ePathWay
THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA

MARCH 2013 | Published by RCPA

Issue #023

In This Issue

Interesting Facts

149

The number of speakers at the Pathology Update 2013 Melbourne under the Microscope conference held from 22-24 February at the Melbourne Convention Centre

Introduction

This month's edition is inspired by the Pathology Update 2013 Melbourne under the Microscope conference held from 22-24 February at the Melbourne Convention Centre. Three of the articles - SIDS and genetics, orphan diseases and rotavirus – were written from presentations at the conference, while the fourth article looks at pathology registrar training which is also an important component of the conference. Trainee pathologists can meet their examiners in dedicated 'Meet the Chief Examiner' morning sessions.

The topics at this year's conference also generated interest from both consumer and trade media with around 50 articles secured across media outlets including The Australian, The Sydney Morning Herald, The Australian Financial Review, ABC News, medical trade publications and websites.

It was also a perfect chance to road test the college's first steps into the world of social media. Twitter was a firm favourite for delegates who were using #PathUpate and #RCPA to tweet about a wide range of subjects from the conference. Top 'tweeters' were our very own CEO, Dr Debra Graves, medical journalists from Australian Doctor, Medical Observer and New Zealand Doctor, and our public relations account director Linsey Brown from S2i Communications.

And don't forget, you can always get the latest news from the RCPA by 'liking' their Facebook page www.facebook.com/TheRoyalCollegeOfPathologistsOfAustralasia, and by following Dr Graves on Twitter (@DebraJGraves).

SIDS and genetics – are we there yet?

1223

The total number of delegates at the Pathology Update 2013 conference

133

The number of delegates who travelled to the Pathology Update 2013 conference from overseas including Canada, China, Fiji, Germany, Ireland, Malaysia, New Zealand, Papua New Guinea, Philippines, Saudi Arabia, Singapore, South Africa, Sri Lanka, Thailand, UK and USA

Source: RCPA

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The first international conference on Sudden Infant Death Syndrome (SIDS) was held in Seattle in 1963. Fifty years later, advances in genetics may help to identify some of the causes, but we're not there yet.

"Genetic causes of SIDS are apparently uncommon but are probably under-represented at this stage," explains Professor John Christodoulou, Director of the Western Sydney Genetics Program based at the Children's Hospital at Westmead in Sydney. "We still need a careful family and personal history, and a timely post mortem is critical, but next generation sequencing (NGS) technologies look set to transform the execution of a 'genetic autopsy'."

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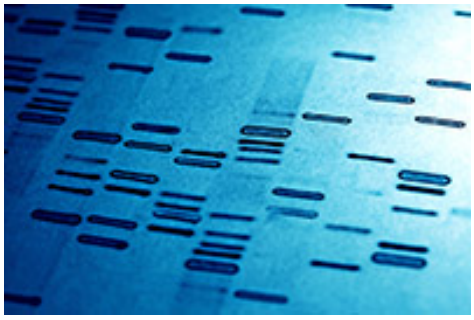
The genetic revolution offers hope to the 'orphans' of the health system



The paradox about rare or 'orphan' diseases is that, collectively, they are not uncommon. There are thousands of identified rare diseases affecting about 350 million people worldwide. This includes six to eight percent of all Australians, which equates to about 1.2million people. Yet diagnosis can be difficult, especially if there are perhaps just five other known cases of a particular disease in the world.

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Rotavirus update highlights the importance of its discovery 40 years ago

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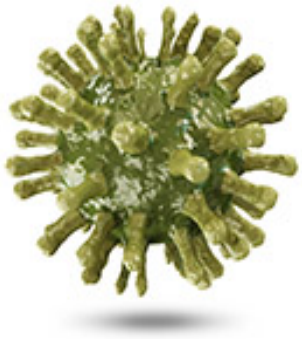

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Pathology training in Australasia leads to world class graduates

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022 - February 2013

021 - December 2012

020 - November 2012

019 - October 2012

018 - September 2012

017 - August 2012

016 - July 2012

015 - June 2012

014 - May 2012

013 - April 2012

012 - March 2012

011 - February 2012

010 - December 2011/January 2012

009 - November 2011

008 - October 2011

007 - September 2011

006 - August 2011

005 - July 2011

004 - June 2011

003 - May 2011

002 - April 2011

001 - March 2011

« Back to Home Page

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SIDS and genetics – are we there yet?

The first international conference on Sudden Infant Death Syndrome (SIDS) was held in Seattle in 1963. Fifty years later, advances in genetics may help to identify some of the causes, but we're not there yet.

"Genetic causes of SIDS are apparently uncommon but are probably under-represented at this stage," explains Professor John Christodoulou, Director of the Western Sydney Genetics Program based at the Children's Hospital at Westmead in Sydney. "We still need a careful family and personal history, and a timely post mortem is critical, but next generation sequencing (NGS) technologies look set to transform the execution of a 'genetic autopsy'."

NGS refers to the development of high-throughput sequencing technologies that can produce thousands of gene sequences at once. It is the result of major technological advances in sequencing technologies leading to a marked reduction in the cost of DNA sequencing.

One example of NGS is the potential for whole exome sequencing to focus on a set of genes that are known to be associated with a particular clinical problem, such as genes known to cause epileptic encephalopathy (a severe brain disorder) in children.

Prof Christodoulou says the genetic causes of SIDS have so far focused on two aspects: 1) identification of genetic predisposing factors, and 2) finding primary genes that caused the death.

“There are proposed classes of genes which mainly predispose people to SIDS. Previous studies have implicated abnormalities in nicotine metabolising enzymes, serotonin transport, regulation of the autonomic nervous system, regulation of inflammation and the regulation of energy production,” he says. “Changes in some of these genes have also been associated with an increased risk of developing SIDS, although this area remains controversial because of conflicting results in multiple studies.”

On the other hand, Prof Christodoulou says single genes may be a major cause of SIDS and could account for up to 10–20% of cases. One example is the long QT syndrome (LQTS) which is caused by mutations in genes which encode the cardiac ion channels[*].

Another genetic contender is a mutation in the medium-chain-acyl-CoA dehydrogenase (MCAD) gene which causes MCAD deficiency. This is a genetic disorder caused by the deficiency of an enzyme which breaks down fats to give us energy, and accounts for about one percent of SIDS cases.

“There is also a grab bag of other rare disorders, but we also need to look at the clues that it’s not SIDS. This includes looking at whether there is a clinical history of perhaps hypoglycaemia (low blood sugar) in the family, or whether the autopsy revealed any findings such as something structurally wrong with the heart muscle. It is devastating for families to lose more than one baby to SIDS, but it does happen, and it is rarely due to infanticide.”

Prof Christodoulou says if a genetic autopsy is performed, it should be accompanied by a careful family history (including a three generation family tree if possible), a past personal history and a detailed history of the 48 hours before death. He also notes that newborn screening cards are important for picking up a range of metabolic disorders known to affect newborns.

The incidence of SIDS has declined by 80% in Australia since a safe sleeping program[1] was introduced in 1990. This program educates parents, caregivers and health care providers about ways to reduce the risk for SIDS and other sleep-related causes of infant death under one year of age.

Despite the decline in deaths, answers are still needed when, after a thorough investigation including a complete autopsy, and review of the circumstances of death and the clinical history, there is still no definitive cause of death. There were 81 SIDS deaths in Australia in 2010, which is 81 reasons to keep investigating possible genetic causes, even if we’re not there yet.

[*] Ion channels are present in the membranes of all cells and basically conduct and control the flow of ions (charged particles). Cardiac ion channelopathies affect the electrical functioning of the heart without affecting its structure.

[1] SIDS and Kids - www.sidsandkids.org/safe-sleeping

« Back to Home Page

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index

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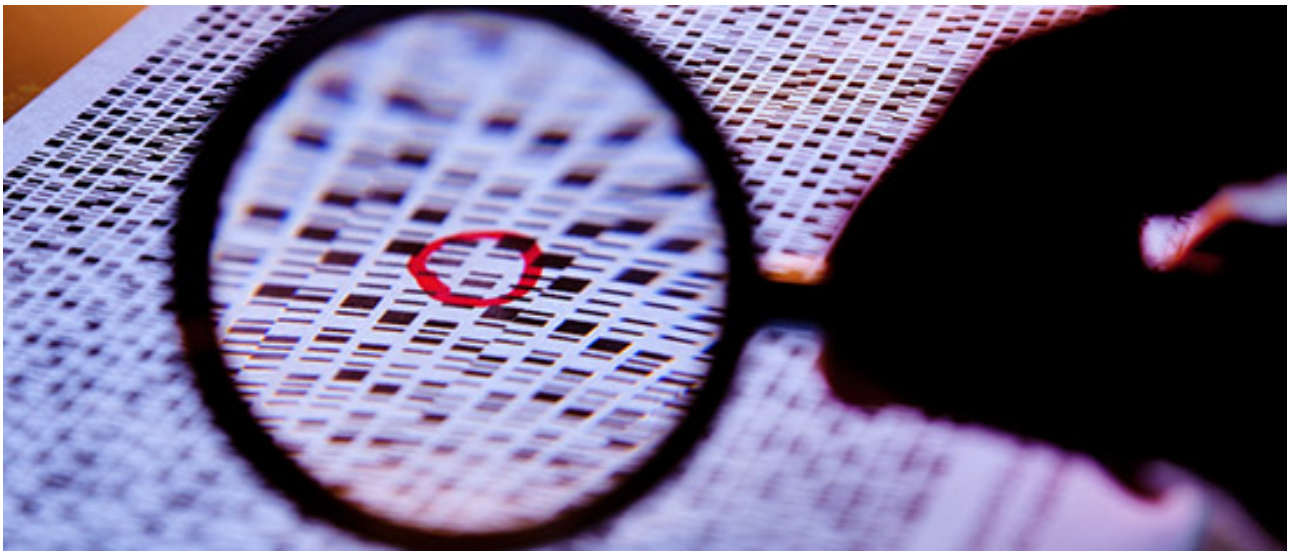
The genetic revolution offers hope to the 'orphans' of the health system



The paradox about rare or 'orphan' diseases is that, collectively, they are not uncommon. There are thousands of identified rare diseases affecting about 350 million people worldwide. This includes six to eight percent of all Australians, which equates to about 1.2million people. Yet diagnosis can be difficult, especially if there are perhaps just five other known cases of a particular disease in the world.

"Many parents or patients describe the 'diagnosis odyssey' which often involves telling their story over and over to many different clinicians," explains Dr Tracy Dudding, consultant clinical geneticist at Hunter Genetics. "All they want is a diagnosis yet around 60 percent of individuals with intellectual disability remain undiagnosed while others have a delayed or inaccurate diagnosis."

Rare diseases share a low prevalence of less than one in 2000. They are serious chronic diseases which are usually life threatening, and are often referred to as 'health orphans' because they are neglected with respect to research. About half of these diseases begin in childhood, and about half again involve neurological and intellectual disabilities.

Despite having different diseases, many patients face similar difficulties in their quest for a diagnosis, relevant information and proper direction towards qualified professionals. Dr Dudding says that while there may not be a cure, or even treatment available, a diagnosis is vitally important.

“Once people have a diagnosis they says things like ‘now I know that I didn’t do anything wrong’, ‘we can meet other parents’ and ‘now I have an answer when people ask what is wrong’. The importance of having a name for the disease cannot be underestimated.”

“One of my patients presented with severe intellectual handicap, seizures, brain malformations and incontinence,” she says. “He was also wheel chair bound and, because of overgrowth, weighed 80kg when he was 12 years old. His parents were in financial and emotional crisis because both of them had to give up work to care for their son.”

Families living with rare diseases are often in crisis, but there is some light on the horizon. New high output genetic technology may revolutionise the diagnostic process by outlining a genetic basis for these diseases, and potentially a therapy. Present estimates suggest that 80% of rare diseases may have an underlying genetic basis.

“The International Rare Diseases Research Consortium (IRDiRC) is also working towards 200 new therapies for rare diseases, and the means to diagnose most rare diseases, by the year 2020,” says Dr Dudding. “But help is also needed now.”

Dr Dudding is a founding board member of Rare Voices Australia[1] which is a not-for-profit organisation established in 2012 to provide a unified voice for all Australians living with a rare disease. The New Zealand Organisation for Rare Disorders (NZORD)[2] was set up in 2000, while orphanet[3] is the reference portal for information on rare diseases and orphan drugs for all audiences.

Without a diagnosis or treatment, patients living with a rare disease, or those caring for someone with a rare disease, can feel like the orphans of the health system. The pivotal role of genetics in the diagnosis, treatment and contribution to the understanding of rare diseases is yet to be realised, but it does give rise to a new era of hope for the millions of people affected by this collectively common plight.

[1] Rare Voices Australia – www.rarevoices.org.au

[2] New Zealand Organisation for Rare Disorders – www.nzord.org.nz

[3] orphanet - www.orpha.net

« Back to Home Page

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RCPA Manual
The Royal College of Pathologists of Australasia

The RCPA Manual aims to:

- Help you understand your clinical problems
- Help you understand pathology tests

Testing Cycle Information

- Requests and Collection
- Blood Collection
- Anatomical Pathology
- Unexpected Results
- Interpretation Guides
- Validity and Reliability
- Predictive Value

General Information

- How to use the Manual
- Publication details
- Glossary
- Acknowledgements
- Foreword
- Useful Links

Find a Clinical Problem

If you know the clinical problems and want to find the pathology test for it, you can find it by either searching or browsing.

Browse Clinical Problems or Enter Clinical Problem... Search

Find a Pathology Test by Name

If you know the name of the pathology test, you can search for it; otherwise, you can browse the pathology test listing.

Browse Pathology Tests or Enter Pathology Test... Search

Find a Pathology Decision Support Tool

If you know the name of the Pathology Decision Support Tool (PDST for short), you can search for it; otherwise, you can browse the Pathology Decision Support Tool listing.

Browse PDSTs or Enter PDST... Search

The RCPA wishes to acknowledge the Australian Government Department of Health and Ageing for their generous support of the RCPA Manual.
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Back to the Top

- RCPA
- RCPA Manual Home
- Terms and Conditions
- Privacy Statement
- Disclaimer

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All you need to know about pathology testing

Pathology tests are essential to modern medicine. They provide the information needed to make diagnoses, screen for illnesses and monitor treatment and medication. We're here to help you understand them with up-to-date information written by practising Australian pathologists and senior laboratory scientists.

Who we are: The website is managed by the Australasian Association of Clinical Biochemists (AACB) with support from the Royal College of Pathologists of Australasia (RCPA). It has been funded under the Quality Use of Pathology Program of the Commonwealth Department of Health and Ageing.

See Features and Services for guidance on how to use this site

Topics in the News

New Blood Test for Detecting Fetal Abnormalities Available in Australia

March 7, 2013

A non-invasive maternal blood test that can detect certain fetal chromosomal disorders, including Down syndrome, early in pregnancy is gaining attention as a potential new method of prenatal screening.

Human Genetics Society of Australasia Essay Competition for Australian and New Zealand High School Students.

March 6, 2013

2013 marks the 60th anniversary of the discovery of the double helix of DNA by James Watson and Francis Crick and the 10th anniversary of the first sequencing

of the human genome. To mark this occasion the HGSA has established a competition encouraging Australian and New Zealand high school students to submit an essay and compete for cash prizes and the honour of having the best essay published in the scientific journal Twin Research & Human Genetics.

New guideline for diagnosing diabetes in pregnancy

February 18, 2013

The Australasian Diabetes in Pregnancy Society (ADIPS) had previously set out guidelines for the testing and diagnosis of gestational diabetes mellitus (GDM) in 1991. However, as a result of the accumulation of more evidence about the incidence and effects of diabetes in pregnancy, ADIPS have revised the guidelines to reflect this new information.

Go

List of all tests and synonyms
Test not listed?
5-HIAA
17-Hydroxyprogesterone
ACE
ACTH
AFB Smear and Culture

List of all conditions/diseases
Acidosis and alkalosis
Addison's disease
Adrenal insufficiency
Alcoholism
Allergies
Alzheimer's disease

List of screening recommendations
Newborns
Infants
Children
Young Adults
Adults
Adults 50+

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Arbovirus Testing
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« Back to Latest Issue

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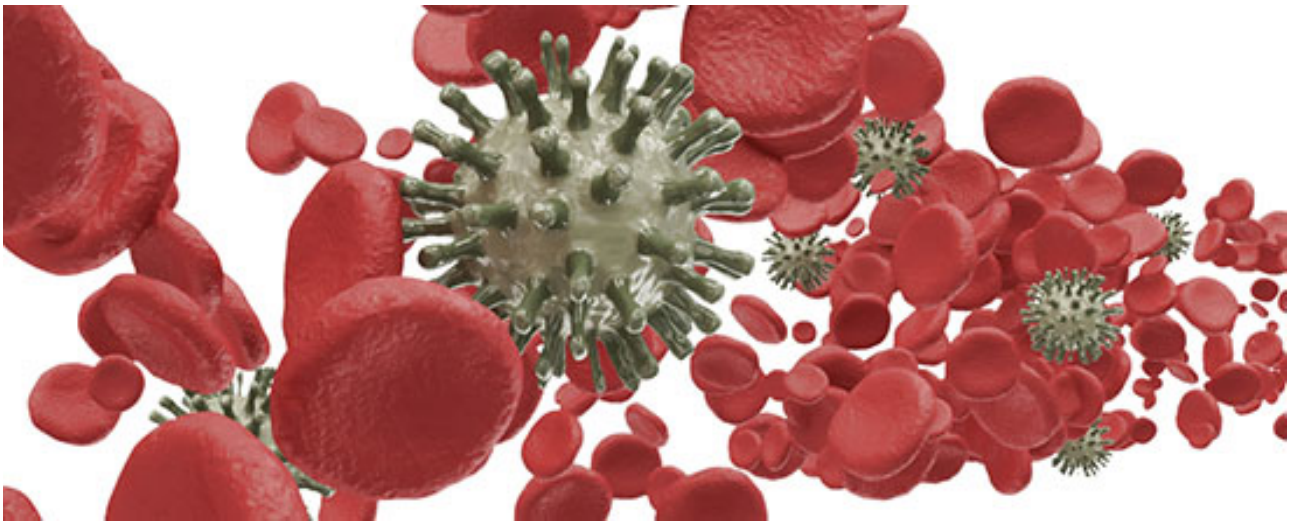
Rotavirus update highlights the importance of its discovery 40 years ago



Rotavirus is the most common infectious cause of severe gastroenteritis in infants and young children around the world. The causative organism was discovered in 1973 by a group of researchers led by Professor Ruth Bishop at the Royal Children's Hospital and University of Melbourne's Department of Microbiology. This eminent professor was on hand to listen to a presentation by Associate Professor Carl Kirkwood at last month's Pathology Update 2013 conference on where rotavirus stands today in the world of infectious diseases.

The name rotavirus comes from the characteristic wheel-like appearance (from the Latin rota meaning 'wheel') of the virus when viewed by electron microscopy. Infection is usually suspected based on symptoms, with diagnosis confirmed by testing a sample of the child's stools in a pathology laboratory. The virus kills about 500,000 children each year globally, although it is vaccine preventable.

A/Prof Kirkwood, who leads the Australian Rotavirus Surveillance Program and World Health Organization (WHO) Collaborating Centre, says there are two rotavirus vaccines commercially available which are administered orally, and both are highly effective in preventing severe rotavirus disease.

"Before the introduction of vaccines into the National Immunisation Program, rotavirus was responsible for around 120,000

doctor visits, 25,000 emergency department visits, 10,000 hospitalisations and three to five deaths each year in Australia," he says. "There has been a significant decrease in rotavirus associated gastroenteritis post vaccine introduction."

These decreases include at least a 71% decline in rotavirus-coded hospitalisations in infants as well as a 38% decline in non-rotavirus coded acute gastroenteritis hospitalisations. This indicates the disease burden from this virus was greater than previously thought.

"More than 100 countries have licensed one or both vaccines, and 42 countries have introduced the vaccine into their national program. It has been available in Australia since 2006, and was introduced into the National Immunisation Program in 2007."

Rotavirus vaccinations are available in New Zealand at a cost to the patient.

A/Prof Kirkwood says the introduction of rotavirus vaccines will increase population immunity, although, as with any virus, it has an innate ability to change.

"Before the vaccine there were five common types of rotavirus which caused more than 95 percent of the disease globally," he explains. "Post vaccine these types still occur, but there have been changes in the dominant strains every year which means changes in the genotype distribution."

Researchers are looking to see if the strains differ genetically from those described before the vaccine was introduced, with recent data from rotavirus outbreaks in the Northern Territory suggesting this is the case.

"Since the vaccine was introduced we have seen changes in the distribution of wild type strain population and a slight increase in rare and unusual types. Importantly, the vaccine strains are rarely associated with the disease."

The first step to developing a vaccine is identifying the causative organism, and in this regard Prof Bishop and her team contributed a huge legacy to child health worldwide. It was therefore fitting that she should be sitting with her peers listening to a presentation of where her work has led, in the same city where she made her discovery four decades ago.

« Back to Home Page

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Pathology training in Australasia leads to world class graduates

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"Pathologists first must obtain a medical degree which can take six to seven years, and then they must complete a minimum of one year of clinical practice before embarking on five years of training as a pathology registrar," explains Dr Debra Graves, CEO of the Royal College of Pathologists of Australasia (RCPA). "However, we prefer our trainees to have at least two years of clinical practice behind them."

The five years of specialist training to become a pathologist is an apprenticeship system in one of the 450 National Association of Testing Authorities (NATA) and RCPA accredited laboratories dotted around Australasia. Pathology specialties include anatomical pathology, chemical pathology, clinical pathology, forensic pathology, general pathology, genetic pathology, haematology, immunopathology, microbiology and oral and maxillofacial pathology, and each trainee dedicates their time to their chosen area.

Dr Joanna Glengarry is a recent graduate in anatomical pathology who now works in the Department of Forensic Pathology at Auckland Hospital doing further training to become a forensic pathologist. She attests to how tough the training regimen is.

“It takes a long time and the training is very intense. The RCPA really want to make sure we come out with world class qualifications so they really set the bar high,” she says. “There are a number of very tough exams that require up to a year of study and preparation, and even then not everyone passes.”

Dr Graves says the major exams occur in the first, third and fifth years of training, and the RCPA also participates in joint training with the Royal Australasian College of Physicians (RACP) in haematology, immunology, microbiology and infectious diseases, chemical pathology and endocrinology. There is also a joint program in genetics on the horizon.

Microbiologist and paediatric infectious diseases physician Dr Brendan McMullan says that joint training as a physician or paediatrician and a pathologist is a great way to appreciate two sides of the same problem.

“I look after children with infections and my pathology training helps me understand how to make the best use of the laboratory and tests to work out the diagnosis and treatment,” he says.

Dr Graves says that most people don’t realise that pathologists are involved in the diagnosis and monitoring of all acute and chronic illnesses and diagnose every detected cancer in the world, while Dr Glengarry sees her role as providing answers to not just other doctors, but to patients and their families.

“They come to us with a diagnostic problem and we have the ability to help them solve it. Our answer may determine the patient’s treatment or prognosis. Just this morning I took a call from a doctor who doesn’t know the cause of death of one his elderly patients and would like a postmortem examination to help him and the family understand what happened,” she explains.

“Forensic pathologists also work alongside the police and the coroner in evaluating the circumstances and causes of death or injury. It is a very diverse specialty and very interesting.”

Dr Glengarry says if there is one thing she would like people to know, it’s that pathologists are also doctors, a fact that some people don’t always realise. This means they can interpret their findings in terms of the whole patient.

And while the actors on CSI might solve their crimes in front of millions of viewers, most pathologists work behind the closed doors of their laboratories diagnosing, advising and monitoring millions of cases of disease and illness each year. To do this, they spend at least 13 years training and studying to practice medicine at its purest level.

« Back to Home Page

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