



# PathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA



March 2015 | Published by RCPA

Issue #045

## In This Issue

- RCPA launches world-first genetic testing standards
- Forensic pathologists use their skills to identify NPS-related deaths
- Expert guidelines needed to stem folate testing flurry
- Molecular tests to detect colorectal cancer could be a 'rescue strategy'

## Welcome to the March edition of ePathWay

Pathology Update 2015 is over for another year, but there is still a buzz in the air thanks to its huge success.

Apart from the outstanding program and speakers, the conference generated excellent media coverage with articles and radio pieces placed across Australia and New Zealand to a potential audience of over half a million people.

The four articles in this edition are from presentations given at the conference. Once you've read them, put Friday 26 to Sunday 28 February in your diary for Pathology Update 2016 A Diamond Date.

More conference coverage and information can be found on our [Facebook](#) page. And don't forget to follow our CEO Dr Debra Graves (@DebraJGraves) or the College (@PathologyRCPA) on Twitter.

## Interesting Facts

# 1290

The number of delegates at Pathology Update 2015 Conference 'Diagnosis by the Yarra'.

# 1150

The number of delegates who registered for all 3 days of the Pathology Update 2015 Conference.

## RCPA launches world-first genetic testing standards

18

The number of countries that delegates travelled from to attend the Pathology Update 2015 Conference, including Brazil, Bhutan, Israel, Sri Lanka and Qatar.

Sources: RCPA

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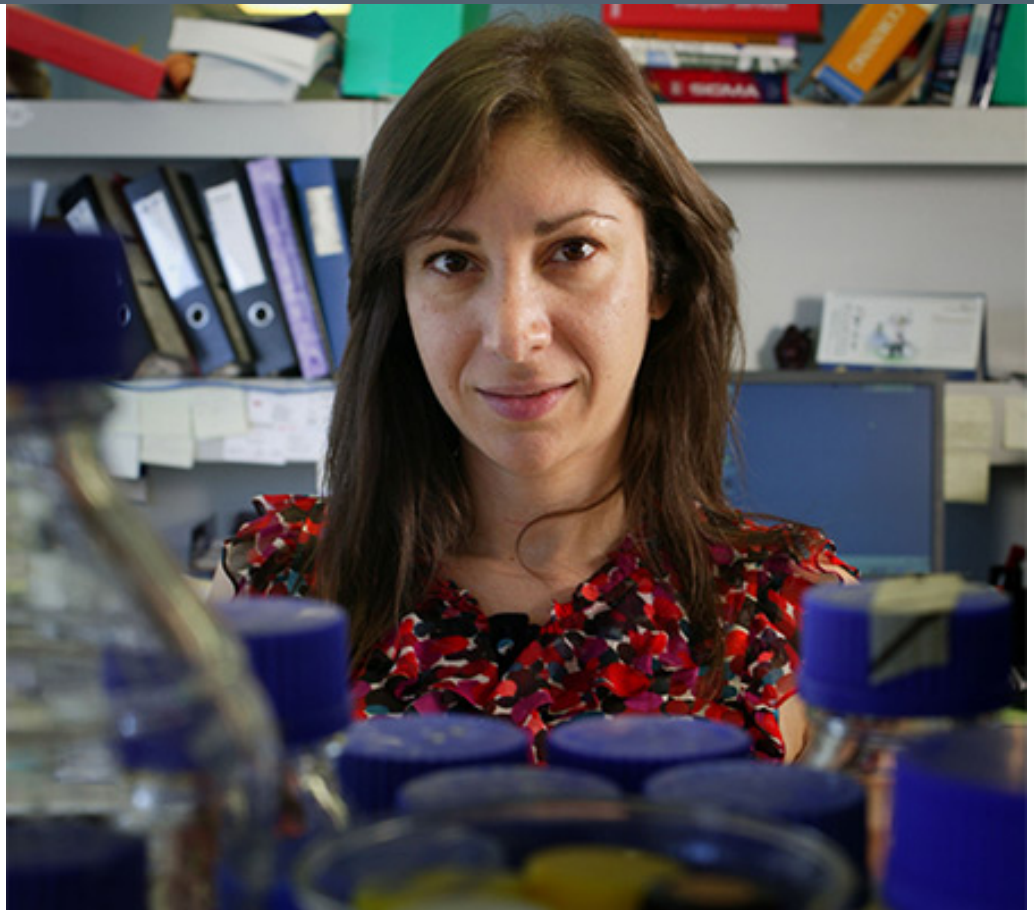
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*Dr Melody Caramins*

The Royal College of Pathologists of Australasia (RCPA), together with the Human Genetics Society of Australasia (HGSA) and the Human Variome Project, has launched a set of new standards to be applied across clinical databases used in genetic testing. These world-first standards aim to ensure a consistent approach in analysing the results of genetic tests to patients' ultimate benefit.

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## Forensic pathologists use their skills to identify NPS-related deaths

Keeping up with the illicit drug trade is like shadow boxing on a global scale. New psychoactive substances (NPS) are throwing extra challengers into the ring, and these drugs are usually legal until national controls are put in place. Fatalities from using NPS are becoming more frequent, and it often falls to forensic pathologists to identify the offending compounds through toxicology tests.



Dr Victoria Kueppers, trainee forensic pathologist at PathWest Laboratory Medicine in Western Australia, says NPS have gained international interest over the past decade as their potential threat to public health increases.

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## Expert guidelines needed to stem folate testing flurry

A recent Australian Bureau of Statistics (ABS) survey found less than one per cent of its 20,000 participants had a folate deficiency. Nevertheless, about \$60 million per year is spent on folate testing in Australia, and one expert says many of these tests are redundant.

Professor Ken Sikaris, spokesperson for the Royal College of Pathologists of Australasia (RCPA), says

increasing numbers of patients are presenting to their doctors with tiredness and weakness, and this appears to be the main driver for the continued unnecessary testing.



[read more »](#)

## Molecular tests to detect colorectal cancer could be a 'rescue strategy'

It's time to call a spade a spade; people don't like collecting poo samples. But it's also time to get over it, because a 2014 report showed that only 40 per cent of Australians over 50 years of age participated in the National Bowel Cancer Screening Program (NBCSP), despite its proven effectiveness.



Professor Graeme Young AM, gastroenterologist and guest speaker at the recent Royal College of Pathologists of Australasia (RCPA) annual scientific meeting, says the purpose of a screening test is to detect abnormalities that might require further investigation.

[read more »](#)

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## Previous Editions



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- Diagnosis by the Yarra is a record breaker
- RCPA Foundation nurtures next generation pathologists
- Fellowship awardees gain international experience in genomic medicine
- Negative test results often

## Welcome to the February edition of ePathWay

Pathology Update 2015 kicks off this Friday at the Melbourne Convention Centre. Attendance is almost at record-breaking numbers, and the outstanding program reflects this year's conference title of *Diagnosis by the Yarra*.

Go to [Pathology Update 2015](#) for more information or attend on-site if you still wish to register. We've also interviewed RCPA CEO Dr Debra Graves about this year's conference.

The RCPA Foundation funds initiatives to help us gain a greater

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## RCPA launches world-first genetic testing standards



The Royal College of Pathologists of Australasia (RCPA), together with the Human Genetics Society of Australasia (HGSA) and the Human Variome Project, has launched a set of new standards to be applied across clinical databases used in genetic testing. These world-first standards aim to ensure a consistent approach in analysing the results of genetic tests to patients' ultimate benefit.

"It's our goal to improve the quality and integrity of genetic databases and the results they help to provide," explains Dr Melody Caramins, Chair of the Genetics Advisory Committee for the RCPA. "This will help ensure patients are in a position to make better informed health decisions which may affect them, their children and their potential offspring."

To date, genetic databases have lacked any recognised international guidelines for accuracy and clinical interpretation. Only now, with financial assistance from the Australian Government Department of Health, have such standards been created. They will initially be implemented in Australia and New Zealand this year, but plans are in the pipeline to expand their reach.

"Introducing a recognised framework will provide laboratory scientists, pathologists and clinicians with a benchmark for genetic analysis, and will also enable us to begin discussions internationally with the intention to use this platform on a wider scale," explains Dr Caramins.

Spokesperson for the RCPA, Vanessa Tyrrell, spoke about the accreditation of DNA Sequence Variation Databases at the Pathology Update 2015 conference in Melbourne last month.

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Dr Victoria Kueppers, trainee forensic pathologist at PathWest Laboratory Medicine in Western Australia, says NPS have gained international interest over the past decade as their potential threat to public health increases.

“The general consensus is that these drugs are posing a threat to public health in Australia and overseas, and are linked to many drug deaths. NPS are usually marketed as legal alternatives to illicit drugs, but are often sold and purchased via underground Internet websites.

New derivatives keep emerging to avoid legislation and meet consumer demand, and they also have different street names which makes tracking them even more difficult.”

NPS are not necessarily new ‘drugs’. For example, piperazine was originally developed in the 1950s as an anti-worming agent. Its derivative 1-benzylpiperazine (BZP) was found to have amphetamine-like effects when it was studied as a potential antidepressant in the 1970s and 1980s. Piperazine’s derivatives are now marketed as ‘party pills’ or ‘legal ecstasy’.

“NPS can also be disguised as ‘research chemicals’, ‘plant food’, ‘herbal incense’ or ‘bath salts’ without appropriate labelling of the contents, let alone the amount or concentration of the substances contained in the product,” explains Dr Kueppers.

“They can also be sold as well-known illicit drugs which means people might take them thinking they are taking something else. This increases the risk of toxicity and overdose. The chemical compositions of NPS are being constantly altered as well



to try and stay one step ahead of the law.”

Dr Kueppers says synthetic cannabis is more commonly detected in post mortem toxicology tests, and it's usually found alongside other drugs such as alcohol and non-synthetic cannabis.

“Forensic pathologists need to know about NPS, including their reported effects and potential harms, so that deaths caused by these substances can be identified and reported. This requires good channels of communication between toxicology laboratories and other stakeholders, and the ability to test for the relevant substances.”

Dr Victoria Kueppers presented the talk *Beyond Booze, weed and speed: emerging drug trends* last month at the Pathology Update 2015 conference in Melbourne.

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Professor Ken Sikaris, spokesperson for the Royal College of Pathologists of Australasia (RCPA), says increasing numbers of patients are presenting to their doctors with tiredness and weakness, and this appears to be the main driver for the continued unnecessary testing.

"Folate deficiency affects only a very small section of the population with these symptoms. Overall, people who have flour in their diet are very unlikely to be folate deficient due to Australia's mandatory fortification of flour introduced in 2007," explains Prof Sikaris.

"As a result, only two groups definitely require folate testing. They are people who have coeliac disease and other gut diseases, and patients who suffer from macrocytic anaemia. We also continue to advise women to take folate supplements during conception and pregnancy as an important way to reduce the chance of the foetus developing neural tube defects."

The Australian Government introduced Medicare restrictions last November to minimise unnecessary folate testing. Despite these steps, Prof Sikaris says the rate of folate testing is too high.

"The issue we face is that B12 and folate are too often considered a combined test. If you test for one, you test for the other too. However, with the government's introduction of the additional new improved B12 tests, GP's have better ways of diagnosing B12 deficiency while avoiding unnecessary testing for folate."

Professor Ken Sikaris spoke about vitamin B12 and folate at the Pathology Update 2015 conference in Melbourne.

The RCPA will release a position statement on B12 and folate testing later this year.

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Professor Graeme Young AM, gastroenterologist and guest speaker at the recent Royal College of Pathologists of Australasia (RCPA) annual scientific meeting, says the purpose of a screening test is to detect abnormalities that might require further investigation.

"In an ideal world these tests should be simple, non-invasive and easy enough for people to undergo without anxiety or discomfort. But, by their very nature, they are not perfect."

The current front-line screening test for colorectal (bowel) cancer in Australia and New Zealand is the Faecal Immunochemical Test (FIT). It detects occult (hidden) blood in a person's stool, which can be a sign of precancerous polyps or even cancer.

"The FIT is the most accurate non-invasive simple screening test that is widely available. When positive, it identifies the subgroup of people who should go on to have a colonoscopy," explains Prof Young.

Molecular tests to detect bowel cancer are being developed, but they are not ready to replace the FIT.

"There are two types of molecular tests to detect colorectal cancer. These are stool DNA and blood DNA. The stool DNA test is more sensitive than the FIT, but is significantly more expensive. It also has a significant loss of specificity in terms of false positive results, and it's not ready for use yet," explains Prof Young.

“Blood DNA tests are not quite as sensitive as the FIT, but have more specificity. They are more expensive than the FIT but not as expensive as stool DNA tests, and because it would require a blood test and not a stool sample, might prove to be a useful ‘rescue strategy’ for people who won’t undergo stool tests or colonoscopy.”

So what is the current advice for bowel cancer screening?

“Stick with the FIT until further notice. Molecular tests for colorectal cancer are still works in progress, but if we can get a molecular signature for cancer that is not just blood, it will probably prove better than the FIT. But we know the FIT is an effective screening method in identifying colorectal cancer, and it’s still the screening test of choice.”

Professor Graeme Young AM presented the talk *Molecular tests in stool and blood for colorectal cancer* last month at the Pathology Update 2015 conference in Melbourne.

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