

Australia identifies stillbirth and chronic disease link in world's 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 Centre for Cancer Biology (CCB) based at the University of South Australia (UniSA) and SA Pathology. The findings have been published 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 Centre 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 centres) 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 unrecognised, role for MDFIC in the lymphatic vasculature", added Harvey.