving lives.



The WIMR Difference


At WIMR our great strength is research that is:

Transdisciplinary – we're working across disciplines and tackling some of the most serious disease challenges of our time.


Translational – research is informed by our researchers' direct experience of working with patients and moves rapidly from bench to bedside.

Transformative – our priority is research that leads to dramatic improvements in health outcomes and quality of life.

WIMR is leading the way in embedding the concept of Precision Medicine into medical research, pioneering new approaches to enable personalised diagnosis and treatment strategies for each individual.

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