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Issue #033

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- 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 workforce
- Sunshine vitamin keeps raining on research

## Interesting Facts

## 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 in two Bond movies, and his size and stature is due to the hormonal disorder acromegaly. We've put this condition – not Kiel's famous alter ego - into the spotlight.

We've also looked at our genetic testing capability in the wake of Angelina Jolie's preventative double mastectomy. Her decision prompted a wave of enquiries – dubbed the 'Angelina effect' - to already stretched genetic services.

Our final article doesn't have a celebrity in sight, but vitamin D is keeping researchers on their toes. Two studies have caught the attention of the medical community, and we look at why they are of special interest.

To find out what's happening at Pathology Update, 'like' our Facebook page and join the online pathology community at [www.facebook.com/TheRoyalCollegeOfPathologistsOfAustralasia](http://www.facebook.com/TheRoyalCollegeOfPathologistsOfAustralasia), or follow our CEO Dr Debra Graves (@DebraJGraves) or the College (@PathologyRCPA) on Twitter.

Save the date: Brain Awareness Week is March 10-16 this year. You can find more information about this initiative at <http://brainfoundation.org>.

143

The number of speakers at this year's Pathology Update which runs over three days from February 21-23.

1199

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

Over 170

The number of poster displays at this year's Pathology Update.

Source: RCPA

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## Annual Pathology Update a true multidisciplinary affair



*Sean Connery relaxes on the bumper of his Aston Martin DB5 during the filming of location scenes for 'Goldfinger' in the Swiss Alps. Copyright Notice - © 1964 Danjaq, LLC and United Artists Corporation. All rights reserved.*

The logistics involved in staging the Royal College of Pathologists of Australasia's (RCPA's) annual Pathology Update are impressive. Eight specialist committees, each made up of six to eight pathologists, work on the content of the scientific program for about 10 months. This effort culminates in 143 speakers (for this year) delivering presentations across eight pathology disciplines. But it's not all work and no play, since James Bond is also on this year's 'guest' list!

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## Bond's larger than life nemesis suffers from hormonal disorder

While attendees at this month's Pathology Update<sup>[1]</sup> dinner are viewing *Designing 007: Fifty Years of Bond Style* exhibition as part of the evening at the Melbourne Museum, they will probably come 'face-to-face' with one of Bond's most memorable nemeses – steel-toothed Jaws. Played by actor Richard Kiel, his distinctive height and features are caused by a hormonal disorder called acromegaly.



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## 'Angelina effect' highlights the importance of building a sustainable and specialist genetic workforce

Angelina Jolie certainly has clout. Since she went public last year about her preventative double mastectomy because of a genetic mutation, there has been a surge in referrals to genetic pathologists to discuss cancer genetics.



Professor Graeme Suthers, genetic pathologist and recent Chair of the Genetics Advisory Committee at the Royal College of Pathologists of Australasia (RCPA), says while the 'Angelina effect' generated a three to four-fold increase in referrals to discuss cancer genetics, the demand for

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## Sunshine vitamin keeps raining on research

Vitamin D<sup>[1]</sup> has become a kind of ‘holy grail’ for medical researchers hunting an absolute link between the sunshine vitamin and non-skeletal disease. However, two new studies on vitamin D have caught the collective attention of the medical community because of their scale and intent.

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



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## Annual Pathology Update a true multidisciplinary affair



The logistics involved in staging the Royal College of Pathologists of Australasia's (RCPA's) annual Pathology Update are impressive. Eight specialist committees, each made up of six to eight pathologists, work on the content of the scientific program for about 10 months. This effort culminates in 143 speakers (for this year) delivering presentations across eight pathology disciplines. But it's not all work and no play, since James Bond is also on this year's 'guest' list!

"The conference dinner has a James Bond theme because it is being held at the Melbourne Museum where the *Designing 007: Fifty Years of Bond Style* exhibition is being held," explains Ms Eve Propper, Events and Sponsorship Manager at the RCPA. "This means the delegates will be able to wander through this exhibition as part of the dinner experience."

While the famous agent might be 'shaken not stirred' about the breadth of forensic pathology expertise present at Pathology Update - which could potentially put him out of business – pathologists from all of the pathology disciplines<sup>[1]</sup> gather together over the three days making this conference a unique annual gathering.

"It's the only multidisciplinary pathology meeting within Australasia where pathologists can mix with their colleagues from different disciplines, and have the opportunity to attend scientific sessions outside of their specialty," says Ms Propper.

Dr Ian Clark, anatomical pathologist at Capital Pathology and Chair of the Pathology Update Overseeing Committee, says this aspect of the conference is very important.

"Pathology Update is also about the business of pathology," he explains. "It's an opportunity to talk to the profession's leaders about issues involved with running pathology across Australasia."

Conferences are also about sharing knowledge, and Dr Clark says his committee awards two scholarships to international pathologists to enable them to attend Pathology Update each year.

“The Project Steering Committee assesses the applications for the scholarships. We have decided to focus on the Asia Pacific region so two pathologists from Indonesia will be attending this year’s Pathology Update as guests of the RCPA,” he says.

While the lecture theatres of the Melbourne Convention Centre will be filled with pathologists and scientists attending sessions over the three days, Ms Propper says when attendees are not in session they have the opportunity to explore the industry exhibitions set up outside the theatres.

“The industry exhibition gives delegates the opportunity to view the latest laboratory equipment and to talk to the experts – who are often scientists - about its use and application to their laboratories,” she says. “The sponsorship provided by the industry exhibition is an important component in running the conference.”

Ms Propper says Pathology Update is a continuing project. She confirms international speakers 18 months to two years in advance, while one speaker presenting at next year’s conference was booked four years ago.

“I have also booked the dinner venues for 2015 and 2016,” she explains. “Then for each conference I have a core team of about 15 people who make sure it all runs smoothly over the three days.”

This year’s Pathology Update is from February 21-23 at the Melbourne Convention Centre. More information can be found at <http://www.rcpa.edu.au/Events/Pathology-Update>.

Updates during the conference can be found at [www.facebook.com/TheRoyalCollegeOfPathologistsOfAustralasia](http://www.facebook.com/TheRoyalCollegeOfPathologistsOfAustralasia), or by following our CEO Dr Debra Graves (@DebraJGraves) or the College (@PathologyRCPA) on Twitter.

[1] The scientific program encompasses anatomical pathology, chemical pathology, forensic pathology, genetics, haematology, immunopathology, microbiology and oral and maxillofacial pathology.

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ISSUE #032

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- Solid evidence should be the catalyst for change when it comes to heart disease
- Study finds sunscreen protects our superhero gene
- Natural disasters reach further than the eye can see

### Introduction

The holiday season is here and Christmas is just days away. Before you tuck into Christmas dinner, check out our article on the facts about heart disease, dietary fats and cholesterol.

You might also slather sunscreen on with extra vigour these holidays after reading our article about how it protects our superhero gene.

We also look at how medical services – including pathology – are rebuilt after natural disasters in the wake of typhoon Haiyan. And for some families, increased genetic capability will deliver a much-anticipated outcome for their children with developmental disabilities.



## 2013

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## Bond's larger than life nemesis suffers from hormonal disorder



While attendees at this month's Pathology Update<sup>[1]</sup> dinner are viewing *Designing 007: Fifty Years of Bond Style* exhibition as part of the evening at the Melbourne Museum, they will probably come 'face-to-face' with one of Bond's most memorable nemeses – steel-toothed Jaws. Played by actor Richard Kiel, his distinctive height and features are caused by a hormonal disorder called acromegaly.

Acromegaly occurs when the pituitary gland produces too much growth hormone during adulthood<sup>[2]</sup>. The pituitary is a small gland located at the base of the brain behind the bridge of the nose. Growth hormone is important for managing physical growth, although the growth associated with acromegaly occurs so slowly the disorder often goes unnoticed for many years.

"Acromegaly is not usually identified early because the signs and symptoms evolve slowly," explains Dr Paul Glendenning, consultant endocrinologist and chemical pathologist at PathWest at the Royal Perth Hospital. "Once diagnosed, patients do become circumspect and can notice physical changes which weren't initially obvious to them. I ask patients to gather old photographs of themselves so they can see the emerging signs over time."

Dr Glendenning says the most common cause of acromegaly is a tumour in the pituitary gland. Signs and symptoms vary, but generally include enlarged hands and feet, coarsened enlarged facial features, pain and limited joint mobility, enlarged tongue and snoring due to an obstructed upper airway.

"We now know that sleep apnoea and colorectal cancer are strongly associated with acromegaly so we commonly test for these conditions as well," explains Dr Glendenning. "In fact, pathology is crucial to the diagnosis and management of

acromegaly."

Diagnosis is based on clinical assessment and the levels of growth hormone and insulin-like growth factor 1 (IGF-1) in the blood. Some people may need a growth hormone suppression test to determine whether growth hormone is suppressed by high blood sugar.

"High blood sugar levels lower or 'suppress' blood growth hormone levels in healthy people. This suppression doesn't occur in people with acromegaly and, in some cases, growth hormone paradoxically rises making this a useful test for confirming the diagnosis," explains Dr Glendenning.

Treatment options include surgery and/or radiotherapy to remove the pituitary tumour, although Dr Glendenning says medical therapy has provided further options.

"We had only surgery and radiotherapy as treatment options in the past, but medical therapy in the form of medicines, that effectively lower or block the action of growth hormone, is now available."

Another initiative in the pipeline is standardising the unit of measure<sup>[3]</sup> used to report growth hormone.

"Two different units of measure for growth hormone are currently used across Australasia, and this can cause confusion," says Dr Glendenning.

The Australasian Association of Clinical Biochemists (AACB) Endocrine Working Group has established a subgroup called the Growth Hormone Standardisation Group for Australasia (GHSGA) to find a way forward on this issue.

There have been many steps forward in the diagnosis and treatment of acromegaly since Richard Kiel appeared as Jaws in 1977 and 1979. Apart from leaving a lasting impression thanks to his memorable character, Kiel also gave this hormonal disorder a public profile - which is arguably a far greater legacy than celluloid fame.

[1] Information on Pathology Update can be found at <http://www.rcpa.edu.au/Events/Pathology-Update>

[2] Children who are still growing and produce too much growth hormone suffer from gigantism.

[3] Different units of measure is covered in these past issues of ePathWay – [ePathWay Issue 019](#) and [ePathWay Issue 027](#)

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## ‘Angelina effect’ highlights the importance of building a sustainable and specialist genetic workforce



Angelina Jolie certainly has clout. Since she went public last year about her preventative double mastectomy because of a genetic mutation, there has been a surge in referrals to genetic pathologists to discuss cancer genetics.

Professor Graeme Suthers, genetic pathologist and recent Chair of the Genetics Advisory Committee at the Royal College of Pathologists of Australasia (RCPA), says while the ‘Angelina effect’ generated a three to four-fold increase in referrals to discuss cancer genetics, the demand for services had already outstripped supply.

“Hats off to Angelina for raising such an important and beneficial issue, but there is still a way to go to get the balance right in terms of demand for tests against workforce capacity and capability,” he explains.

“For example, we don’t know most of the genes associated with a familial predisposition to breast cancer yet, so there is only a very small proportion of women where genetic testing will give them clinically useful information at present.”

The RCPA is committed to building solid foundations to ensure the unfolding benefits of the genetic revolution can flow to as many people as possible. High on the College’s ‘to do’ list is working with the government to have genetic pathology recognised as a specialty area of pathology.

“This is a very important issue because it means those doctors who train as genetic pathologists will have a career path in this essential specialty. This also means we will retain these specialists in the area of genetics because they will have a career in a recognised specialty,” explains Dr Debra Graves, CEO of the RCPA.

Dr Graves says the RCPA is actively identifying possible sites for future trainees and specialists in genetic pathology to ensure Australia and New Zealand can continue to participate in, and contribute to, the benefits of advances in genetics.



“There are only 17 genetic pathologists in Australia and New Zealand at present. We have identified that we need about 22 more to ensure we can meet, and keep up with, the demand for genetic testing,” explains Dr Graves. “These pathologists will then continue to build capacity and capability through training the next generation of genetic pathologists.”

Genetic scientists already play a vital role in providing high quality care, and more are needed to ensure sustainability in the future. Prof Suthers says assembling the right workforce and resources is vital to not only handle the current workload, but to also respond to future demand.

“We also need to design an appropriate pathway to educate GPs about when and how to guide patients towards meaningful genetic testing. This is because many of these important primary care doctors were in medical school before the genetic revolution happened,” he says. “We also need to build a fairer system of billing for genetic tests so that people are not disadvantaged according to where they live.”

The evolving field of genetics certainly presents challenges, especially when advances occur so rapidly that it seems like the road is being paved as we go. But there are also enormous gains to be had, especially in the area of preventative health and targeted treatments. Creating a sustainable specialist genetic workforce is therefore vital to ensure the genetic revolution delivers clinically useful information to current and future generations.

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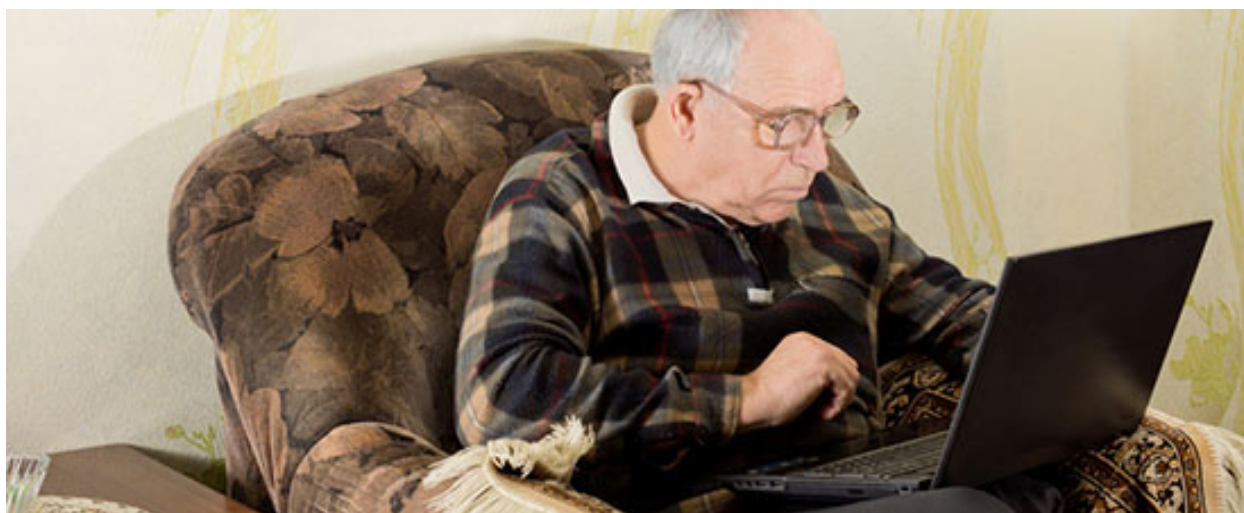
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## Sunshine vitamin keeps raining on research



Vitamin D<sup>[1]</sup> has become a kind of 'holy grail' for medical researchers hunting an absolute link between the sunshine vitamin and non-skeletal disease. However, two new studies on vitamin D have caught the collective attention of the medical community because of their scale and intent.

"There are about 36,000 studies published on vitamin D so far, and about 4000 of these were published last year alone," explains Dr Paul Glendenning, consultant endocrinologist and chemical pathologist at PathWest at the Royal Perth Hospital. "It's hard to keep up with that amount of information, but these two studies do stand out because one attempted to minimise previous research bias, and the other is large enough to provide meaningful data."

One study is a systematic review published in *The Lancet Diabetes and Endocrinology*<sup>[2]</sup> last month. Lead author Philippe Autier and colleagues reviewed the evidence from 290 observational studies which suggest that high serum concentrations of vitamin D confers a protective effect, and compared these findings to the results of 172 randomised controlled trials. They concluded that low vitamin D is not a cause but a consequence of ill health.

The Lancet's editorial on this study says large clinical trials to assess the effects of vitamin D on non-skeletal health problems are justified given the large number of people who take vitamin D supplements. But, it cautions that the scale of further research must be large enough to produce clinically relevant outcomes to avoid the neutral effect reported by most clinical trials to date. This is why the D-Health study about to kick off in Australia is potentially significant.

The D-Health study will follow 25,000 Australians aged 60-79 years for five years to establish the role vitamin D plays in preventing a range of diseases. It is being coordinated by the QIMR Berghofer Medical Research Institute in Brisbane. Dr Glendenning says this study will look at both skeletal and non-skeletal diseases, and will hopefully provide answers about many aspects of vitamin D including whether it has local or paracrine actions in the body.

Although vitamin D has been studied, debated, studied again, debated again and so on for a number of years, Dr Glendenning says there is still no evidence to support widespread screening or treatment with supplements.

The Royal College of Pathologists of Australasia (RCPA) released a position statement<sup>[3]</sup> on the *Use and Interpretation of Vitamin D Testing* last year. It asserts that testing should only be used to detect cases where treatment is likely to be of benefit such as in osteoporosis or other medical conditions, or severe and chronic lack of UVB sunlight exposure through dark skin pigmentation, or being confined indoors.

Since the final results of the D-Health study won't be known for many years, countless research studies will be published during that time if current trends continue. But until research proves an absolute link between vitamin D and non-skeletal disease, the sunshine vitamin will continue to rain on the research, and keep its complex role in health and disease hidden behind the clouds.

[1] Vitamin D of measure is covered in these past issues of ePathWay – [ePathWay Issue 003](#) and [ePathWay Issue 026](#)

[2] Autier, Philippe, Mathieu Boniol, Cécile Pizot, and Patrick Mullie. "Vitamin D Status and Ill Health: A Systematic Review." *The Lancet Diabetes & Endocrinology* 2.1 (2014): 76-89. Print.

[3] The position statement can be found at <http://www.rcpa.edu.au/Library/College-Policies/Position-Statements/Use-and-Interpretation-of-Vitamin-D-Testing>

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