

Australian breakthrough in stem cell kidney research

May 4 2018



Credit: Murdoch Children's Research Institute

New research led by the Murdoch Children's Research Institute (MCRI), has combined gene editing technology with stem cell kidney regeneration to correct a patient's gene mutation.

This is the first time a patient has had kidney regenerated from their stem [cells](#) in Australia.

The research, is part of a regenerative medicine project in which human stem cells are used to develop mini-kidneys with a view to discovering new [genes](#) and treatments for inherited [kidney disease](#).

In this new study, published in the *American Journal of Human Genetics* and involving hospitals and laboratories in Brisbane, Sydney and Melbourne, stem cells derived from a child with genetic kidney [disease](#) were grown into two sets of living mini-kidney organoids – one with her kidney disease and one in which her [gene mutation](#) was corrected.

The stem cells were created from a skin biopsy taken from 12-year-old Alexandria, who suffers from Mainzer-Saldino Syndrome, a rare genetic condition causing progressive retinal degeneration and end-stage kidney disease.

"Within the patient's mini-kidneys, we discovered abnormally shaped cellular antennae. This showed that Alexandria's disease exists within the mini-kidney and proves that we can use these regenerated tissues to learn about her disease," said MCRI researcher and paediatric nephrologist, Dr. Tom Forbes.

"We then used gene editing technology to correct the genetic mutation in Alexandria's stem cells. These 'gene-corrected' stem cells were then grown into another set of mini-kidneys which showed the antennae were now a normal shape. So the gene-correction stopped the development of the disease within the mini-kidney."

"By comparing the two mini-kidneys, we now have a better understanding of how this disease develops."

Alexandria's is the first reported case in which the effect of a gene mutation in kidney disease has been investigated using gene editing of patient stem cells to correct the genetic mutation.

"The fact that we can make [kidney tissue](#) from human stem cells and then correct any [genetic mutations](#) to study the way the disease develops is a very promising step towards developing personalised future

treatment," said MCRI senior research officer Dr. Sara Howden.

At least 50% of children who require dialysis or a [kidney transplant](#) have a genetic cause for their kidney disease. For almost all of these conditions, very little is known about how the disease develops and no specific treatment exists.

"By growing mini-kidneys from a patient's stem cells we're hoping to find new genes responsible for kidney disease. We also hope we can test them for possible new treatments for that patient's specific disease."

While this latest research won't impact Alexandria's current condition, it does pave the way for possible treatments in future for other patients with similar renal diseases.

"There is a long way to go to make personalised treatments, but knowing we can study inherited kidney disease using a patient's [stem cells](#) is a breakthrough step," said Dr. Howden.

Alexandria's story:

When she was six months old, Alexandria was diagnosed with a degenerative vision impairment, however the cause was unknown. Five years later, with only three per cent kidney function, Alex suffered renal failure and was immediately placed on dialysis.

At six years of age, Alex underwent a renal transplant using a [kidney](#) donated by her grandmother. While the transplant was a success and Alex's kidneys began to function well, the cause of her renal failure was also unknown.

Alex was 10 years old when genetic testing undertaken by Genetics Health Queensland finally led to an answer. Alex was diagnosed with a

[rare genetic condition](#) called Mainzer-Saldino Syndrome. This was very exciting news for Alex and her family as it has given them some resolution to her diagnosis and access to current research into the condition.

Now in year seven, Alex loves taekwondo, swimming, dancing, performing, reading and trying new things. She is a very determined young lady and an inspiration to everyone she meets.

Provided by Murdoch Children's Research Institute

Citation: Australian breakthrough in stem cell kidney research (2018, May 4) retrieved 3 May 2024 from <https://medicalxpress.com/news/2018-05-australian-breakthrough-stem-cell-kidney.html>

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