



# PathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA



MARCH 2014 | Published by RCPA

Issue #034

## In This Issue

- The future and the present have finally merged to create the era of molecular testing
- Are pathologists the patients' physicians?
- Genetics delivers precision medicine in cancer care
- Sudden death in young athletes

## Introduction

Pathology Update is over for another year. Pathologists from 17 countries, including the Netherlands, Kenya, France, Mauritius and China, gathered at the Melbourne Convention Centre from February 21-23 to listen to their choice of presentations delivered by 143 speakers.

Three of the articles in this month's edition are from lectures delivered at Pathology Update. They include the suggestion that pathologists have become the patients' physicians, the advent of personalised cancer genomics, and the issue of sudden death in young athletes.

The fourth article examines the impact of the genetic revolution on the way pathology and medicine is practised, which was an emerging theme at the conference.

More information from Pathology Update can be found on our Facebook page at [www.facebook.com/TheRoyalCollegeOfPathologistsOfAustralasia](http://www.facebook.com/TheRoyalCollegeOfPathologistsOfAustralasia).

You can also keep up-to-date with what's happening in pathology every day by following our CEO Dr Debra Graves (@DebraJGraves) or the College (@PathologyRCPA) on Twitter.

## Interesting Facts

# 1224

The number of delegates at this year's Pathology Update.

# 1074

The number of delegates from within Australia.

The future and the present have finally merged to create the era of molecular testing

150

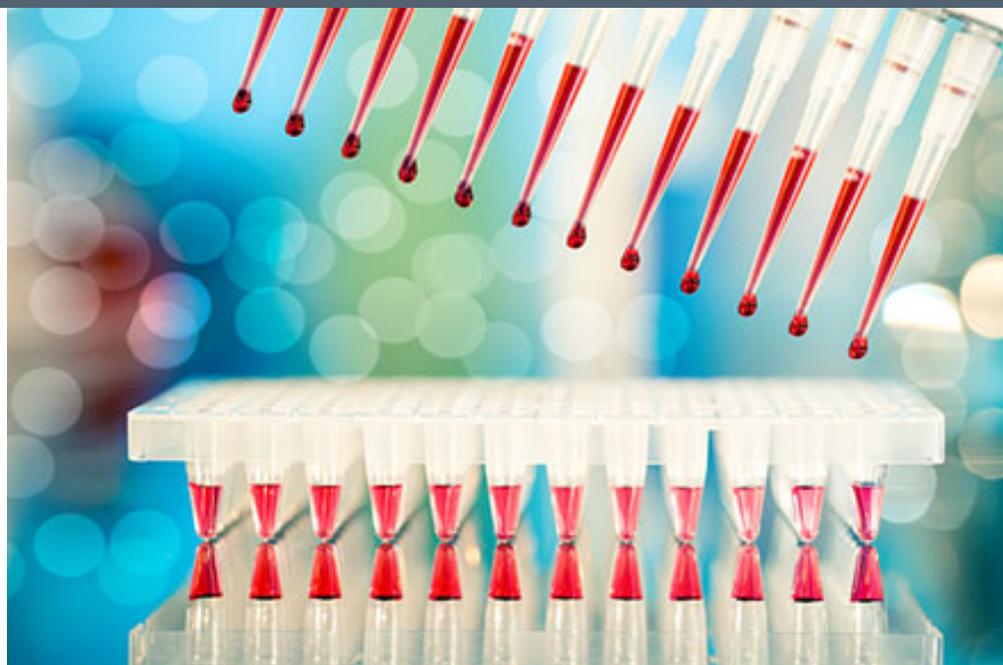
The number of delegates from overseas.

Source: RCPA

## Important Message



has an important message for you. [Click to see the message!](#)



Molecular testing once represented 'the future' of pathology and medicine. At some point the future becomes the present, and it seems that time has come. Responding to the issues that accompany the molecular testing age is the next challenge for pathologists.

[read more »](#)

## Suggest to a friend

Know someone who might be interested in this website? Why not [suggest the website](#) to them.

## Previous Editions

Did you miss something from last month? You can view our [previous editions](#) at any time.

## Subscribe Now!

Subscription is easy! Simply fill in our [subscription form](#).

## Links

[RCPA Manual](#)

[LabTest Online](#)

## Are pathologists the patients' physicians?

The traditional view of the pathologist sitting behind a microscope in the hospital's basement is being blown away by the winds of change. Technology has changed medicine, and pathologists are usually the patrons of these new technologies. Think genome sequencing, molecular biology, bioprofiling and targeted therapies – to name a few - and you get a picture of a profession that is changing medicine.



[read more »](#)

## Genetics delivers precision medicine in cancer care

The era of precision medicine has arrived. Our increasing ability to precisely characterise a person's genomic drivers for developing cancer, coupled with the rise of molecular cancer medicine, means cancer genomics has become personal.





Dr Elaine Mardis, professor of genetics and molecular microbiology, and co-director of the Genome Institute at Washington University School of Medicine, says a person's genotype can now be searched against available targeted therapies for a possible match that may deliver better treatment options.

[read more »](#)

---

## Sudden death in young athletes

While sudden and unexpected deaths during sport are rare, especially for young athletes, they do happen. It's difficult to determine the exact numbers, but each death attracts widespread attention because the person was young and healthy. International studies show that sporting participants are three times more likely to suffer from an undiagnosed cardiac (heart) condition compared to people who died at the same age for other reasons, which may increase their chance of sudden death during sport. Is this the case in Australia?



[read more »](#)

---

Copyright © 2014 The Royal College of Pathologists of Australasia

RCPA - Durham Hall - 207 Albion St Surry Hills NSW 2010 AUSTRALIA | (+61) 2 8356 5858 | [www.rcpa.edu.au](http://www.rcpa.edu.au)

[Privacy Policy](#) | [Legal](#) | [Disclaimer](#)

[Unsubscribe](#)



# PathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA



Published by RCPA

## RCPA Message



**Pathology gives life's most important answers.**



[« Back to Latest Issue](#)

Copyright © 2013 The Royal College of Pathologists of Australasia

RCPA - Durham Hall - 207 Albion St Surry Hills NSW 2010 AUSTRALIA | (+61) 2 8356 5858 | [www.rcpa.edu.au](http://www.rcpa.edu.au)

[Privacy Policy](#) | [Legal](#) | [Disclaimer](#)

[Unsubscribe](#)



# ePathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA

Published by RCPA

## Previous Editions



**FEBRUARY 2014 | PUBLISHED BY RCPA** **ISSUE #033**

---

### IN THIS ISSUE

- Annual Pathology Update a true multidisciplinary affair
- Bond's larger than life nemesis suffers from hormonal disorder
- 'Angelina effect' highlights the importance of building a sustainable and specialist genetic

### Introduction

Welcome to the first edition of ePathWay for 2014 which could be called 'the celebrity issue'!

The major event on the calendar this month is Pathology Update from February 21-23 at the Melbourne Convention Centre. Our article gives a snapshot of this event including why delegates who attend the dinner can also get up close and personal with James Bond!

Other celebrities featured this month include Richard Kiel and Angelina Jolie. Richard Kiel played the daunting steel-toothed Jaws

2014

[033 - February 2014](#)

## 2013

[022 - February 2013](#)

[023 - March 2013](#)

[024 - April 2013](#)

[025 - May 2013](#)

[026 - June 2013](#)

[027 - July 2013](#)

[028 - August 2013](#)

[029 - September 2013](#)

[030 - October 2013](#)

[031 - November 2013](#)

[032 - Dec 2013/Jan 2014](#)

## 2012

[010 - Dec 2011/Jan 2012](#)

[011 - February 2012](#)

[012 - March 2012](#)

[013 - April 2012](#)

[014 - May 2012](#)

[015 - June 2012](#)

[016 - July 2012](#)

[017 - August 2012](#)

[018 - September 2012](#)

[019 - October 2012](#)

[020 - November 2012](#)

[021 - December 2012](#)

## 2011

[001 - March 2011](#)

[002 - April 2011](#)

[003 - May 2011](#)

[004 - June 2011](#)

[005 - July 2011](#)

[006 - August 2011](#)

[007 - September 2011](#)

[008 - October 2011](#)

[009 - November 2011](#)

[« Back to Home Page](#)

---

Copyright © 2014 The Royal College of Pathologists of Australasia

RCPA - Durham Hall - 207 Albion St Surry Hills NSW 2010 AUSTRALIA | (+61) 2 8356 5858 | [www.rcpa.edu.au](http://www.rcpa.edu.au)

[Privacy Policy](#) | [Legal](#) | [Disclaimer](#)

[Unsubscribe](#)



# PathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA



Published by RCPA

---

## Subscription Form

Full Name:

Email address:

## Unsubscription Form

Email address:

[« Back to Home Page](#)

---

Copyright © 2013 The Royal College of Pathologists of Australasia

RCPA - Durham Hall - 207 Albion St Surry Hills NSW 2010 AUSTRALIA | (+61) 2 8356 5858 | [www.rcpa.edu.au](http://www.rcpa.edu.au)

[Privacy Policy](#) | [Legal](#) | [Disclaimer](#)

[Unsubscribe](#)



Prof Stewart says interpreting genetic tests is not black and white because we currently only know what less than 10% of the genome does. The role the rest of it plays in health and disease is still unknown, so there is still much to learn. He says the data from these tests needs the correct interpretation in all areas of pathology practice, and it should add value to patients' results.

"It's time to address these issues because technology has advanced so quickly that full genome sequencing is now offered in Australia for \$1500. The RCPA is ready to respond to the challenges associated with the molecular testing age, and to facilitate the changes in practice required to take pathology, and medicine, into the future."

[« Back to Home Page](#)

---

Copyright © 2014 The Royal College of Pathologists of Australasia

RCPA - Durham Hall - 207 Albion St Surry Hills NSW 2010 AUSTRALIA | (+61) 2 8356 5858 | [www.rcpa.edu.au](http://www.rcpa.edu.au)

[Privacy Policy](#) | [Legal](#) | [Disclaimer](#)

[Unsubscribe](#)



# PathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA

MARCH 2014 | Published by RCPA

Issue #034

## Are pathologists the patients' physicians?



The traditional view of the pathologist sitting behind a microscope in the hospital's basement is being blown away by the winds of change. Technology has changed medicine, and pathologists are usually the patrons of these new technologies. Think genome sequencing, molecular biology, bioprofiling and targeted therapies – to name a few - and you get a picture of a profession that is changing medicine.

“Pathology is embracing new discoveries and bridging the gap between science and clinical medicine,” explains Professor Wendy Erber, Head and Professor at the School of Pathology and Laboratory Medicine at the University of Western Australia. “Pathologists will use their understanding of the biology of the individual and of their illness to ‘personalise’ pathology.”

It could be argued this is happening already through what Prof Erber terms the ‘five P’s’ that pathologists already deliver. These stand for:

1. Pronouncements or diagnoses through integrated testing and reporting.
2. Prognoses based on patient profiles and available therapies.
3. Prescribed therapies to ensure the right drug to the right patient at the right time is given, or bioprofiling tumours, or identifying targeted therapies for breast cancer.
4. Progress of patients to monitor their response to therapy or if they have relapsed, using a multitude of tools such as tumour markers or chromosomal changes.
5. Preventing disease which mostly falls under the watch of genetic pathologists.

“There is a sixth ‘P’ which stands for post-mortem, because information from these procedures supports teaching, training

and research,” she says.

Prof Erber says patients don't want to hear information third hand anymore. They want it from the source, and that usually means from the pathologist – be it their pronouncement, prognosis, prescribed therapy, progress or preventative disease strategies.

She predicts pathologists will be in outpatient clinics in the near future to consult with patients – a situation already occurring in Toronto, Canada.

“The profession is changing around us. Pathologists are already translating scientific discoveries into patient care through methods such as biomarkers at the tissue, cellular and molecular level. Doctors and their patients seek pathologists' expertise. Pathologists know the diagnosis, they understand the patient's bioprofile and prognosis, they advise on their treatment, monitor their progress, and often know more about the patient's disease than their physician. That makes the pathologist the patient's physician.”

Professor Wendy Erber delivered the Eva Raik Plenary Lecture titled *The pathologist: The patient's physician* at Pathology Update in Melbourne on February 22, 2014.

[« Back to Home Page](#)

---

Copyright © 2014 The Royal College of Pathologists of Australasia

RCPA - Durham Hall - 207 Albion St Surry Hills NSW 2010 AUSTRALIA | (+61) 2 8356 5858 | [www.rcpa.edu.au](http://www.rcpa.edu.au)

[Privacy Policy](#) | [Legal](#) | [Disclaimer](#)

[Unsubscribe](#)



# PathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA



MARCH 2014 | Published by RCPA

Issue #034

## Genetics delivers precision medicine in cancer care



The era of precision medicine has arrived. Our increasing ability to precisely characterise a person's genomic drivers for developing cancer, coupled with the rise of molecular cancer medicine, means cancer genomics has become personal.

Dr Elaine Mardis, professor of genetics and molecular microbiology, and co-director of the Genome Institute at Washington University School of Medicine, says a person's genotype can now be searched against available targeted therapies for a possible match that may deliver better treatment options.

"Cancer genomics gives us a microscope of exceptional resolution," she explains. "In the past we dispensed chemotherapy and didn't really know who would and wouldn't respond. Now we can match the new classes of targeted therapies to the patient based on their tumour genotype."

Dr Mardis says whole genome sequencing<sup>[1]</sup> enables genetic pathologists and scientists to find cancer mutations that are unique to each patient's disease. Coupling these mutations with information from ribonucleic acid (RNA) sequencing reduces the number of candidates because not all mutated genes are expressed as RNA.

"Genes drive cancer and everyone has different drivers," she says. "Next generation sequencing<sup>[2]</sup> accelerated cancer discovery, and it is now being used in clinical translation. It gives us information that is very comprehensive and available in a short space of time which is useful in cancer treatment. It can also aid a difficult diagnosis by adding data about tumour mutations to the conventional pathology assays of tumours."

A key factor in the explosion of personalised genomic medicine is the rapidly declining cost and speed of genome sequencing. The cost plummeted from the \$billions required to sequence the Human Genome Reference only ten years ago, to just a couple of thousand dollars today. Consequently, the progress in personalised genomics has rocketed. The speed of data generation means a human genome's worth of data can be produced overnight.

The benefits to cancer medicine from this reduction of cost and time are huge. For example, Dr Mardis and her team have sequenced more than 1000 tumour/normal pairs and placed this information into the public domain.

“We are also working on the use of genomics to drive the development of personalised cancer immunotherapy,” she says.

Precision medicine enabled by genomics is now a reality. Dr Mardis says cancer patients have already received modified treatment regimes based on personalised genomics; an approach a world away from the ‘one size fits all’ cancer therapies of even five years ago. It will be interesting to see what unfolds over the next five years when even a week can be a long time in cancer genomics.

Dr Elaine Mardis delivered the Pathology Innovations lecture *Making Cancer Genomics Personal* at Pathology Update in Melbourne on February 23, 2014.

[1] Whole genome sequencing is a laboratory process that determines the complete DNA sequence of an organism's genome at a single time.

[2] Next generation sequencing is a term used to describe a number of different modern sequencing technologies.

[« Back to Home Page](#)

---

Copyright © 2014 The Royal College of Pathologists of Australasia

RCPA - Durham Hall - 207 Albion St Surry Hills NSW 2010 AUSTRALIA | (+61) 2 8356 5858 | [www.rcpa.edu.au](http://www.rcpa.edu.au)

[Privacy Policy](#) | [Legal](#) | [Disclaimer](#)

[Unsubscribe](#)



# PathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA



MARCH 2014 | Published by RCPA

Issue #034

## Sudden death in young athletes



While sudden and unexpected deaths during sport are rare, especially for young athletes, they do happen. It's difficult to determine the exact numbers, but each death attracts widespread attention because the person was young and healthy. International studies show that sporting participants are three times more likely to suffer from an undiagnosed cardiac (heart) condition compared to people who died at the same age for other reasons, which may increase their chance of sudden death during sport. Is this the case in Australia?

Dr Sarah Parsons, forensic pathologist at the Victorian Institute of Forensic Medicine, is leading a study to determine if the cause of death during sporting activity in young athletes in Australia mirrors the international data.

"A young athlete is generally defined as a person younger than 35 years of age," explains Dr Parsons. "Our retrospective study is looking at people aged five to 35 years who died during a sporting activity in the past 12 years. We included young athletes who were training, exercising or involved in a sporting activity."

Dr Parson's team identified 77 cases that fitted their criteria in Victoria, Tasmania, Northern Territory and the Australian Capital Territory (data from other states is still pending). Of these, autopsy reports found 12 died from an undetermined cause, 34 died from non-cardiac causes and 31 died from cardiac causes – mostly ischaemic heart disease (reduced blood supply to the heart) and coronary artery thrombosis (a blood clot in a main heart artery).

"One interesting fact uncovered so far is that none of the young athletes in Australia who died from ischaemic heart disease had a preexisting heart condition that would have been picked up with routine screening tests," Dr Parsons explains.

Young athletes are currently screened for predominantly cardiac abnormalities before they can participate in sporting activities in other countries.

"Pre-participation screening for young competitive athletes occurs in Europe and America to varying degrees," says Dr Parsons. "For example, if you want to participate in a competitive sport in Italy, you need to be cleared to do so by a pre-

participation screening test. About 10 percent of Italy's population is screened every year for this purpose.”

Dr Parsons says these tests usually involve an electrocardiogram (ECG) which detects cardiac (heart) abnormalities by measuring the electrical activity generated by the heart as it contracts. But, is a screening regime a double-edged sword?

“There is the question of whether restricting a young person's sporting activity will lead to increased inactivity, which can lead to obesity and possible coronary artery disease later in life. I suppose it's a benefits versus risks equation and there is no simple answer,” she says.

“There is also the controversial issue of whether pre-participation screening should even be considered in Australia. Would screening give young athletes and their families a false sense of security, and would screening tests pick up every pre-existing condition that could lead to sudden death in young athletes?”

Dr Parsons says one study of 12 to 25 year old athletes participating in competitive sport in Italy found their sporting activity enhanced their risk of death by a factor of 2.5. Early data from Dr Parson's study suggests that young males in their twenties are predominately dying from ischaemic heart disease rather than other conditions, although interpretations will be made once the data from all states is correlated.

The actual rate of sudden death in sports is also uncertain because the studies to date are retrospective, mostly carried out in America and Europe, and the exact circumstances of their death are not always known. There are also more questions than answers about sudden deaths in young athletes in Australia at this stage. Working through these issues can seem like a marathon, but it's an issue that needs investigating to produce a home run in terms of detecting and preventing sudden death in young athletes in Australia.

Dr Sarah Parsons delivered the lecture *Sudden death in young athletes* at Pathology Update in Melbourne on February 23, 2014.

[« Back to Home Page](#)

---

Copyright © 2014 The Royal College of Pathologists of Australasia

RCPA - Durham Hall - 207 Albion St Surry Hills NSW 2010 AUSTRALIA | (+61) 2 8356 5858 | [www.rcpa.edu.au](http://www.rcpa.edu.au)

[Privacy Policy](#) | [Legal](#) | [Disclaimer](#)

[Unsubscribe](#)