The Royal College of Pathologists of Australasia

MEDIA RELEASE **2 MARCH 2018**

Australian team global-first to use the whole epigenome and single cell genomics to study rare breast tumours

Professor Sandra O'Toole, breast cancer researcher and pathologist at the Garvan Institute of Medical Research, will speak at the Royal College of Pathologists of Australasia's (RCPA) annual conference, 'Pathology Update 2018'. She explains that, in a global first, an Australian team has utilised cutting edge technologies for whole genome epigenetic analysis and single cell genomics to study Phyllodes tumours, a rare type of breast tumour.

The results of this first assessment of genome-wide DNA methylation in Phyllodes tumours, offers a unique opportunity to identify biomarkers to better classify Phyllodes tumours and provides a strong potential for translation into improved treatment outcomes for patients.

Prof O'Toole, explains,

"Through early access to new technology, via collaboration with Dr Clare Stirzaker, Prof Susan Clark and their team at the Garvan Institute, we have been able to profile the whole epigenome of Phyllodes tumours. We have identified that there are striking differences in the patterns of methylation between malignant Phyllodes tumours and the less aggressive subtypes. We hope to exploit these differences to develop a diagnostic test for improved Phyllodes tumour stratification."

Using single cell transcriptomic technology at the new Garvan-Weizmann Centre for Cellular Genomics with A/Prof Alexander Swarbrick's team, they have also been able to analyse the very rare malignant Phyllodes tumour at a single cell level looking at all of the expressed genes from each individual cell. Previous technology only allowed study of the whole tumour as a mixture, potentially missing important variations from cell to cell. This technology is especially useful to study the response of the person (host) to the tumour, such as determining the types of immune cells present.

The study, which is funded through the National Breast Cancer Foundation and the Sydney Breast Cancer Foundation, has captured 4,500 cells from one highly aggressive malignant Phyllodes tumour with an individual read out of each cell. This has provided unique insights in relation to what drives these tumours.

"By introducing cutting edge technology to the analysis of Phyllodes tumours, we have been able to analyse the changes at the single cell level to see what makes them tick. In exciting preliminary data, we have observed that inflammation might play an important role in driving these tumours. Although it is early days, we're hopeful that with more work, an immune treatment approach may prove to be effective, as malignant Phyllodes tumours may have a very poor outcome and tend to be resistant to traditional chemotherapy and radiation treatment. We would like to follow up this study on Phyllodes tumours, and are seeking funding to be able to study these changes in larger groups of women, a critical step to move this research finding into the clinic," explains Prof O'Toole.

Phyllodes tumours are rare and there is still currently no effective treatment if the tumours cannot be controlled surgically. By utilising the latest technology to understand Phyllodes tumours, it is hoped that the study will offer improved ways to diagnose these tumours.

"For patients with these rare tumours their cancer may be misdiagnosed. The tumours can show considerable variability so a patient may have a biopsy that shows only benign tissue, but the malignant tissue could be missed. If the tumour recurs and cannot be controlled by surgery, there is a lack of effective treatments.

"The importance of research to identify better diagnostic tests and improved treatments cannot be emphasised enough. A good example of this is more common breast cancers - by studying the biological changes in the cancer cells, we understood that the hormones and pathways that drive tumour growth, can be blocked. We now have very effective treatments against these types of breast cancer, because we understand the biology.

"Conversely, rare tumours are under-analysed because they are under-recognised and patients haven't had a loud voice until recently advocating for research. If you were to add up all the individual types of rare and uncommon cancers, as a group, it would become one of the most common cancers. Unfortunately there are far worse outcomes for patients with rare cancers because we don't know much about them and therefore we can't diagnose or treat them effectively therefore further research is absolutely critical," says Prof O'Toole.

Prof O'Toole will be a speaker at the RCPA's Pathology Update 2018 conference, 'A Bridge to the Future', which takes place at the ICC Sydney from 2nd until 4th March 2018.

ENDS

About the Royal College of Pathologists of Australasia:
The RCPA is the leading organisation representing pathologists in Australasia. Its mission is to train and support pathologists and to improve the use of pathology testing to achieve better healthcare.

Media enquiries:
Dr Debra Graves
Chief Executive Officer
The Royal College of Pathologists of Australasia
+61 2 8356 5858
Debrag@rcpa.edu.au

Linsey Brown
Managing Director
The Public Relations Agency
+61 425 514 005
Linsey@thepublicrelationsagency.com.au