

SA Scientist of the Year 2019 is Professor Jozef Gecz

Our state's top scientist of 2019 is a global leader in the genetic and biology of childhood disability including epilepsy, autism and cerebral palsy.

Clare Peddie, Science Reporter, The Advertiser

August 9, 2019 10:00pm

A dream to find a drug for multiple medical conditions is coming true for Adelaide's top scientist striving to prevent childhood disability.

University of Adelaide neurogenetics Professor Jozef Gecz was named SA Scientist of the Year at last night's Science Excellence Awards dinner in the Adelaide Convention Centre.

The professor is a global leader in the genetics and biology of childhood disability, including epilepsy, autism and cerebral palsy.

Over 25 years he has helped discover more than 200 relevant genes, with many now used in routine diagnostic tests worldwide.

"My work has changed the understanding of the causes of neurodevelopmental disability, for early diagnosis and personalised interventions," Dr Gecz said.



"These things can be diagnosed early, technically they can be prevented and some of these disorders are treatable, a small number of them but I am sure that number will continue to grow.

"My research focus is to find the causes, understand the mechanisms and go into finding the drugs which are going to help these individuals."

Right now he's "very excited" about the Phase III clinical trial of a drug for "girls clustering epilepsy", a rare and debilitating disorder that arises within the first three years of life.

In 2008, he discovered the offending gene (PCDH19), which he then patented, and he brought together scientists, patients and their families to form an international community.

Recognising the potential for an existing drug (ganaxolone) to be repurposed to treat this disorder, he proposed and instigated clinical trials with Marinus Pharmaceuticals.

The drug corrects a deficiency in cases of girls clustering epilepsy.

"Interestingly this molecule, a neurosteroid, is involved also in post-traumatic stress disorder, in Alzheimer's, in post-partum depression, so in women who are depressed after birth," he said.

“So clearly, we are tapping into a fundamental mechanism in the brain, which is somehow broken and these genetic findings are really leading the way to understand and also perhaps help treat multiple different groups of disorders, eventually. I guess that would be my dream, to discover another Aspirin or something like that which generally is going to help, across a whole range of medical conditions.”

Minister for Innovation and Skills David Pisoni said Professor Gecz was a research pioneer with an enviable track record in gene discovery in intellectual disability, epilepsy, autism and more recently, cerebral palsy.

“Congratulations to Professor Gecz, whose work has helped to establish South Australia as an international leader in the field of gene disease discovery,” Mr Pisoni said.

“His research has had worldwide impact and his work has transformed the understanding of causes and underlying mechanisms of neurodevelopmental disability, leading to clinical trials, informed national and international policy, and improved health care delivery.”

Full list of winners

South Australian Scientist of the Year: Professor Jozef Gecz, University of Adelaide

Excellence in Research Collaboration: “B Part Of It” Meningococcal B vaccine team

STEM Professional of the Year: Dr Mark Skanes, Principal Engineer Dommar Pty Ltd

STEM Educator of the Year in Tertiary Teaching: Associate Professor Tom Raimondo, UniSA

STEM Educator of the Year in School Teaching: Ms Anthea Ponte, Hawthorndene Primary School

PhD Research Excellence: Dorothea Dumuid, UniSA

South Australian Tall Poppy of the Year: Dr Nigel Rogasch, University of Adelaide and SAHMRI

Unsung Hero of South Australian Science: Zoos SA Research Laboratory

Unsung Hero of South Australian Science Communication: Dr Philip Roetman, City of Burnside