



# PathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA



AUGUST 2018 | PUBLISHED BY RCPA

ISSUE #083

## IN THIS ISSUE

- Sepsis, a medical emergency
- Pathology, it's in the blood
- Pathology and cancer research
- RCPA's concerns over direct to consumer genetic testing

## INTERESTING FACTS

### 8 Million

The number of deaths caused by sepsis worldwide annually<sup>1</sup>.

### 50%

The percentage by which early recognition can reduce sepsis mortality<sup>1</sup>.

### 100%

The percentage of cancers diagnosed by pathologists.

Source:

[1] <https://www.world-sepsis-day.org/about/>

## Welcome to the August 2018 edition of ePathWay

This month's issue of *ePathway* will look at the following:

- Sepsis, a medical emergency
- Concerns over direct to consumer genetic testing
- Daffodil Day 2018
- Pathology, it's in the blood

Sepsis is a medical emergency and remains one of the main causes of death worldwide. It is a potentially life threatening response by the body to infection and can be triggered in any part of the body. We speak to Dr Timothy Gray ahead of World Sepsis day which takes place on 13 September.

In a new feature for ePathway, 'Pathology, it's in the blood' takes a look at families who have more than one pathologist. In this issue, brothers Doctor Ian Beer and Doctor Brian Beer discuss how they both came to be in the pathology profession.

Ahead of Daffodil Day 2018, we speak to Anatomical Pathologist Associate Professor Chris Hemmings to discuss the important role that pathologists play in the diagnosis and management of cancer, and the impact that national fundraising days, such as Daffodil Day, have on the vital area of cancer research.

And finally, to the world of genetics and direct to consumer genetic testing in newborn babies: the RCPA explores these complex medical tests and its new position statement in this area.

Remember to follow us on [Facebook](#) (@TheRoyalCollegeofPathologistsOfAustralasia), Twitter (@PathologyRCPA) or on Instagram (@the\_rcpa). CEO, Dr Debra Graves can be followed on Twitter too (@DebraJGraves).

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## Sepsis, a medical emergency

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We spoke to Dr Timothy Gray, Staff Specialist Microbiology and Infectious Diseases at Concord and Canterbury Hospitals, ahead of World Sepsis Day, an initiative by the Global Sepsis Alliance. The awareness day takes place on 13 September each year, providing an opportunity for people around the world to unite in the fight against sepsis.

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## Pathology, it's in the blood

Doctor Ian Beer is an anatomical pathologist and is Director/Managing Pathologist at Pathlab Waikato, New Zealand. His brother, Doctor Brian Beer is a forensic pathologist, based in NSW, Australia at Newcastle Department of Forensic Medicine, John Hunter Hospital. Growing up in a small provincial town in New Zealand, the brothers both attended high school in Christchurch before going on to study medicine in Auckland, with Ian becoming the first to enter into the profession, and Brian following in his footsteps three years later.



*L-R Dr Brian Beer and Dr Ian Beer*

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## Pathology and cancer research

It is a little known fact that pathology affects each and every one of us. Pathology determines the cause, origin and nature of disease and, sometimes unknowingly, touches the lives of many during their lifetime, particularly those that have been affected by cancer.



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## RCPA's concerns over direct to consumer genetic testing

The Royal College of Pathologists of Australasia's (RCPA) forthcoming new position statement on genetic testing is due to be finalised and released next month. It will outline the College's stance on genetic tests that are marketed directly to consumers, genetic testing for medical purposes, and genetic testing for non-medical purposes, important considerations for all genetic tests and important additional considerations for all direct to consumer tests. The statement will also summarise categories of genetic testing in order to assist in discussions of which tests are, or are not, appropriate for access through a direct to consumer route.



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## Sepsis, a medical emergency



We spoke to Dr Timothy Gray, Staff Specialist Microbiology and Infectious Diseases at Concord and Canterbury Hospitals, ahead of World Sepsis Day, an initiative by the Global Sepsis Alliance. The awareness day takes place on 13 September each year, providing an opportunity for people around the world to unite in the fight against sepsis.

Dr Timothy Gray said:

“Sepsis is a medical emergency and remains one of the main causes of death worldwide. Sepsis occurs when chemicals released by the body to fight infection trigger inflammatory responses, resulting in damage to tissues and organs and causing them to fail. It is a potentially life threatening response by the body to infection and can be triggered in any part of the body. Most commonly it is caused by bacteria; however, viral and fungal infections can also cause sepsis.

“A person with sepsis will often present with a high fever, but occasionally the body will respond by dropping to a low temperature. A person with sepsis may find their heart racing or have a rapid breathing rate. A person with severe sepsis may be confused, have difficulty breathing and have pain at the site of infection. If no treatment is sought, the sepsis patient’s blood pressure will drop, which can lead to dizziness or sometimes collapse.”

Sepsis accounts for at least 8 million deaths worldwide annually<sup>[1]</sup>. Those who are at the highest risk include the very young and the very old, as well as people with an impaired immune system, and those suffering from a chronic or serious illness such as diabetes or cancer.

“While anybody can develop sepsis it is most common in the elderly and those with



weakened immune systems. It is also more dangerous in these individuals. In Australia, approximately 12% of patients admitted to the Intensive Care Unit have a diagnosis of sepsis. With early recognition, though, the clinical course can be changed and the majority of patients with sepsis do not require admission to the intensive care unit”, said Dr Gray.

Sepsis should be treated in-hospital as a medical emergency, as quickly and efficiently as possible. A 2006 study showed that the risk of death from sepsis increases by 8% with every hour that passes before treatment begins [2]. Early recognition and treatment reduces sepsis mortality by 50% [1].

“The laboratory plays a central role in diagnosing sepsis and is important in guiding prompt and appropriate treatment. Many branches of pathology, including biochemistry, haematology and microbiology, all play key roles in diagnosing sepsis. In particular, the microbiology laboratory focuses on detecting the microorganism (most commonly a bacterium) that is causing the triggering infection. The microbiology results help guide the choice of an appropriate antibiotic.

“There has been a renewed focus in the healthcare system to ensure that a person with sepsis is recognised early and timely management is implemented. The use of appropriate antibiotics as soon as sepsis is recognised has been shown to be a key intervention.

“The treatment of sepsis has not changed for a long time. The mainstay of treatment involves the prompt administration of antibiotics, removal of infected devices or drainage of pus, intravenous fluid therapy, organ system support with drugs that support blood pressure, mechanical ventilation and dialysis as required, and oxygen if levels are low. It is important to perform investigations, such as culturing the blood, early, and to establish the site of infection, as this can help guide treatment.

“Most patients with sepsis respond to treatment and do well. Dying from sepsis is more common among the elderly, those with pre-existing medical problems and those who require admission to the intensive care unit. In fact, of patients admitted to the intensive care unit with sepsis, approximately one in five will succumb despite the best treatment,” said Dr Gray.

“One of the major challenges going forward is the increased number of drug resistant bacteria. Although Australia has remained a lucky country with generally lower rates of drug resistant bacteria compared to the rest of the world, the numbers are increasing. When an infection caused by a drug resistant bacterium triggers sepsis, this can be more difficult to treat and the outcome for the person with sepsis is worse. This is a major challenge for all people involved with the treatment of sepsis. The microbiology laboratory plays a key role in identifying these drug resistant bacteria and supporting doctors in the treatment of their patients with sepsis,” said Dr Gray.

[1] <https://www.world-sepsis-day.org/about/>

[2] <https://www.australiansepsisnetwork.net.au/community-awareness/treatment>

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## RCPA's concerns over direct to consumer genetic testing



The Royal College of Pathologists of Australasia's (RCPA) forthcoming new position statement on genetic testing is due to be finalised and released next month. It will outline the College's stance on genetic tests that are marketed directly to consumers, genetic testing for medical purposes, and genetic testing for non-medical purposes, important considerations for all genetic tests and important additional considerations for all direct to consumer tests. The statement will also summarise categories of genetic testing in order to assist in discussions of which tests are, or are not, appropriate for access through a direct to consumer route.

Ahead of the new position statement being released, we spoke to Dr Melody Caramins, Chair of the RCPA Genetics Advisory Committee, who, along with five other medical professionals/pathologists, wrote the soon-to-be released position statement.

"Genetic factors are recognised to play a role in almost every aspect of health and disease. Our understanding of these genetic influences is increasing, as is our ability to test for them. Many people are interested in using genetic tests; however, genetic tests can be utilised for medical and non-medical purposes. They can be used to determine ancestry, predict medication sensitivity, predict the likelihood of developing particular diseases and of passing this predisposition to their children, and identify, in the oncology setting, acquired genetic changes which may help determine prognosis or treatment. However, genetic test results can have significant health implications, not only for the individual being tested, but potentially also for their relatives when testing for heritable genetic changes," said Dr Caramins.

“There are many important considerations for all genetic tests. For those which are used for any medical purpose it is a legal requirement in Australia that all such tests must be performed in an accredited laboratory. This ensures that the results are analytically correct and meet appropriate quality standards, and that the test meets criteria for scientific validity. It also ensures that there is appropriate clinical supervision and oversight of the testing process and its interpretation. For other genetic tests that may not necessarily be used for a medical purpose, it is strongly recommended that such tests should also be performed in an accredited laboratory, for the same reasons of ensuring appropriate standards of analytical accuracy and quality.”

Last month, the RCPA expressed concern over direct to consumer genetic testing for newborns. This followed recent reports that a new online genetic test was available direct to parents which can identify over 63 treatable conditions which occur in children, babies, and young adults.

The RCPA strongly advocates that complex medical tests always be requested by, and after discussion with, an experienced medical practitioner or other appropriately qualified health practitioner. This approach applies to all medical tests. It is particularly relevant for complex genetic tests that predict the medical future of a child. It is not appropriate for genetic tests that deal with significant clinical issues to be marketed directly to patients.

“We are greatly concerned at reports indicating that direct to consumer genetic testing for babies is taking place without the parents having had a discussion with their doctor prior to the test being done. The RCPA acknowledges that expanded screening of newborns could potentially provide information that may assist parents in caring for their child or making further reproductive plans. The College also supports the broad provision of information about genetic tests to the public. However, the provision of tests which predict the future of a person’s health carries significant ethical and social implications and requires appropriate professional oversight,” said Dr Caramins.

The development of a newborn screening test requires careful consideration in order to assess what the test will achieve for the child and parents. Some of the key considerations that should be addressed before providing such a test are as follows:

- The selection of and justification for the genes included in the test;
- The performance of the test in terms of both detecting genetic mutations and in the impact on the child’s health. Note that not all mutations cause disease.
- The benefit or overlap of the genetic test, versus current newborn screening programs that are already provided free nationwide;
- The integration of such tests in the overall patient care pathway, including access to other tests, medical assessment, and treatments that may be required;
- Consultation with professionals involved in the established care pathways for babies, such as obstetricians, clinical geneticists, paediatricians, and pathologists;
- The education of requestors; and
- Access to professional genetic counselling.

The National Pathology Accreditation Advisory Council (NPAAC) dictates the standards required of all Australian laboratories delivering medical genetic tests. The accreditation standard, *‘Requirements for Medical Testing of Human Nucleic Acids’*, states that a key consideration prior to initiating a medical genetic test is that *‘The Laboratory must provide medical nucleic acid testing only in the context of a clinical service provided by a medical practitioner.’*<sup>[1]</sup>

The National Health and Medical Research Council (NHMRC) has also released concise guidelines on direct to consumer (DTC) genetic testing, which highlight the importance of professional involvement and education. It explains that *‘The NHMRC encourages the involvement of health professionals, including medical practitioners, clinical geneticists and genetic counsellors in the genetic testing process’* and *‘Professional involvement is important in providing individuals and their families with balanced information and an appropriate clinical context.’*<sup>[2]</sup>



Dr Caramins concluded,

“The standards for good medical practice in pathology place the patient’s welfare at the centre of all that we do. To ensure that this focus is maintained, it is essential that an experienced and independent medical practitioner be involved in guiding the selection, requesting, and use of complex medical tests. The RCPA does not support any organisation which offers direct to consumer testing without the involvement of the patient’s medical practitioner.”

References:

[1] National Pathology Accreditation Advisory Council, Requirements for medical testing of human nucleic acids, Second edition 2013, NPAAC Tier 4 Document.

[2] Direct to Consumer Genetic Testing, A statement from the National Health and Medical Research Council (NHMRC), December 2014

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## Pathology and cancer research



It is a little known fact that pathology affects each and every one of us. Pathology determines the cause, origin and nature of disease and, sometimes unknowingly, touches the lives of many during their lifetime, particularly those that have been affected by cancer.

Pathologists are specialist medical practitioners who study the cause of disease and the ways in which diseases affect our bodies by examining changes in the tissues and in blood and other body fluids. Some of these changes show the potential to develop a disease, while others show its presence, cause or severity or monitor its progress or the effects of treatment.

Pathologists are constantly working 'behind the scenes' to diagnose 100 percent of all cancers. They play a pivotal role in the diagnosis, treatment and management of cancer.

This year, Daffodil Day will take place across Australia on Friday, 24 August 2018 and in New Zealand on 31 August 2018, to raise funds for life-saving cancer research.

We spoke to Anatomical Pathologist, Associate Professor Chris Hemmings, Department of Anatomical Pathology, Canterbury Health Laboratories, to discuss the important role that pathologists play in diagnosing and managing all cancers, the importance of awareness days, such as Daffodil Day, and the role of pathology in cancer research.

"Essentially all cancer is diagnosed by a pathologist – be it blood cancers which are normally diagnosed by haematologists, or cancers of solid organs (including those in children) which are normally diagnosed by anatomical pathologists, which is what I am. I diagnose cancer every day in my job, but there's more to it than that – we also assess cancers that have been removed through surgery to determine not only the particular

type of cancer but how bad it is, how far it has spread and other factors that may influence treatment – such as genetic testing to determine susceptibility to particular drugs or other therapy.

“We also contribute to multidisciplinary cancer meetings, which is where all the doctors involved in cancer care (pathologists, radiologists, surgeons, oncologists) and other healthcare workers (cancer nurses, social workers, dietitians, etc.) get together to determine the best treatment for a particular patient. We can also advise on the best type of biopsy to make the diagnosis in the first place.”

Awareness days aim to immerse the public in important health information, promote efforts at prevention, and raise money for the cause. Daffodil Day is one of the best recognised awareness events with so many people knowing the significance of the yellow flower. Each year, supporters and volunteers come together in the community to sell daffodils and merchandise and to collect donations which help to fund vital research.

“Daffodil Day is a fantastic awareness day. What it does very well is raise general awareness of cancer in the community, educate people about both prevention (for example, not smoking, weight control and moderation of alcohol intake) and also the warning signs that should prompt them to see their doctor and of course raise important funds for cancer research too.

“In addition to Daffodil Day, which focuses on cancer broadly, there are also a number of other groups that focus on specific cancer types. These groups help to support patients and their families, and also organise fundraising efforts to support research. Of course, there’s a limited pool of research funding out there, so tailored grants can be a big help in supporting research into these less well known cancers. For example, the Unicorn Foundation is a great support for people with neuroendocrine (“carcinoid”) tumours, and several groups such as Rainbows For Kate do the same for sarcoma cancers. In my particular interest areas of rare cancers, these groups are particularly helpful in providing research funding for areas that are less well known in the public arena and tend not to attract the big dollars,” said A/Prof Hemmings.

“Pathology is fundamental to cancer research – in terms of understanding the biology of cancer, developing and tailoring new treatments, and also in supporting other forms of cancer research such as clinical trials. The first step in researching a cancer is to make sure that the diagnosis is actually correct, and that requires a pathologist. As well as conducting our own research, we often contribute to other research being performed by surgeons, oncologists, drug companies and so on. Pathologists also support the collection of tissues for research – such as The Canterbury Tissue Bank here in Christchurch.”

A/Prof Hemmings is a surgical pathologist with subspecialty interests in gut pathology and in rare cancers including sarcoma, GIST and neuroendocrine tumours. Her current primary research focus is aspects of radiation response in locally advanced rectal cancer.

“There are so many interesting projects going on, it’s hard to pick just a few to mention. My own area of research interest has been in looking at why different rectal cancer patients get a better or worse response to radiation treatment. I have found a particular marker that seems to predict for good or poor response, but now I need to go on and verify that finding in a second study, to prove that it wasn’t just a one-off for some reason.

“Other researchers here in Christchurch have recently identified a particular type of bacteria that, if present in the bowel, seems to predispose to the development of bowel cancer. If that finding holds true, it has huge implications for monitoring people at risk of one of our most common cancers. It’s a hugely exciting and rewarding area but it can also be frustrating because we want to just get on and find the answers, and you have to go down a lot of blind alleys before you get the results,” said A/Prof Hemmings.

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## Pathology, it's in the blood



Doctor Ian Beer is an anatomical pathologist and is Director/Managing Pathologist at Pathlab Waikato, New Zealand. His brother, Doctor Brian Beer is a forensic pathologist, based in NSW, Australia at Newcastle Department of Forensic Medicine, John Hunter Hospital. Growing up in a small provincial town in New Zealand, the brothers both attended high school in Christchurch before going on to study medicine in Auckland, with Ian becoming the first to enter into the profession, and Brian following in his footsteps three years later.

Ian Beer:

“Brian and I both went to Shirley Boys’ High School in Christchurch which was a very good school in terms of encouraging work ethic and training. Following this, I lived in Auckland whilst I was on the general pathology training program and Brian followed along two years after me – he thought it was a good idea too. We both secured jobs in provincial towns, I got a job in Tauranga, and he ended up in Gisborne initially and life changed from then on! I have now become an anatomical pathologist with some coronial pathology which means I do Coroner’s autopsies.

“We don’t come from a professional family; my parents ran a grocery shop before the supermarkets came in and ran them out of business. I think a good dose of sibling rivalry led us into medicine. I was first, but I suspect Brian followed in my footsteps to do something similar.

“My first appointment was to Greenlane Hospital as a Surgical Registrar with the view that a budding surgeon should understand what is required from a pathologist, as surgeons send a lot of pathology to the lab. Effectively, I had about six months there



while I realised that becoming a Surgical Pathologist was really what I wanted to be.

“I wouldn’t say that Brian and I have similar personalities but I guess the one thing you need to become a pathologist is to be fixated on detail, which we both are. There are quite a few people that I can think of in whom pathology runs in their family, both siblings and parents and their children. I think it’s probably an example of wanting to do something similar to your sibling/parent, combined with finding that it is intrinsically interesting. I’m 65 now and I’m still enjoying it. Every day there are a new set of challenges, certainly diagnostic challenges that really intrigue me. There’s a lot of routine, of course, but I guess around 5% of the work will be quite challenging and I’m very grateful to have colleagues to show things to.

“Pathology is the hinge on which the diagnostic door turns. You can’t really practise medicine without pathology. Every cancer diagnosis has a pathology test or examination to determine what kind of cancer it is and to assess how it should be managed. More broadly, there are a huge variety of tests performed on a patient to arrive at a diagnosis, which are often based on blood chemistry, immunology, microbiology and haematology, looking at the white cells or red cells and so on.

“And now of course, genomics is becoming a major focus and that applies to almost every discipline because there is a genetic basis to a large number of diseases, especially cancers.”

Brian Beer:

“My earliest memories of Ian are from the town in New Zealand that we grew up in. Our parents owned a dairy (a corner shop) and my earliest memories go back to then and also going to our uncle or granddad’s farms for the holidays, particularly in August which was lambing season. Some of my clearest and vivid memories are of my brother and I helping my uncle or granddad with lambing early in the morning with frost everywhere and lambs running around bleating.

“I was very interested in pathology from medical school and Ian had also made a start in pathology during my latter years in medical school which put the specialty onto my radar as what to do. As part of my last year at The University of Auckland School of Medicine, I did the final year three-month elective in the forensic department. At that particular time the Air New Zealand DC-10 air crash into Mt Erebus in the Antarctica occurred and I was a small part in the forensic team doing the DVI (Disaster Victim Identification) exercise for that. This unique experience whetted my appetite for pathology and into general pathology training.

“Doctors have skill sets/personality traits that suit them to a particular specialty, and in anatomical and autopsy pathology, pattern recognition and attention to detail are critical, which fits with the way my brain operates. This, along with my initial experience of post mortems particularly, drew me to pathology.

“I have practised as a general pathologist for most of my career in regional towns where there is a need for someone who can cover all disciplines in the lab and often as part of that role, I would do the Coronial post-mortems. Later I was able to easily transition to forensic pathology. Training as a general pathologist has allowed me a lot of flexibility in my pathology career over the years.

“Pathology is critical to the current practice of medicine, underpinning many of the advances in medicine, which is something most people don’t appreciate as the discipline is not visible to them. If you didn’t have a good quality laboratory system in healthcare, much of what doctors currently do now wouldn’t be possible.”

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