CHILDREN'S MEDICAL RESEARCH INSTITUTE
Jeans for Genes®

Finding cures for children's genetic diseases

Under the microscope

Arato's story

There's one huge goal that we share, and that is to ensure every child has a chance to grow, succeed and live a healthy, happy life.

Arato, 12, is just one of the many kids we're working together to help. He lives with genetic blindness and is a shining success story of his own making!

We shared Arato's story just before Christmas last year, and so many of you wrote kind messages wishing him luck in his next tennis tournament. We passed these messages on to him, and you can see for yourself just how much he appreciated reading them.

Arato had some other huge milestones in the first weeks of this year: he started high school and appeared on centre court at the Australian Open!

"He was a bit nervous about starting high school, because he didn't know anyone, and he had to learn to navigate the new surroundings, but he loved it from day one," Arato's mum Junko said.

Another highlight of the New Year was getting to toss the coin at the Australian Open quarter-final, and signing with Tennis Australia to play in the Low Vision Competition.

"It was an amazing experience for him. Many high-profile players never get to step onto centre court, so he was incredibly lucky."

Watch Arato reading your well wishes

AUTUMN EDITION 2024

From the Director

Welcome to our latest edition of Under the Microscope.

I am happy to inform you of some great progress our scientists are making in 2024, with new research discoveries in the areas of cancer and gene therapy published in world-leading scientific journals.

One of these discoveries is the identification of a new cancer treatment target, a molecule that can be disrupted in cancer cells in order to kill them – another significant success achieved by Professor Hilda Pickett and team.

We recently held a major event to celebrate 65 years of impact at Children's Medical Research Institute (CMRI). Our Patron, the Governor General of Australia, generously opened his Sydney home, Admiralty House, to host this evening for many distinguished guests. The Governor General spoke of his confidence in the great future that CMRI's scientists are helping to create, together with you, our community, for generations of children.

Working with the community is crucial to our continued success, not only for raising vital funds for life-saving research, but also for awareness and advocacy. This was reinforced by some of our clinical colleagues during a Rare Disease Family Day held at CMRI. They explained the vital role CMRI plays in connecting with patients and their families, advancing science, and working with clinicians in the health system to progress these scientific discoveries into treatments for the children who need them most.

I always look forward to hearing from you and invite you to write to us at research@cmri.org.au to share your thoughts and hopes for the future.

Roger Reddel AO
Lorimer Dods Professor and Director, CMRI

Professor Hilda Pickett

Our Research

Improving outcomes for all cancers

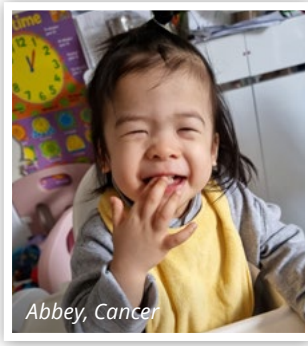
We're thrilled to share a new discovery with you, recently made by one of our talented researchers. This could have a huge impact for cancer patients, leading to improved cancer treatments that have fewer harmful side effects on normal cells.

Professor Hilda Pickett, who leads our Telomere Length Regulation Unit, said this was an exciting development for Dr Alex Sobinoff and other team members.

"This project builds on over a decade of work by my team and our collaborators. The results are promising and have future potential to improve long term health outcomes for all types of cancer."

Dr Sobinoff found a compound that disrupts growth in cancer cells while sparing non-cancerous cells.

"These compounds represent fantastic new tools for fundamental research," Dr Sobinoff said.



Abbey, Cancer

Mother of two, Irene, has watched her 2-year-old daughter, Abbey, go through cancer treatment including surgery after receiving her diagnosis for Wilms tumour. Her genetic disease, WAGR syndrome, puts her at risk of the cancer returning, so Irene may have to face this again. Irene understands how past medical research saved Abbey's life, and she's hopeful that more discoveries will lead to better treatments.

"Abbey was only 18 months old when she was diagnosed, and she was just starting to walk. Then she had all the treatment, and suddenly she couldn't even stand and didn't have the energy to walk. If this cancer comes back again, we don't want her to face extra struggles every time she is treated. That's why research is so important."

Find out more at bit.ly/cancer-news-march

14 million
children and adults
worldwide are diagnosed with cancer each year

2 Australian
children
and their families face a cancer diagnosis every day

Children
aged 4-14
face cancer as a leading cause of death

"Whatever I can contribute towards funding that research seems to me to be a very worthwhile legacy, and that's why I'm leaving a gift in my Will."

- Lindsey Freeman

Part of the Team

"My name is Lindsey Freeman - I know two children that were born with a genetic disease - one is my son, and the other was my nephew's son. It has made me appreciate the profound impact these diseases can have, and the critical importance of ongoing medical research to achieve previously-unimaginable positive outcomes.

My son, Richard, was diagnosed with cystic fibrosis and was in and out of hospitals leading up to the diagnosis. The shock and sadness of learning that our dearly loved little boy was unlikely to live to adulthood and would be unwell and vulnerable all his short life, was unbearable. We grieved the loss of the hopes parents have for a child and their future.

At that time there was no genetic testing for carriers of the cystic fibrosis gene, and we were advised that there was a very high risk that any subsequent children of ours would have cystic fibrosis too. So we opted not to have more children - another blow. Now, thanks to research, that situation has changed, and the risk can be managed.

I've seen what can be achieved by medical research in relation to identifying the gene for cystic fibrosis and its early diagnosis and treatment. But also, I have seen the great need for research to enable better and earlier diagnosis and treatment to save and improve the lives of the many children with as-yet incurable genetic disorders."

Associate Professor Leszek Lisowski with Spela and Samo Miseovic

Talking to a young visitor at the Family Day

Community & Committees

Rare Disease Family Day

Spela Miseovic

Families, researchers, and clinicians came together for our first ever Rare Disease Family Day to talk about the progress of our gene therapy research.

A special guest of the event was Slovenian mother, Spela Mirosevic, who together with her husband, Samo, started the International CTNNB1 Foundation after her son, Urban, was diagnosed with this rare condition. Many children with this condition cannot walk or talk.

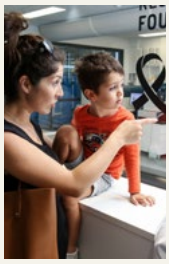
"In the ideal world, you would go to the doctor, the doctor would give you the diagnosis and then they would give the treatment," Spela said. "Unfortunately, we do not live in that ideal world...at least not yet. However, we live in a time where there is hope with gene therapy.

"We started with a simple wish - to become parents. But along the way, we've grown into so much more. We've become advocates, tirelessly working to build a better future for our children and lending our voices to others in need.''

The CTNNB1 Foundation reached out to CMRI after hearing about our work from families and rare disease experts three years ago. Since then, our very own Associate Professor Leszek Lisowski, Head of CMRI's Translational Vectorology Research Unit, has been working on an experimental gene therapy for CTNNB1 syndrome.

"There are over 6,000 known genetic disorders, which combined affect an estimated 1 in 20 children," Leszek said.

"I'm very excited that the gene therapy technology has reached a level of maturity which allows us now to develop treatments, or even cures, for many of those disorders."



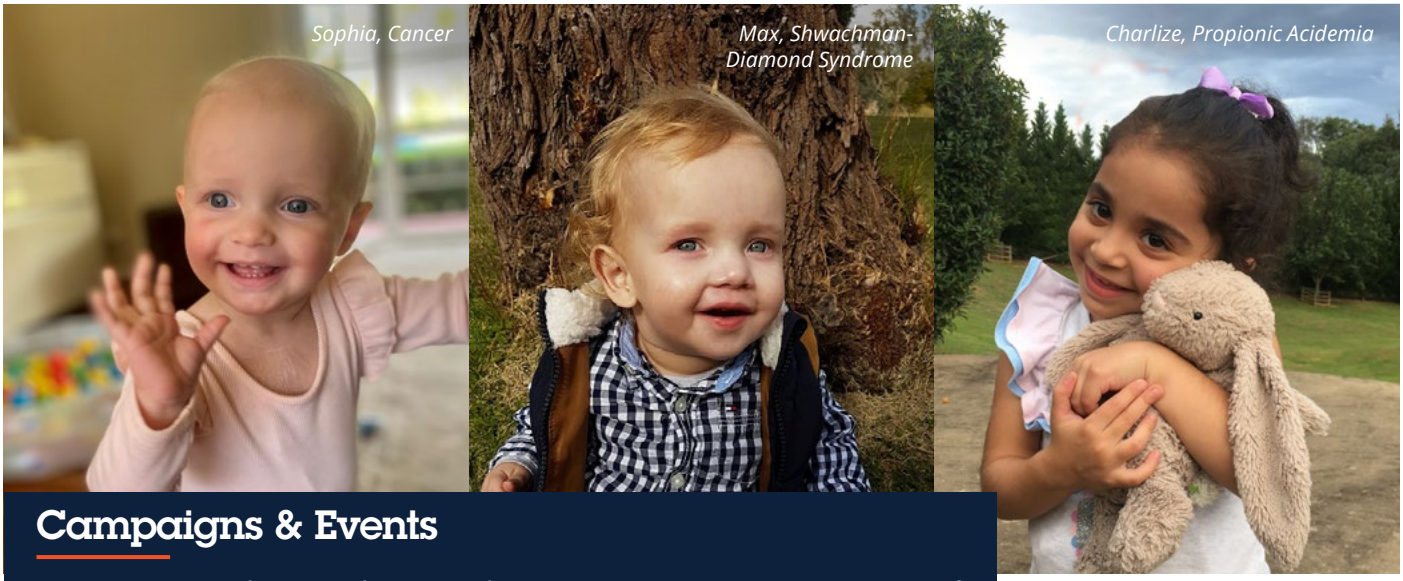
You're invited to our Discovery Day this year! We have different dates available throughout the year where you can tour our labs, learn about our research, and enjoy a morning tea and a light lunch.

Call us on 1800 346 347 to register

If you love getting involved in running, cycling, walking, or swimming events, you could be doing it to support medical research and the children who benefit from it!

These events suit all fitness levels.
Find out more at https://cmri.grassrootz.com

Jeans for Genes is on Friday 2nd August this year!



Campaigns & Events

What's next for the future of kids with a genetic disease?

We've analysed the results from a survey we sent out last year, and you've asked to hear more about our future plans. We're thrilled you asked!

We've already made some huge achievements over the years.

We've improved survival of premature babies, which used to be a leading cause of infant death. We've pioneered microsurgery, enabling organ transplants in children. We've contributed to the Rubella vaccine, the first heart and lung life-support machine, made major advancements in gene therapy for genetic diseases, and have developed a world-first, rapidly increasing 'big data' library of cancer information that will transform how cancer is diagnosed and treated.

Many of these milestones have already helped countless children, both in Australia and around the world.

You might be wondering what's next, and here at CMRI, we face our challenges head-on because we know you'll be by our side.

The goal has always been simple – give kids the best chance at living a long, healthy, and happy life. But until we can treat or better yet, cure, all childhood genetic diseases – the work isn't done. It's only with your support that we can take these next steps together.

We need to further improve our understanding of genes that cause disease and the way we test for genetic diseases, such as for the 1 in 1,000 people affected by inherited blindness.

We also need to continue developing new gene therapies for a range of conditions – there are over 6,000 known genetic diseases so there's an enormous amount to do, but we're starting with serious conditions like cystic fibrosis, deadly liver metabolic diseases, and neurodevelopmental disorders.

8 million people die from cancer worldwide every year – an unacceptably high number. We need to make cancer treatments more personalised for a greater chance of successful treatment with fewer side effects.

Thank you for standing by our side and sharing the same goals – you're showing sick kids just how much you care, and how hopeful you are for their futures.

1800 436 437

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