

Stillbirth and chronic disease link identified in world first discovery

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In a world first discovery, South Australian researchers have identified a genetic mutation responsible for a lymphatic disorder that may cause stillbirth or severe, chronic disease in affected children.

An anomaly in the development of lymphatic vessels in unborn children, leading to fluid accumulating in the heart, lungs and other organs, has been uncovered by scientists from the Center for Cancer Biology (CCB) based at the University of South Australia (UniSA) and SA Pathology.

The findings are published today in the journal *Science Translational Medicine*.

CCB Director Professor Natasha Harvey says a genetic study of six families affected by stillbirth or lymphoedema revealed the link between a mutated protein coding gene called MDFIC and fluid accumulation in vital organs and tissues.

This has demonstrated that MDFIC is important for controlling the growth and development of the lymphatic vessels in the fetus for the first time.

The Center for Cancer Biology collaborated with scientists and clinical teams from the Women's and Children's Hospital, University of Adelaide, Belgium, Germany, the United States and Iran to make the breakthrough.

"The lymphatic system is a network of vessels (pipes) and nodes (filters and control centers) important for maintaining fluid balance in our tissues and transporting infection-fighting [white blood cells](#) throughout our bodies," Prof Harvey says.

"We determined that MDFIC controls cell migration, an important early event during the formation of the lymphatic vessel valves. The genetic variants we have found in our study reveal a crucial, previously unrecognized, role for MDFIC in the lymphatic vasculature.

"If the lymphatic valves don't form properly, lymph fluid accumulates in

critical organs such as the heart and lungs, causing major respiratory problems that may eventuate in stillbirth or chronic disease."

An SA Pathology research team headed by Professor Hamish Scott initially found the genetic link in an Australian family. Their international colleagues in Belgium, Germany and the US reported variants in the same gene, MDFIC, in several patients with the same [lymphatic disorder](#).

Prof Harvey says the disorder, known as central conducting lymphatic anomaly (CCLA), is one of a group of severe lymphatic disorders and may result in stillbirth, or severe chronic disease in affected children.

Few effective treatments are available but with continued identification of the genetic causes of CCLAs, Prof Harvey says the next step is to develop new therapeutic drugs to combat the disease.

"There are existing drugs that may be used to treat these disorders, but we need to make sure that the [signaling pathway](#) that's treated by those drugs is the same pathway that is affected in our patients.

"This project is about the power of collaboration at a local level, national level and international level, so that's been really important. Science is done in teams and we have great research teams here in Adelaide at the Center for Cancer Biology. Our Ph.D. students and postdoctoral fellows have been integral to this work. They have really driven it and we're delighted to be working with such a group of talented people."

More information: Pathogenic variants in MDFIC cause recessive central conducting lymphatic anomaly with lymphedema, *Science Translational Medicine* (2022). [DOI: 10.1126/scitranslmed.abm4869](https://doi.org/10.1126/scitranslmed.abm4869)

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