



ePathWay

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA

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Issue #090

In This Issue

- Call to end freeze on Medicare rebates for pathology testing
- Increasing testing for Familial Hypercholesterolemia (FH)
- Pathology, it's in the blood: Dr Julie Lokan and Dr Anand Murugasu
- Diagnosing and treating Haemophilia

Interesting Facts

20+

how many years the freeze on Medicare rebates for pathology testing has remained in place.

1,000+

the number of molecular genetic tests in routine diagnostic use today. Only 30 of these are covered on the MBS.

Welcome to the April issue of ePathWay

ePathway is an e-magazine designed for anyone interested in their health and wellbeing and the integral role pathology plays in the diagnosis, treatment and management of diseases.

This month's issue of ePathway looks at the following:

- Call to end freeze on Medicare rebates for pathology testing
- Increasing testing for Familial Hypercholesterolemia (FH)
- Pathology, it's in the blood: Dr Julie Lokan and Dr Anand Murugasu
- Diagnosing and treating Haemophilia

The Royal College of Pathologists of Australasia (RCPA) has called on the Government to end the freeze on Medicare rebates for pathology testing, which has been in place for over 20 years. This follows the announcement from the Government that the current indexation freeze on all GP services on the MBS will be lifted from 1 July 2019, along with various diagnostic imaging rebates from 1 July 2020. RCPA President, Associate Professor Bruce Latham expresses his concerns on this ongoing issue.

The RCPA has submitted an application to MSAC in relation to genetic testing for Familial Hypercholesterolemia (FH) which is currently being reviewed. We speak to Associate Professor David Sullivan to learn more about this common genetic disorder, currently affecting at least 65,000 people in Australia. He explains that increased genetic testing, as well as treating those affected by FH with routine treatments is a very cost effective strategy, almost to the point of saving money for the healthcare system.

In this month's Pathology, It's in the blood feature, we speak to Doctors Julie Lokan and Anand Murugasu about what it's like to be a married couple working in the same profession. Julia and Anand met during their training to be Anatomical Pathologists in August 2004 when Anand concocted a ruse to call and get in touch. They and are now married with three children and currently work as Anatomical Pathologists at Royal Melbourne Hospital and Austin Health.

To tie in with World Haemophilia Day which took place on 17 April, we spoke to Associate Professor Chris Barnes to discuss this potentially life-threatening, genetic bleeding disorder. A/Prof Barnes provides an interesting insight into diagnosing and treating haemophilia, including a look at the challenges involved which are being addressed through a number of new treatments.

2,800

The number of people diagnosed with haemophilia in Australia¹. Recent studies suggest it affects over 430 New Zealand residents².

\$1,500

the amount it currently costs a patient for a genetic test for Familial Hypercholesterolemia (FH)

Source:

[1] <https://www.haemophilia.org.au/about-bleeding-disorders/fast-facts>

[2] <http://www.haemophilia.org.nz/bleeding-disorders/haemophilia/>

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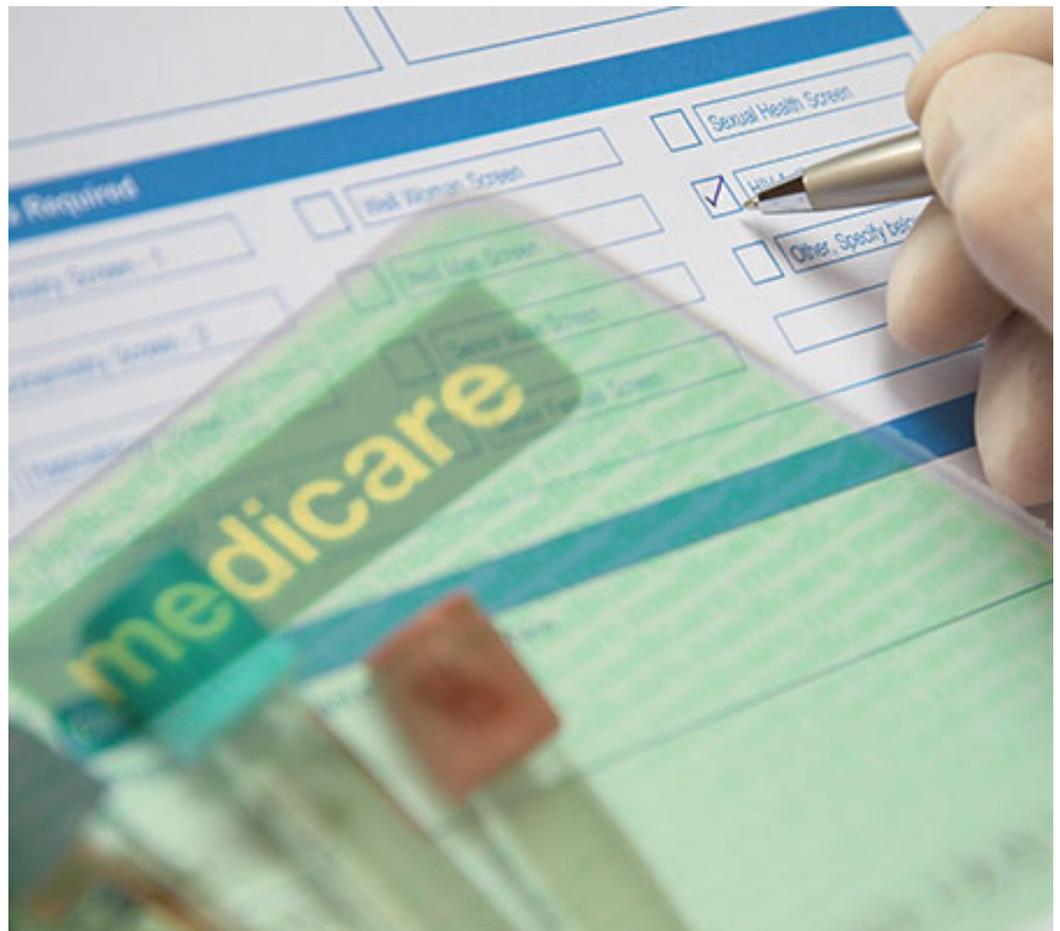
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Call to end freeze on Medicare rebates for pathology testing



[The Royal College of Pathologists of Australasia](#) (RCPA) has called on the Government to end the freeze on Medicare rebates for pathology testing, which has been in place for over 20 years. This follows the announcement from the Government that the current indexation freeze on all GP services on the MBS will be lifted from 1 July 2019, along with various diagnostic imaging rebates from 1 July 2020.

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Increasing Testing for Familial Hypercholesterolemia

Familial Hypercholesterolemia (FH) is a common genetic disorder in which the ability to remove low density lipoprotein (LDL) cholesterol from the blood is severely reduced. This results in high levels of LDL cholesterol, which can form plaques known as 'atheroma' on the arteries of the cardiovascular system, blocking the flow of blood and increasing the risk of heart attack and stroke. We spoke to Associate Professor David Sullivan to discuss the need for increased testing.



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Pathology, it's in the blood: Dr Julie Lokan and Dr Anand Murugasu

Doctors Julie Lokan and Anand Murugasu met during their training to be Anatomical Pathologists in August 2004. In 2005, Anand made the leap and moved to Melbourne where Julie was already living. They are now married with three children and live together in Melbourne where they work as anatomical pathologists, Anand at Royal Melbourne Hospital, and Julie at Austin Health.



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Diagnosing and treating Haemophilia

This year, World Haemophilia Day took place on 17 April, a worldwide initiative to increase awareness of haemophilia and other inherited bleeding disorders. We took the opportunity to speak with Associate Professor Chris Barnes, consultant haematologist at



the Royal Children's Hospital Precinct in Melbourne, to discuss how haemophilia is diagnosed and treated.



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IN THIS ISSUE

- Brain Awareness Week – Brain Cancer
- New nationwide research on genetic testing in Australia
- Seafood borne parasitic diseases - A single health approach is needed
- Diagnosing adrenal insufficiency

Welcome to the March issue of ePathWay

ePathway is an e-magazine designed for anyone interested in their health and wellbeing and the integral role pathology plays in the diagnosis, treatment and management of diseases.

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- Brain Awareness Week – Brain Cancer
- New nationwide research on genetic testing in Australia
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- Diagnosing adrenal insufficiency

Brain Awareness Week took place from 16-22 March. This is a global campaign to increase public awareness of the progress and benefits of

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Call to end freeze on Medicare rebates for pathology testing



[The Royal College of Pathologists of Australasia](#) (RCPA) has called on the Government to end the freeze on Medicare rebates for pathology testing, which has been in place for over 20 years. This follows the announcement from the Government that the current indexation freeze on all GP services on the MBS will be lifted from 1 July 2019, along with various diagnostic imaging rebates from 1 July 2020.

Associate Professor Bruce Latham, President of the RCPA, said,

“It is disappointing to see a continuation of the freeze on Medicare rebates for pathology testing. Pathology is central to many of the health initiatives announced in the Federal Budget 2019, including personalised treatment of cancers; diabetes prevention, care and research; tackling antimicrobial resistance; diagnosis of blood borne viruses and STIs; genomics; and biobanking.”

Beginning on 1 July 2019, the indexation of rebates for 119 general practitioner (GP) item numbers on the MBS will be reintroduced, whilst rebates on X-rays and ultrasounds will increase from mid-2020. The freeze on rebates for pathology testing remains in place.

“Whilst we welcome the intention to more adequately fund diagnostic imaging, oncology and surgery, it is important to note that none of these modalities actually diagnose cancer. One hundred percent of all cancer diagnoses are made by medical specialist pathologists and, despite the dramatically increasing complexity and expense involved in cancer diagnosis, MBS

rebates for all pathology tests remain frozen.”

“Personalised cancer treatment requires molecular genetic analysis of tumour cells. It is not true to say that genetic analysis is only in the future or that it is currently just a research issue. Over 1,000 molecular genetic tests are in routine diagnostic use today, but only about 30 of these items are covered on the MBS. These tests are absolutely vital in guiding treatment choices, however, due to their cost are not accessible to all who need them” said A/Prof Latham.

Recent nationwide research conducted by the RCPA for the Department of Health, showed that genetic and genomic testing is becoming increasingly integrated into healthcare, reflected by a 73% increase in test requests over the past 5½ years. More than 660,000 genetic/genomic tests were reported over the one-year survey period (April 2016 to March 2017). Of the 79, 000 tests requested for cancer the vast majority were funded by patients themselves or public hospitals.

“The more than 20-year rebate freeze has created two significant problems that threaten the future of pathology services for Australians. One is that newer, mainly genetic, tests are not funded by Medicare at all; and the other is that established tests are now critically underfunded. The high quality and timely pathology testing to which Australians are entitled is threatened to the point of near-extinction. The Government must intervene and end the rebate freeze,” said A/Prof Latham.

The RCPA continues to work with the Government to ensure that quality pathology services are accessible and affordable for all Australians.

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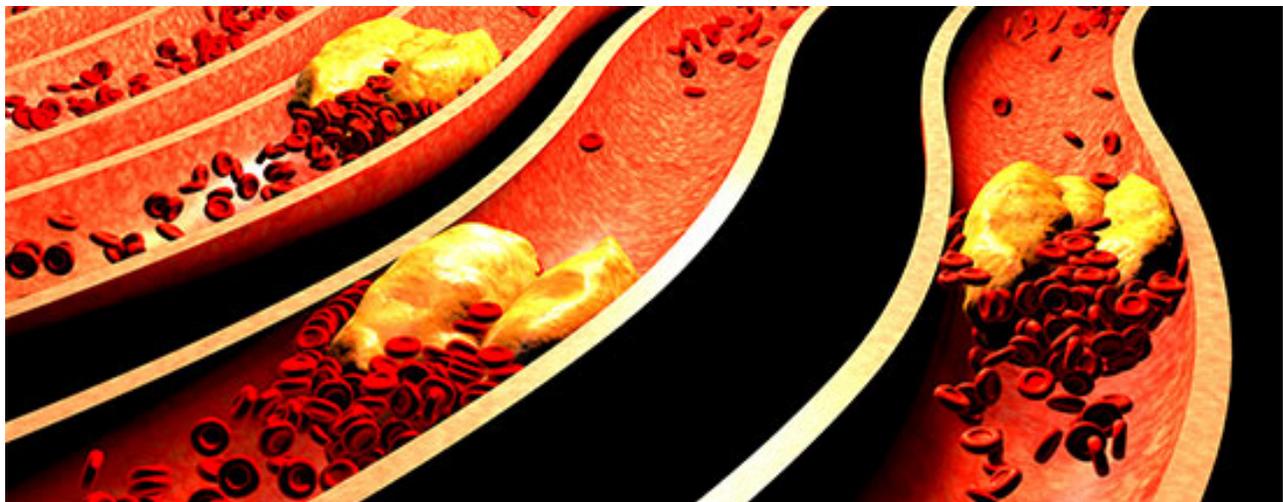
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Increasing Testing for Familial Hypercholesterolemia



Familial Hypercholesterolemia (FH) is a common genetic disorder in which the ability to remove low density lipoprotein (LDL) cholesterol from the blood is severely reduced. This results in high levels of LDL cholesterol, which can form plaques known as 'atheroma' on the arteries of the cardiovascular system, blocking the flow of blood and increasing the risk of heart attack and stroke. We spoke to Associate Professor David Sullivan to discuss the need for increased testing.

"FH is caused by major variations in the genes, particularly those affecting the LDL receptor. This includes the gene for the LDL receptor itself, the gene for the protein that links to the receptor, and also the gene responsible for the fine-tuning of the receptor, called PCSK9. There are also much rarer causes of FH.

"Just one faulty gene can result in problems with the body removing cholesterol from the blood, causing these levels to increase to up to twice what they should be. This can bring forward the risk of heart attack or stroke by about 20 to 40 years," said A/Prof Sullivan.

Although untreated (severe) FH can become clinically evident in adulthood, the disorder can be more difficult to identify at a younger age because cholesterol deposits in body tissues take time to develop. In Australia, it is estimated that at least 65,000 people have FH, with the vast majority of them being undiagnosed and/or inadequately treated.^[1] Many of those who do receive treatment have not been diagnosed with FH, so the familial implications may not have been recognised.

FH can be diagnosed with a genetic test which will assess whether or not a pathological change in the gene is present. In

those patients suspected of having inherited high blood cholesterol, cascade testing can be conducted on family members. Cascade screening is a mechanism for identifying people at risk for a genetic condition by a process of systematic family tracing.

The RCPA has submitted an application to MSAC in relation to genetic testing for FH, which is currently being reviewed. If approved, then it will be possible to refer patients with probable or definite FH for MBS-reimbursed genetic assessment. If a pathological genetic change is detected, cascade screening of close family members will be also be available.

“At the moment, for someone in whom the family genetic pattern is unknown, the cost of this test would be around AU\$1,500. Once we identify the change within a family, we can look for that change in other family members through cascade testing,” said A/Prof Sullivan.

In those family members tested, around 50% are likely to have FH as well. Detection early in life will allow those people to make lifestyle changes and also seek drug therapy to lower their blood cholesterol, thereby preventing or reducing the severity of cardiovascular disease. The other 50% of family members will be reassured that they don't have the inherited condition.

“The associated cost of the test has been very carefully examined. Diagnosing the condition with genetic testing, using that genetic result to test other family members, as well as treating those affected by FH with the routine treatments is a very cost effective strategy, almost to the point of saving money for the healthcare system.

“Once a patient is diagnosed, the condition is actually very treatable. The routine cholesterol lowering treatments, such as statins and intestinal cholesterol absorption inhibitors, all work very well. These are now supported by exciting new biological treatments which are very effective. The first family of these to be approved is called the PCSK9 inhibitors, such as evolocumab and alirocumab,” said A/Prof Sullivan.

References:

[1] Bellgard MI, Walker CE, Napier KR, et al. Design of the Familial Hypercholesterolaemia Australasia Network Registry: Creating Opportunities for Greater International Collaboration. *J Atheroscler Thromb*. 2017;24(10):1075-1084.

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Pathology, it's in the blood: Dr Julie Lokan and Dr Anand Murugasu



Doctors Julie Lokan and Anand Murugasu met during their training to be Anatomical Pathologists in August 2004. In 2005, Anand made the leap and moved to Melbourne where Julie was already living. They are now married with three children and live together in Melbourne where they work as anatomical pathologists, Anand at Royal Melbourne Hospital, and Julie at Austin Health.

Julie Lokan:

"I was interested in pathology as an undergraduate, it was my favourite subject because it seemed to me that the essence of clinical medicine was the study of disease. I guess I discovered an affinity for microscopy even back as far as second year histology classes. When some of my classmates were complaining about indecipherable 'pink and blue dots' I was seeing a wonderland of colours and patterns. I have a very visual brain, and used to notice small details in the world around me from an early age. In retrospect it is no surprise that I gravitated towards pathology. Nonetheless, I needed to experience clinical medicine for a while before the idea to do pathology took hold during my third year of medical residency. A position became available in microbiology, so I leapt right in!

"Microbiology was very interesting. For a while I contemplated doing microbiology as a speciality, but soon realised it was fast becoming the domain of infectious diseases physicians rather than pathologists. I then switched to Anatomical Pathology. I

joined the department at the hospital where I had done most of my medical resident rotations. They were lovely, welcoming, and inspiring teachers of the craft! I distinctly remember a calm and quite joyous feeling of knowing I had finally made the right career choice; it came to me when I was selecting tissue to examine under the microscope from an autopsy I had just performed, of all things! I really loved the challenge of tying all of the pieces of information from the medical history with what I had found at autopsy into a coherent story, which would hopefully provide answers to the treating team, and reassurance to the family. Detective work, problem solving, endless new learning, attention to detail, looking at pretty pictures down the microscope, it was everything I enjoyed!

“Anand and I met during our training, introduced through a mutual pathology friend of ours, who facilitated our interstate connection and encouraged Anand to make the leap of faith and move to Melbourne. We are both eternally grateful!

“Pathology husband and wife teams are not unheard of, but they are perhaps not common. It is really nice to have a life partner whom I can share my day with, and know he will really understand what I am describing. We also share cases with each other, and even ask each other’s advice about cases from time to time, as we have different areas of expertise. We work in different departments, and I suspect this is essential to maintaining harmony, and autonomous careers! Our personalities are quite different. Anand is very much an extrovert, and I am more introverted, though we are capable of meeting in the middle when required. Also, the way we practice pathology is slightly different, albeit equally valid, as we are in a different place on the “obsessiveness” spectrum!

“We have always found that our personality differences are complementary to each other - we fill in each other’s gaps, and this has made us a great team in life! In spite of our differences, our values and goals are essentially the same. We have found that having roughly the same career paths has provided us with a similar road to follow, which has worked very well for us and our family. We have a very equal relationship, an equality that extends both into our professional lives, our earning capacity, and our domestic responsibilities.

“We are both very passionate about the career we have chosen, and feel very lucky to have practiced morphological diagnosis in its “heyday”. We both enjoy teaching trainees and sharing our enthusiasm with them, hopefully to “pay forward” some of the enthusiastic teaching we received from our senior colleagues. Pathology is undergoing some changes as a specialty, with digital microscopy and molecular diagnostics evolving so rapidly, but we are looking forward to new challenges. No doubt we will be able to help each other evolve in the new diagnostic era.”

Dr Anand Murugasu

“My dad was a pathologist and my mum is a GP so I knew I wanted to do something medical, but pathology wasn’t ever something I thought hard about at Med School. I thought I would work in anaesthetics or in ICU but I tried that for a year and found I didn’t really enjoy it much. My Dad really enjoyed his job as a pathologist so I applied for a job and got on the program. I thought I would give it a year but after six months I found I really enjoyed it.

“The thing I love about Anatomical Pathology is that you can still be very general and have to see disease processes over lots of organ systems, and you get a real overview of it. Working in the hospital is really clinical. You’re involved with students and clinicians, and there is lots of research, so there are lots of opportunities to work across everything.

“I think pathology has something for everyone. There are certain traits that I think you do require to be a pathologist but Julie and I have very different personalities! There are a few husband and wife pathologists around but I wouldn’t say it is that common. Julie and I met through a friend who was a pathologist, but we met on a ski trip rather than through work. I saw Julie and thought she was pretty. She was a year ahead of me so I thought to ring her up and ask for exam advice, but really it was so I could ask her out.

“What I would say is that out of all the specialties, I think pathology and general practice are probably the easiest to be husband and wife because they are actually quite family friendly. You can both have pretty high-powered careers and still manage a family. At home we find we do talk about work a lot and use each other for advice – well, I use Julie!

“It is a really exciting time, not just in pathology but specifically Anatomical Pathology. In Oncology especially, there are big advances coming, which will be in understanding the molecular make-up of tumours. This will hopefully lead to some more targeted therapies, especially in breast cancer and prostate cancer, for example. The issue for these tumours, which sometimes get over-treated, is working out which of the tumours won’t do badly. So then you don’t over-treat them. The answer to this is through pathology.”

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Diagnosing and treating Haemophilia



This year, World Haemophilia Day took place on 17 April, a worldwide initiative to increase awareness of haemophilia and other inherited bleeding disorders. We took the opportunity to speak with Associate Professor Chris Barnes, consultant haematologist at the Royal Children's Hospital Precinct in Melbourne, to discuss how haemophilia is diagnosed and treated.

"Haemophilia is a potentially life-threatening, genetic bleeding disorder where the blood does not clot properly due to a lack of clotting factor (protein). It is caused by a mutation or change in one of the genes located on the X chromosome, meaning that the vast majority of those affected are males. Females can be a "carrier" of haemophilia if they have one affected X chromosome.

"Haemophilia occurs in 1 in 10,000 males. In Australia, there are currently more than 2,800 people diagnosed with varied degrees of severity^[1]. Recent studies suggest that over 430 New Zealand residents have haemophilia^[2]" said A/Prof Chris Barnes.

Symptoms of haemophilia vary depending on the severity of the deficiency of the protein; if the clotting-factor level is only mildly reduced, then bleeding may only occur after surgery or trauma. However, if the deficiency is severe then spontaneous bleeding may occur, including unexplained or excessive bleeding from cuts, pain and swelling in the joints, large or deep bruises, or blood in urine or in stools.

"In most cases, bleeding starts when young boys are moving around as infants and they develop significant bruising or painful

joints from bleeding. When left untreated, this internal bleeding can result in early onset arthritis. Bleeding in the brain is uncommon but might occur in young patients. Once diagnosed, bleeding can be effectively treated with the administration of clotting factor concentrate.”

“Managing the condition may need intravenous injections up to three times a week for some patients. The clotting factor treatment needs to go into the vein, directly into the circulation. We spend the majority of our time in the clinical space, assisting parents and the older boys in the infusion of the clotting factor” said A/Prof Barnes.

Until now, replacement therapy, which provides patients with the missing clotting-factor, has been the mainstay of treatment and is very effective in managing and preventing bleeding events. However, challenges still remain, including breakthrough bleeding, progressive joint disease, the development of inhibitors to the clotting factor. These challenges are being addressed through a number of new treatments.

“New treatments for haemophilia fall into three categories. Firstly, improvements to current clotting factors treatments mean patients do not have to infuse as frequently. Secondly, non-clotting factor treatments which are injected under the skin rather than infused - some of these medications have been trialled and are approved for funding. Thirdly, gene therapy.

“In broad terms, gene therapy uses viruses to provide a genetic treatment which allows patients to produce small amounts of the required clotting protein in the liver. This gene therapy is currently in late trial phase and we’re hopeful that this will ultimately result in a cure.

“Realistically, I’d say in the next three to five years gene therapy is likely to be a reality, but may not be available for all patients. We have to be careful that patients don’t think that one simple injection will cure the condition, it’s a bit more complicated than that. However we would be hopeful that gene therapy in some form, for a significant number of patients, will be available within a small number of years,” said A/Prof Barnes.

References:

[1] <https://www.haemophilia.org.au/about-bleeding-disorders/fast-facts>

[2] <http://www.haemophilia.org.nz/bleeding-disorders/haemophilia/>

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