



# NEWS UPDATE

## NEW RESEARCH ADVANCE IDENTIFIES DRUG TARGETS

Cerebral cavernous malformations (CCM) are the most common cause of stroke in young people. These malformations also known as Cavernomas are present in 0.1-0.5% of the population, with about 60% causing symptoms. Currently there is no drug treatment available for CCM.

However, as opposed to stroke in the elderly where no single genetic defect is implicated, in CCM defect in one of three genes (CCM1, CCM2 and CCM3) causes CCM disease. Correcting the consequences of these defects thus provides a plausible treatment, but hitherto how defects in the CCM genes cause disease has been unknown.

A new study published in Nature (Zhou et al) by Centenary's Dr Xiangjian Zheng's Cardiovascular Signaling group, Sydney Medical School and collaborators at Perelman School of Medicine, University of Pennsylvania, has identified abnormalities in the endothelial lining of brain blood vessels that have raised the possibility of a therapeutic intervention. To test this possibility they used a genetically engineered model to mimic human disease, where they were able to inhibit the expression of the culprit proteins and found they could prevent the formation of CCM and death from the subsequent strokes.

**This study provides three 'druggable targets' for the treatment for CCM disease. Dr Zheng has a long term involvement in understanding the genes causing CCM disease.**

The breakthrough for this study is based on their long time effort to mimic human disease and the recent invention of new micro-CT technology that allows visualization and quantification of the cavernous malformations. This study showcases that basic research can lead to the design of drugs for "untreatable" diseases.

## GETTING TO THE HEART OF SUDDEN DEATH IN EPILEPSY

Getting to the heart of sudden death in epilepsy Researchers from Centenary and the University of Sydney recently released results from the world's largest genetic study into sudden unexpected death in epilepsy (SUDEP), revealing a possible genetic link between the heart and the brain in epilepsy patients.

**The leading cause of epilepsy-related premature mortality is sudden unexpected death, and the cause remains unknown.**

State-of-the-art technology was utilised in this new research to examine all 22,000 genes of participants. Following an analysis of rare variants, it was found that a sizeable proportion of SUDEP cases have clinically relevant mutations in cardiac arrhythmia (irregular heart beat) and epilepsy genes.

Leading this study, Centenary's Dr Richard Bagnall, who is a genetic researcher in our Molecular Cardiology program, said this is a crucial first step in building our understanding of why SUDEP occurs and how we can prevent it. "This study has identified the first possible link between the heart and the brain in epilepsy patients", Dr Bagnall said.

"Understanding the genetic basis of SUDEP may inform the future diagnosis of at-risk family members, as well as provide opportunities for prevention."

University of Sydney Cardiologist Professor Chris Semsarian said this is an exciting development that could help to save lives into the future.

"If we can understand why SUDEP occurs, we can work towards preventing it. These new findings provide a platform to initiate available treatment options, such as antiarrhythmic drugs or implantable defibrillators, with the ultimate goal to prevent SUDEP in the community."

If you are interested in making a donation to a specific area of our research, taking part in or holding a community fundraising event please contact us on 1800 677 977 or email [donations@centenary.org.au](mailto:donations@centenary.org.au)  
**Thank you for your ongoing support!**



## UNDERSTANDING AND PREVENTING THE SPREAD OF BREAST CANCER

Centenary cancer researcher, Dr Shweta Tikoo, from our Immune Imaging program has been awarded new funding from Cancer Australia to investigate the underlying mechanisms that drive the spread of breast cancer. It is hoped that this research will identify new therapeutic targets aimed at preventing the spread of breast cancer to other parts of the body.

**Breast cancer continues to be one of the major causes of death in Australian women. It is estimated that this year alone over 3,000 Australian women succumbed to breast cancer.**

While primary breast cancer can be treated through surgery and chemotherapy, prognosis is much poorer once the cancer metastasises (spreads) to other organs. This is due in part to the rapid development of chemotherapy resistance in metastatic breast cancer cells.

Dr Tikoo said this new research project will investigate the role of "Perivascular Macrophages" in tumour cell metastasis. Macrophages are a type of immune cells which have been widely implicated in tumour progression and metastasis.

"If we can understand the spread of breast cancer at cellular and molecular level, we can develop new therapeutic targets to intervene in the process," Dr Tikoo said.

"This funding support from Cancer Australia will enable us to take a new and inventive approach to understanding and treating breast cancer, which will hopefully improve the patient outcome in future."

This funding was awarded from Cancer Australia's Priority-driven Collaborative Cancer Research Scheme, which aims to support research that reduces the impact of cancer on the community.



Researchers and members of our science support staff took part in the recent Colour Run Sydney raising money for our research.



## AND THE WINNER IS SYDNEY! TO HOST WORLD CONGRESS ON INFLAMMATION

Following a competitive international bidding process Sydney has been announced as the successful City to host the 14th World Congress on Inflammation in 2019.

The Congress will be Chaired by Professor Jennifer Gamble head of Centenary's Vascular Biology program, and Associate Professor Matt Sweet from the IMB, Queensland, with the support of a local organising committee made up of hospitals, universities and research institutes from Australia, Korea, China, Singapore, India, New Zealand and Japan.

This prestigious international conference will bring together scientists from around the world to report on and discuss the latest developments in inflammation research.

**Professor Gamble, said this is a great opportunity to showcase Australia as a world-leader in inflammation research.**

"People understand inflammation in terms of the local response for example, to a thorn in the hand during gardening. But inflammation also drives chronic diseases, such as cancer, cardiovascular disease, liver disease, diabetes, skin diseases, asthma and tuberculosis. Thus understanding how inflammation develops and why it fails to resolve is so important to our understanding of disease. Without that basic understanding of inflammation we will find it hard to develop new treatments" Professor Gamble said.

"Australia is home to some of the best and brightest minds working on understanding inflammation. This successful bid is a wonderful endorsement of Australia as a world leader in this incredibly important field of research.

"The bid was supported by the NSW Minister for Medical Research, the Hon Pru Goward MP and the NSW Minister for Tourism the Hon Stuart Ayres MP.



Dr Andrew Rochford from 7 News Sydney chatted to Dr Ben Roediger about eczema's link to asthma, food allergies and hayfever.

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