ePathWay
THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA

MAY 2018 | PUBLISHED BY RCPA ISSUE #080

IN THIS ISSUE

INTERESTING FACTS

1200

The number of people in New Zealand who die from bowel cancer each year.[1]

12

The approximate percentage of bowel cancer patients in New Zealand whose cancer has been diagnosed at an early stage.[2]

700,000

Welcome to the May 2018 edition of ePathWay

Pathology is the foundation for the clinical practice of medicine and paves the way to the appropriate diagnosis, management and treatment of diseases.

At some point in their life, every person relies on the work of pathologists. More often than not, though, patients don't know how closely involved a pathologist is in their healthcare and the diagnosis of their conditions.

In this month's issue of ePathway, the importance and scope of pathology is apparent as we discuss

- New Zealand's National Bowel Screening Programme;
- The Government's announced intention to focus on genomics and fund trials for genetic diseases
- A new diagnostic test for Polycystic Kidney Disease – another example of WGS (Whole Genome Sequencing) changing medicine; and
- Community acquired pneumonia (CAP) in Australia.

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).

The New Zealand National Bowel Screening Programme

The number of people in New Zealand who will be offered bowel screening every two years, once the national bowel screening programme is fully rolled out.[3]

Source:

[1] https://www.hqsc.govt.nz/our-programmes/health-quality-evaluation/projects/atlas-of-healthcare-variation/bowel-cancer/

[2] https://www.fmhs.auckland.ac.nz/assets/fmhs/sms/ctnz/docs/THE%20PIPER%20PROJECT%20Final%20deliverable%20report%207%20August%202015%20(HRC%2011_764%20FINDLAY).pdf

[3] https://www.beehive.govt.nz/release/national-bowel-screening-roll-out-next-step

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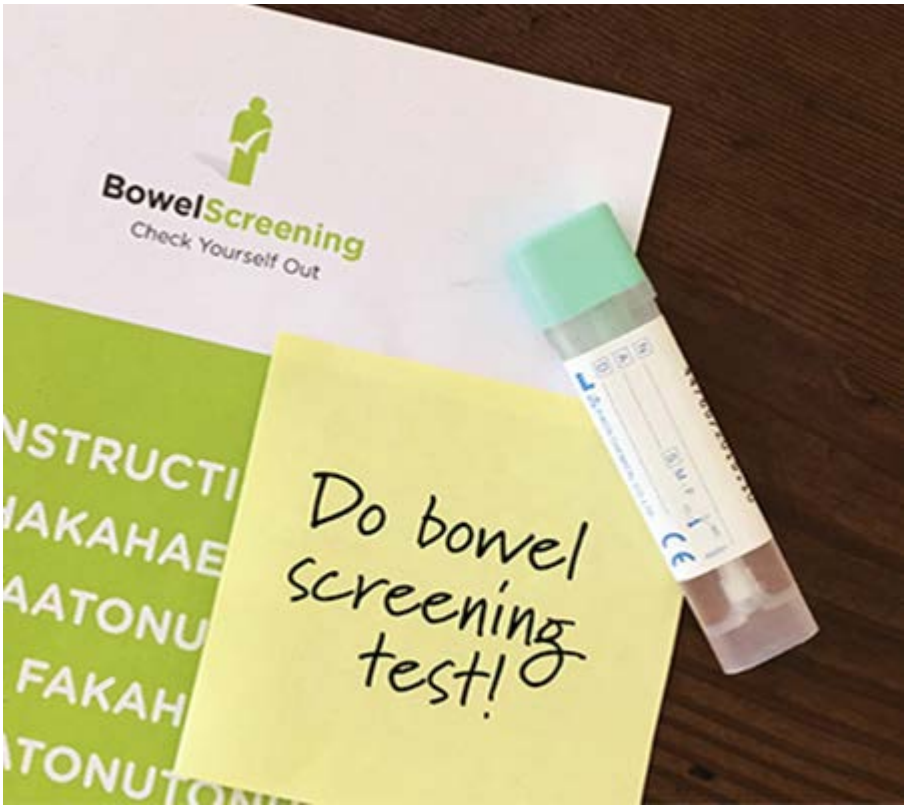

New Zealand has a high rate of bowel cancer[1], having the 16th highest incidence rate and the fourth highest mortality rate among OECD countries.

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New diagnostic test for PKD – another example of WGS changing medicine

Lab Tests Online

Know Pathology Know Healthcare

An advanced research test for polycystic kidney disease (PKD), developed at the Garvan Institute of Medical Research, has been translated through Genome.One into the world's first clinically accredited diagnostic whole genome sequencing (WGS) test for PKD.

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Dr Jenny Robson, Pathologist-in-Charge of Sullivan Nicolaides Pathology's Department of Microbiology and Molecular Pathology, discusses community-acquired pneumonia (CAP).

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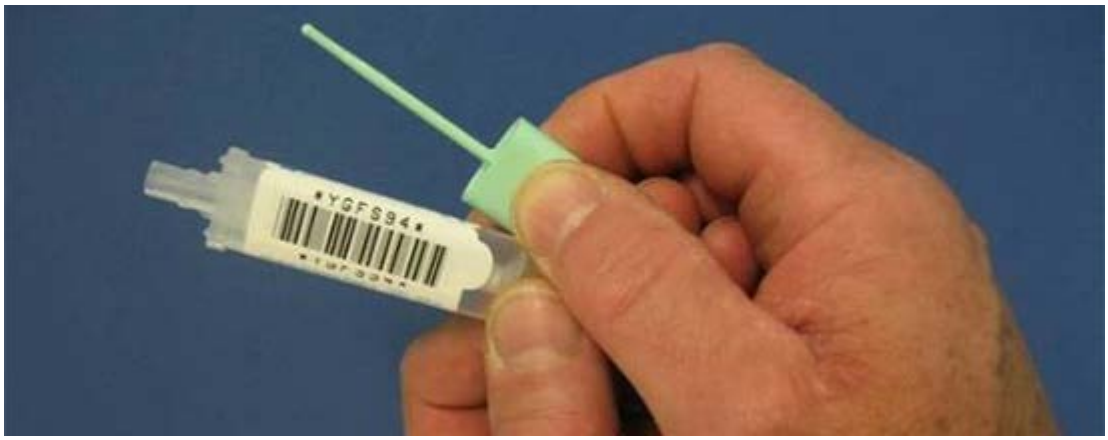
The New Zealand National Bowel Screening Programme



New Zealand has a high rate of bowel cancer [1], having the 16th highest incidence rate and the fourth highest mortality rate among OECD countries.

The National Bowel Screening Programme [2] is currently being rolled out across New Zealand, and will be free for men and women who are eligible for publicly funded health care and aged 60 to 74 years. There is evidence that population screening using regular faecal occult blood testing reduces mortality from bowel cancer, and many countries, such as the United Kingdom and Ireland, start with a restricted age range. In line with this practice, New Zealand is offering screening to those aged 60-74 years and, once the programme is fully rolled out, 700,000 people will be invited every two years. [3]

Dr Nicole Kramer, Anatomical Pathologist at LabPlus, Auckland City Hospital and The Royal College of Pathologists of Australasia's (RCPA) New Zealand Vice President, discusses the introduction of the National Bowel Screening Programme.

"Initially, a bowel screening pilot was implemented from 2012 until 2015 in one region within New Zealand. The pilot was subsequently extended for a further two years and in July last year, Hutt Valley and Wairarapa were the first District Health Boards (DHBs) to join the National Bowel Screening Programme. Following this, Waitemata (which was the area of the pilot) and Southern DHBs joined the programme. New Zealand is divided into 20 regions that are looked after by 20 DHBs that provide public health care in their region. The remaining DHBs will follow in stages with the next one, Counties Manukau DHB, due to commence in the coming months."

Those eligible for the national screening programme will be sent an initial pre-invitation

letter, followed two weeks later by a free bowel screening test kit, with instructions on how to complete the test in the privacy of their own home. Information on who is eligible for the publicly funded health care services is available on the Ministry of Health website.[4]

The completed test kit is then sent to a pathology laboratory. The test aims to detect traces of blood in the stool. If the test is positive, suggesting there is blood in the stool and therefore possibly a bowel problem, the participant is offered a colonoscopy. For the very small proportion of people for whom colonoscopy is not appropriate, a computed tomography colonography (CTC) or virtual colonoscopy is arranged. At colonoscopy, if an abnormality is seen, the physician or surgeon performing the colonoscopy takes a sample or removes a growth and then sends it to anatomical pathology for testing. We use microscopes to look at slides of the tissue to analyse it.

“In the diagnosis of bowel cancer, we are increasingly using molecular tests which provide oncologists and surgeons with better information about potential therapies and the prognosis. There are newer molecular tests for certain situations, coming on line all the time.

“Pathologists are the doctors who, in most situations, make the diagnosis of cancer on these small tissue samples that have been taken at colonoscopy. We also have a critical role in examining the tumour or cancer after surgery. We determine some of the features within the cancer that may define the prognosis and which will guide subsequent therapy for the patient. So the role that we have is pivotal in the diagnosis, prognosis and management of bowel cancer.

“The idea of the bowel screening programme is to capture asymptomatic bowel cancers in people aged between 60 and 74 years. We know that in New Zealand only a small proportion of people who have bowel cancer, approximately 12%, are diagnosed at an early stage[5]. The aim of screening is to detect cancer at an earlier stage, when people are not experiencing symptoms and when it can be more successfully treated.

“Although 80% of bowel cancer occurs in those aged 60 years and over, it is important to remember that anyone who is experiencing bowel symptoms that they are concerned about should speak to their General Practitioner as soon as possible. It’s important not to ignore symptoms.”

Each year, approximately 1,200 people in New Zealand will die from bowel cancer[6]. Therefore being aware of the symptoms that bowel cancer may cause is important. Symptoms include bleeding from the bottom (rectal bleeding) without any obvious reason, or a persistent change in bowel habits, such as going to the toilet more often or experiencing looser stools for several weeks.

“Being involved in bowel screening is really important because it’s one of the most common cancers in New Zealand. Screening provides an opportunity for people to have bowel cancer detected and treated, usually by surgery, at an earlier stage. That means additional treatments such as chemotherapy may not be required and it also means that your chance of survival may be higher.

“There has never been a national bowel screening programme in New Zealand before. Even though it is based on international screening programmes, we operate in a different health environment so there are always going to be ‘country unique’ challenges that arise during a pilot. Part of the reason for a pilot is to identify the most significant potential problems and address these early on before bowel screening is rolled out nationally.

“In New Zealand, we are trying to create a national register of all the potential participants in the bowel screening programme. We invite people using this register which is based on the National Health Index (NHI), a unique health identifier that all New Zealanders get when they enrol and come into contact with the public health system. For this reason, I would encourage everyone to make sure they are enrolled with their local GP so that they can be invited for screening when they become eligible and we can be sure we have their most up to date address.”

[1] https://www.oecd-ilibrary.org/social-issues-migration-health/health-at-a-glance-

2017/colorectal-cancer-mortality-2005-and-2015-or-nearest-years_health_glance-2017-graph103-en and https://www.oecd-ilibrary.org/social-issues-migration-health/health-at-a-glance-2017/survival-and-mortality-for-colorectal-cancer_health_glance-2017-41-en

[2] https://www.timetoscreen.nz/bowel-screening/

[3] https://www.beehive.govt.nz/release/national-bowel-screening-roll-out-next-steps

[4] https://www.health.govt.nz/new-zealand-health-system/eligibility-publicly-funded-health-services

[5] https://www.fmhs.auckland.ac.nz/assets/fmhs/sms/ctnz/docs/THE%20PIPER%20PROJECT%20Final%20deliverable%20report%207%20August%202015%20(HRC%2011_764%20FINDLAY).pdf

[6] https://www.hqsc.govt.nz/our-programmes/health-quality-evaluation/projects/atlas-of-healthcare-variation/bowel-cancer/

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The RCPA supports the Government's announced intention to focus on genomics and fund trials for genetic diseases

The Royal College of Pathologists of Australasia (RCPA) strongly supports the Government's greater focus on genomics in the Budget. This follows the RCPA's successful application [1] to the Medical Services Advisory Committee (MSAC) to introduce wider diagnostic testing in cystic fibrosis (CF).

Furthermore, the RCPA supports the establishment of the Australian Genomics Health Futures Mission and its aim to improve research into genetic disorders.

Dr Melody Caramins, Chair of the RCPA Genetics Advisory Committee, said,

"The Government's decision to allocate funding to both testing and research for genetic diseases will improve general access to genetic testing and, as a result, will offer greater family planning assistance. CF, specifically, is one of the most common genetic diseases with 1 in 25 people carrying the defective gene – and since carriers usually do not display symptoms, most are unaware that they are carriers. In Australia, approximately one in 2,500 babies are born with CF. That's one every four days [2]; therefore, offering the test provides the opportunity to make better informed reproductive choices."

The new test available in Australia will be used to determine the diagnosis of individuals suspected of having CF, as a prenatal test of the fetus, or as a preconception test for partners of CF carriers. This wider access to preconception CF testing will be available in order to determine the carrier status of couples and better information will be provided to assist in decision making. CF is a recessive disease; therefore, to have a child that

suffers from the disease both parents must be carriers.

The main manifestation of CF is lung disease; however, there are other manifestations, including disorders of the pancreas, liver, kidneys and intestine. The major cause of morbidity and mortality among young people with CF is progressive respiratory disease.

“The ability to test for genetic diseases such as CF enables individuals to make informed reproductive choices and certainly there are some potential treatments available which depend on knowing which mutations you have. Whilst treatments can ease symptoms, there is currently no cure. Major advances in medical research over the past several decades have made the condition much more manageable; however, reduced life expectancy is still a reality for the majority of people living with CF, with an average life expectancy of 38 in Australia.”

“The earlier that a decision is made regarding CF carrier status, the more options are available and, with this approval from MSAC, couples who have a child with CF, or who are at high risk because one or both parents is a carrier, will be provided with some improved access to testing via the MBS.”

[1] http://www.msac.gov.au/internet/msac/publishing.nsf/Content/1216-public

[2] http://www.cysticfibrosis.org.au/all/learn/

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New diagnostic test for PKD – another example of WGS changing medicine

An advanced research test for polycystic kidney disease (PKD), developed at the Garvan Institute of Medical Research, has been translated through Genome.One into the world's first clinically accredited diagnostic whole genome sequencing (WGS) test for PKD.

Dr Nicole Schonrock, Medical Science Liaison at Genome.One, discusses the exciting new test which is changing the diagnosis of PKD.

"Polycystic kidney disease (PKD) is an inherited condition in which many fluid filled sacs, also called cysts, form in the kidneys. It's a progressive disorder that gets worse over time as the cysts grow and kidney function declines, and unfortunately, it often causes kidney failure. Autosomal dominant PKD (ADPKD) is the most common form, which is mostly caused by mistakes in two genes called PKD1 and PKD2. Genetic testing for ADPKD is challenging as there are six other genes present in our DNA with an almost identical sequence to PKD1 making it hard to identify the real gene. Traditionally PKD is diagnosed using imaging techniques. A diagnostic genetic test for ADPKD using long-range PCR is available in the UK to sequence PKD1 but this technique cannot look at all types of genetic changes.

"The new PKD test, which is now available in Australia but is not currently available on the Medicare Benefits Schedule (MBS), allows us to diagnose PKD using whole genome sequencing. The patient's entire DNA is sequenced and a group of nine genes that are known to cause large kidney cysts in adult and paediatric patients are analysed. The new test looks at all types of genetic changes in these genes and the individual's entire

genetic information then becomes a lifelong resource to use and re-analyse as needed. For example, this information can provide insights into pharmacogenomics and the patient's response to certain medications, which is information that their doctors could use to guide prescriptions and avoid possible adverse side effects.

"My role in making this test available was as a Medical Science Liaison at Genome.One. I've been working with research, commercial and clinical teams to increase knowledge and awareness and drive translation of the research into a clinically accredited test that is now available to the renal genetics community.

"This test offers benefits to both the patients and their families. For patients, it can help confirm the diagnosis or discover the exact cause of disease if the clinical diagnosis is unclear. It allows clinicians to make more informed decisions and, in certain circumstances, could avoid further, more invasive tests such as kidney biopsies. It can also help an individual to know what to expect. For instance, certain genetic changes in PKD1 are often associated with a more severe and early onset form of the condition, compared with variants in PKD2. Although there is currently no cure for PKD, early detection and management may reduce some of the symptoms and help maintain kidney function. This might take the form of treating hypertension and proteinuria, or implementing lifestyle changes such as drinking more water. The individual's family may also benefit from genetic testing if the results lead to screening of at-risk family members. This can be useful to see if an individual could be a kidney donor. If you are a family member who wants to donate a kidney, clinicians need to be absolutely sure that you don't have the condition. If the individual is a young donor, that might not be possible to determine using imaging technologies but is possible using genomic information. Individuals with PKD and their relatives might use genetic testing results to assist with family planning."

"The test has now been available in the clinic for approximately 12 months. Since then, we have had nearly 80 families come through Genome.One and there were many more involved in the validation process to get the test set up. We are also working closely with PKD Australia, around creating awareness for patients so that they can be partners in the decision making process about their health. A major challenge is that the availability of tests often runs ahead of funding schemes and health providers' awareness."

"The test is accessible from genetics clinics. Kidgen, the renal flagship of the Australian Genomics Health Alliance, has been driving the establishment of fifteen renal genetics clinics across the country, and this number is growing. With nephrologists, clinical geneticists and genetic counsellors working together they understand adult and paediatric renal genetics exceptionally well and are very well placed to help families access this test."

"From my perspective, genetic testing should be another tool in a clinician's suite of tests to help diagnose, manage and effectively treat their patients. It complements current testing and provides additional insights not given by other technologies. Genomic testing has the ability to change medicine from a reactive to a proactive way of dealing with health. However, many challenges still remain in dealing with cost, accessibility and integration into routine medical practice. Genomic testing will impact the health system in a big way and ADPKD is just a beautiful example of a condition that can be diagnosed using this advanced technology."

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Community acquired pneumonia (CAP) in Australia

Dr Jenny Robson, Pathologist-in-Charge of Sullivan Nicolaides Pathology's Department of Microbiology and Molecular Pathology, discusses community-acquired pneumonia (CAP).

"CAP is one of the more common and potentially serious respiratory infections that arise in the community. Respiratory infections are often graded from pharyngitis, bronchitis, and bronchiolitis to pneumonia. CAP is due to an infection of the lower airways involving the lungs and its airspaces and is differentiated from other respiratory infections such as bronchitis, pharyngitis, and viral type illnesses that don't affect the lower airways.

"Symptoms of pneumonia include fever and chills along with shortness of breath, cough and occasionally sputum production. Chest pain can also develop if the surface of the lung is involved, due to pleurisy, which is caused by inflammation of the linings around the lungs.

"It may be mild or severe but tends to be more severe in older patients who may have other significant medical conditions. CAP is quite different from nosocomial or hospital acquired pneumonia (HAP) which develops whilst someone is in hospital. HAP develops in hospitalised patients who are receiving lots of other medical interventions, and the causes of CAP are different from HAP."

Pneumonia is most commonly caused by bacteria but a significant number of cases are due to viruses or other infectious agents. About 50% of the time it is difficult to say what caused the CAP. The bacterium that most commonly causes CAP is *Streptococcus pneumoniae*, also known as the pneumococcus. This bacterium can colonise a person's

airway without causing disease, but on occasions colonisation progresses to a pneumonia, which tends to occur most commonly during winter. Another reason for the winter predominance is that viral infections such as influenza are more common at this time and these can precede or even cause the pneumonia.

There are over 77,500 pneumonia hospitalisations in Australia each year, and the average stay rises with age – from six days for those under 65 to 13 days for those 65+ [1].

“CAP is diagnosed by putting together the symptoms and signs. For instance, there might be signs of fever, rapid respiratory rate and high pulse rate. Then when you auscultate the chest (listen, using a stethoscope) there are often signs in the chest that suggest there is consolidation of the alveoli or air sacs which become filled up with inflammatory cells and other secretions. Then it is confirmed on radiology either via an X-ray or a CAT scan.

“Certainly, pathology allows us to investigate the possible cause of CAP and this helps to determine how it should best be treated. For instance, if it is determined it’s caused by a virus it should not be treated with antibiotics. In this time of great concern for widespread antimicrobial resistance, it’s important not to use unnecessary broad-spectrum antibiotics. *Streptococcus pneumoniae*, the most common cause of CAP on the other hand, is a bacterium and does require antibiotics, so for mild CAP, amoxicillin, or doxycycline or clarithromycin is still effective without the need for broader spectrum antibiotics. What is used in treatment depends on the severity of the pneumonia and there are clinical indicators to tell whether it is mild, moderate or severe and if the patient requires hospitalisation and intravenous therapy.

“In the laboratory, one can perform sputum cultures which can be used to detect and diagnose bacterial lower respiratory tract infections. Sputum cultures have a low sensitivity but can be useful, particularly in those people who haven’t received prior antibiotics. If an individual has had antibiotics then often the flora in the sputum changes and we might isolate things that aren’t really the true cause of the pneumonia. *Streptococcus pneumoniae* can classically be found as little pairs of elongated Gram-positive cocci in the sputum that is collected and that can be a good indicator of CAP, but it’s difficult because *Streptococcus pneumoniae* is also carried in the throat and the respiratory tract. As in lots of infections in microbiology, differentiating between carriage and clinical disease is difficult and correlation of our microbiological results with clinical findings is so important.

“Polymerase chain reaction (PCR) is another technique used in the pathology laboratory. Here a single copy or a few copies of a segment of nucleic acid from the pathogen are amplified across several orders of magnitude, generating thousands to millions of copies of a particular DNA sequence. Nowadays, we can use PCR to detect a whole range of respiratory viruses – influenza A and B, respiratory syncytial virus (RSV), human metapneumovirus (hMPV), paramyxovirus type 1,2,3 and 4, adenovirus and rhinovirus. Many of these viruses not only cause upper respiratory tract infections but can cause pneumonia as well. The same PCR technology is now being applied to detect other less common causes of pneumonia including “atypical” bacteria that may be more difficult to culture. For instance, you can use PCR techniques to look for *Mycoplasma pneumoniae*, *Chlamydophila pneumoniae* and *Legionella* species.

“For individuals who are really unwell, and may require hospitalisation, two sets of blood cultures might also be taken. Again, it’s a bit like sputum in that blood cultures have a low sensitivity but if they are positive, this really helps us to define what the illness is caused by. If an individual has a very severe CAP that takes them to an emergency centre, an X-ray might be done to confirm the pneumonia; then, as well as the sputum, PCR’s and blood cultures we may well collect a urine test too. Here we are looking for two specific bacterial antigens, which when found in the urine in pneumonia can be very useful in confirming a diagnosis. These include the pneumococcus and the less common Legionella pneumophila that can cause quite a severe pneumonia.

“Overall, to protect yourself against CAP, the two most important things you can do from a medical point of view are to ensure you get the flu vaccine and, depending on age, the pneumococcal vaccine. Of all the respiratory viruses, influenza A and B are probably the

most important pathogens so it's important to protect yourself but also others by having the flu vaccine. In adults over 65 years of age, you can consider getting the pneumococcal vaccine without the need for further boosters. It is recommended that indigenous Australians get this vaccine earlier, from 50 years of age, and also receive a booster at 5 years. The other priority is to practice good respiratory hygiene. When you have respiratory symptoms, be sure to take measures to stop the spread of infection by using tissues, not handkerchiefs, and throwing them away after use, covering your mouth and washing your hands."

[1] https://lungfoundation.com.au/patient-support/other-lung-conditions/pneumonia/

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