ePathWay
THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA

APRIL 2016 | Published by RCPA

Issue #057

In This Issue

Interesting Facts

593

The number of Fellows of the RCPA who attended Pathology Update 2016.

250

The number of trainee pathologists who attended Pathology Update 2016.

Welcome to the April edition of ePathWay

This month's edition spotlights talks given at Pathology Update 2016, and while they cover different topics there is also a common thread – they all look towards the future. The articles cover:

- The next chapter for non-invasive prenatal testing.
- The search for a cure for two debilitating blood disorders.
- A possible new approach to diagnosing food allergies in children.
- Infection control in the 21st century.

Don't forget to check our posts on our Facebook page. You can also follow our CEO Dr Debra Graves (@DebraJGraves) or the College (@PathologyRCPA) on Twitter to keep up to date with pathology news.

The next chapter for NIPT shows it has scope outside of the prenatal setting

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The number of retired pathologists who attended Pathology Update 2016.

Source: RCPA

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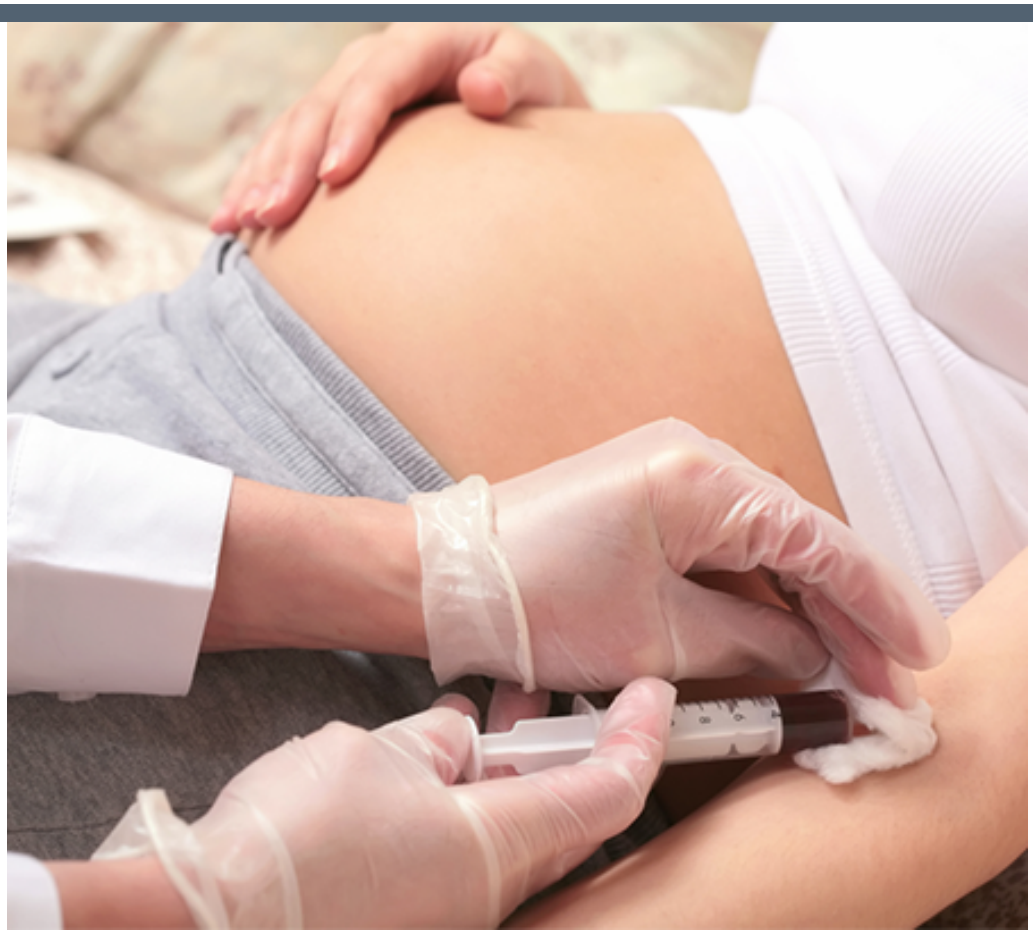

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The next chapter for NIPT shows it has scope outside of the prenatal setting

This article charts the next chapter for non-invasive prenatal testing (NIPT), but it won't be the last. Developed by Professors Dennis Lo and Rossa Chiu at The Chinese University of Hong Kong, NIPT is providing new insights into maternal and placental physiology or pathology, but its revelations are not limited to pregnancy. It's also showing us new aspects of diseases such as autoimmune disorders and cancer.

"We developed the current technology for prenatal non-invasive screening for Down syndrome in 2007. We conducted the first clinical trial in 2011, and by 2012 it was used in several countries including Australia. With the latest breakthrough of plasma DNA tissue mapping, NIPT now has the potential to first detect the presence of abnormal DNA in blood taken from pregnant women, and then identify if this abnormal DNA is from the baby or the mother."

NIPT performed on a maternal blood sample detects increased chromosome 21 DNA content which is a marker for an increased risk of delivering a baby with Down syndrome. Prof Chiu says while this was the original intent of NIPT, it is also uncovering incidental findings such as mosaicism, and diseases not limited to the prenatal setting.

"Cancer and some autoimmune diseases such as systemic lupus erythematosus are also associated with abnormal plasma DNA profiles. We have therefore had cases where DNA abnormalities detected by NIPT have revealed the presence of these diseases in the mothers."

Prof Chiu says her team is also working on other methods to identify sets of DNA variations, or polymorphisms, that tend to be inherited together, and investigating ways to non-invasively map the baby's genome before it is born.

"NIPT may also detect other single gene disorders apart from Down syndrome, but that's still being investigated as well. We are also looking for functional diseases of pregnancy and pregnancy associated disorders such as preeclampsia."

These wish lists mean this certainly isn't the last chapter for NIPT. The RCPA is also planning to submit an application for NIPT to be included in the Medicare Schedule, which is a logical step for a technology that has now changed the practice of prenatal screening around the world.

What is not clear is how much more of an impact NIPT will have on prenatal screening, and on detecting disease outside of the prenatal setting, because these chapters are still a work in progress. Watch this space!

Professor Rossa Chiu is a Fellow of the RCPA. She delivered a talk titled *Non-Invasive Prenatal Testing: What's Next* on Saturday 27 February at Pathology Update 2016 held at the Melbourne Convention Centre.

NIPT is covered in the July 2015 edition of ePathWay as well.

You can also read the RCPA's Position Statement on NIPT, and find more information about this test at Lab Tests Online.

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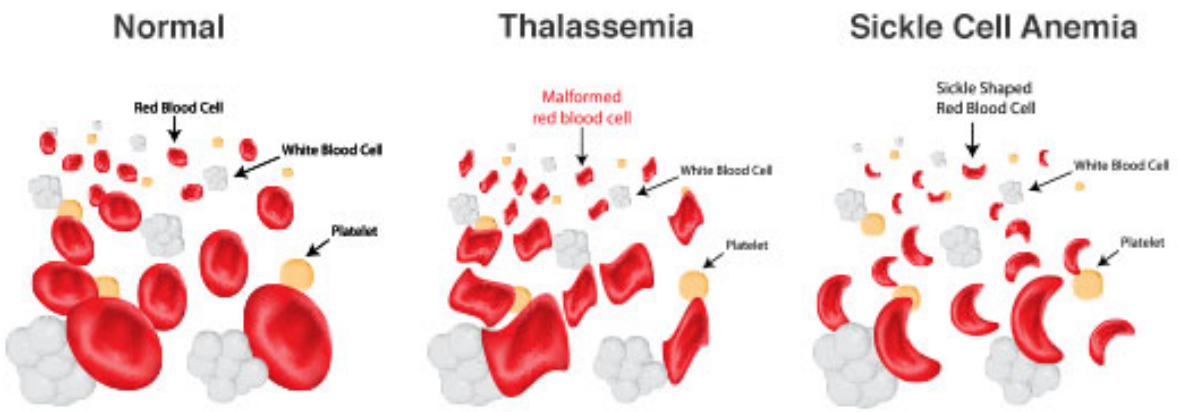

Albert Einstein challenged us all to never stop questioning, and that seems to be the ethos of Professor Merlin Crossley and his team. Their curiosity has them looking for a cure for sickle cell anaemia and thalassaemia, which are debilitating diseases with no known cure, using world-first techniques in gene therapy.

Sickle cell anaemia and thalassaemia are genetic disorders caused when a person inherits two defective adult haemoglobin genes. Haemoglobin is an ingredient in red blood cells that allows them to carry oxygen.

In sickle cell anaemia, abnormal haemoglobin causes red blood cells to lose their shape and flexibility, block capillaries and fail to carry oxygen properly. Similarly in thalassaemia, patient's red blood cells are abnormal and reduced in number. In extreme cases, patients who have these blood disorders need regular blood transfusions and experience frequent pain from blocked capillaries. But there is a glimmer of hope on the horizon.

"We have discovered that through genome editing, such as CRISPR [Clustered, Regularly Interspaced, Short Palindromic Repeats], we can mimic natural mutations that turn on the super haemoglobin gene, which is very exciting," explains Prof Crossley.

Prof Crossley says there are a few families throughout the world who have the super haemoglobin gene turned on, even into adulthood, and don't display major symptoms, even if they carry mutations for sickle cell anaemia and thalassaemia.

"This led researchers to question how the haemoglobin 'switch' works and to assess if these diseases could be reversed in the laboratory using genome therapy. For instance, we wondered if introducing mutations that switch the super haemoglobin gene back on would work in the lab, and it turns out the answer was yes!"

Prof Crossley says haematopoietic stem cells, which are blood-forming stem cells that could come from a bone sample from the patient, would be needed to turn this discovery into an effective treatment.

"If we could achieve a high frequency of editing in enough stem cells to enable repopulation of the patient's blood with genetically enhanced cells, and then transplant them back into the patient, then this should cure these diseases."

Professor Merlin Crossley is an Australian biochemist, former Dean of the Faculty of Science and now Deputy Vice-Chancellor Education at the University of New South Wales. He delivered a talk titled *Editing the genome to introduce beneficial mutations* on Saturday 27 February at Pathology Update 2016 held at the Melbourne Convention Centre.

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"There are enormous challenges facing infection control in the 21st century. Countries across the world are confronted by ageing populations, restricted healthcare resources, demands for modern medicine and increasing antimicrobial resistance," explains Dr Stephanie Dancer, Consultant Microbiologist from NHS Lanarkshire and Professor of Microbiology at Edinburgh Napier University, Scotland.

"Problem pathogens in the community are set to invade hospitals, and those created in hospitals are seeding into the community. And to top it all off, continued use of antimicrobial agents is generating and consolidating resistance to nearly all classes of antimicrobial drugs."

While antibiotics still save lives, Dr Dancer says infections remain the second most common cause of death worldwide. Healthcare-associated infections affect at least one in 10 patients admitted to hospital.

"All of this is occurring against the present backdrop of steadily-increasing antimicrobial resistance. Without international recognition and collaboration on this issue, successful interventions in one part of the world will ultimately be compromised by control deficits in another."

Dr Dancer says our global village is one factor contributing to this conundrum because of the ease by which pathogens traverse the globe courtesy of healthcare tourism, migrant workers, refugees, and business and holiday travellers.

"Our mobile global population are the perfect travel agents for any bug who hitches a ride, especially when every person is also equipped with dual mobile inoculators [i.e. our hands]."

Dr Dancer says the world needs to prepare for a world without effective antibiotics, and that will probably mean going back to the future.

“We have rubbish in the street, dirty cutlery in restaurants, we don’t wear coats in the rain, and we don’t seem to be as careful of avoiding infections as people in past eras because of the expectation that there is a ‘pill for every ill’. Except there isn’t anymore. The bottom line is that we need to reinvent hygiene.”

Dr Dancer says strategies to cope with infection control in the 21st century include focusing on prevention rather than control of infections, educating people about what effective hygiene is, antimicrobial stewardship, research, managerial and political engagement, and good access to routine diagnostic laboratories including introducing rapid diagnostic molecular methods into routine practice.

“There is still time to prepare for a world without effective antibiotics. Revisiting hygiene values of the past might not set pulses racing, but we must reverse any complacency over infection prevention and control in the 21st century to help protect us against untreatable infections.”

Dr Stephanie Dancer presented the talk *Infection control in the 21st century* on Friday 26 February and on Sunday 28 February at Pathology Update 2016 held at the Melbourne Convention Centre.

We also covered her talk *How can antibiotics make us sick and what to do about it* in the March edition of ePathWay.

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Diagnosing food allergies is challenging. But if we know the epigenetic status of some genes is altered in children who develop food allergies then the question is: could targeting these modifications help diagnose food allergies, and distinguish symptomatic and asymptomatic patients?

Before we reveal how this might work, we'd better explain epigenetics. Its literal meaning is *above or on top of genetics* and refers to external modifications to DNA that turn genes 'on' or 'off'. These modifications from factors such as age, environment and lifestyle don't change the DNA sequence, but instead effect how cells interpret the genes, and this might be the key to finding a new way to diagnose food allergies.

"We are commissioning data sets from 100 patients. This is a continuation of a pilot study that took place at the Royal Children's Hospital Melbourne to identify signatures that differentiate children who have active food allergy from those who are tolerant," explains Dr David Martino, researcher from the Murdoch Childrens Research Institute.

"That study, which used a DNA based diagnostic for predicting outcomes, was completed in a small number of children within the hospital for both egg and peanut allergies. Blood samples from the children were taken after an oral food challenge and their DNA analysed to look for methylation modifications because methylation modifies the function of the DNA. The outcomes were quite positive and we are now investigating if this approach could translate into routine clinical use."

Dr Martino says the cause of food allergies must be due to a combination of genes and the environment – epigenetics – because the rapid rise in their incidence can't be explained by genetics alone. He says about 20% of those with allergies are sensitised to common foods, but only 10% have any symptoms.

"Current diagnostic tests are excellent markers of sensitisation but poor predictors of what happens when the food is eaten. By continuing our research thanks to funding contributed by the DHB Foundation, we hope to analyse methylation sensitive genes, look at how they could predict clinical outcomes for children, and assess how useful they might be in diagnosing food

allergies."

Dr David Martino delivered a talk titled *Using methylation signatures to distinguish food allergy from tolerance* on Sunday 28 February at Pathology Update 2016 held at the Melbourne Convention Centre.

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