ePathWay
THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA

FEBRUARY 2019 | PUBLISHED BY RCPA ISSUE #088

IN THIS ISSUE

- Rickets, a preventable skeletal disorder
- The RCPA's new position statement on pharmacogenetics
- Revisions to the RCPA's position on direct to consumer genetic testing
- Pathology Update 2019, The Power of Personalised Pathology

INTERESTING FACTS

11

The number of international speakers presenting at Pathology Update 2019.

1,370

The number of delegates in attendance at Pathology Update 2019.

$7,000

The total amount raised over the Pathology Update weekend for The Snowdome Foundation, in collaboration with Roche and the MCEC.

Welcome to the February issue of ePathway

ePathway is an e-magazine designed for anyone who is 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 will look at the following

- Rickets, a preventable skeletal disorder
- The RCPA's new position statement on pharmacogenetics
- Revisions to the RCPA's position on direct to consumer genetic testing
- Pathology Update 2019, The Power of Personalised Pathology

Rickets is a preventable skeletal disorder that affects infants and young children, causing soft and weakened bones. We spoke to Professor Tony Huynh to discuss the role of pathology in the diagnosis, management and treatment of the disease.

The RCPA has released a new position statement on the significance of genetic testing to predict a patient's response to drugs, also known as pharmacogenetics. We spoke to clinical geneticist and genetic pathologist, Professor Graeme Suthers to find out more.

Last year, ePathway announced that the College had expressed concerns over direct to consumer genetic testing without the involvement of an experienced medical practitioner. Since then, the College has released a revised position statement on genetic tests that are marketed directly to consumers. We spoke to Dr Melody Caramins, Chair of the RCPA's Genetics Council to explain more about these changes.

And finally, to Pathology Update 2019, The Power of Personalised Pathology which took place from 22 to 24th February 2019 at Melbourne Conference and Events Centre (MCEC). We provide an overview of the hugely successful three-day conference which is one of the largest gatherings of pathologists in the world.

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).

4.9/100,000/year

overall incidence vitamin D deficiency in children less than 16 years of age in Australia based on a study published in the Medical Journal of Australia in 2012

IMPORTANT MESSAGE

RCPA has an important message for you. Click to see the message!

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Know someone who might be interested in this website? Why not suggest the website to them.

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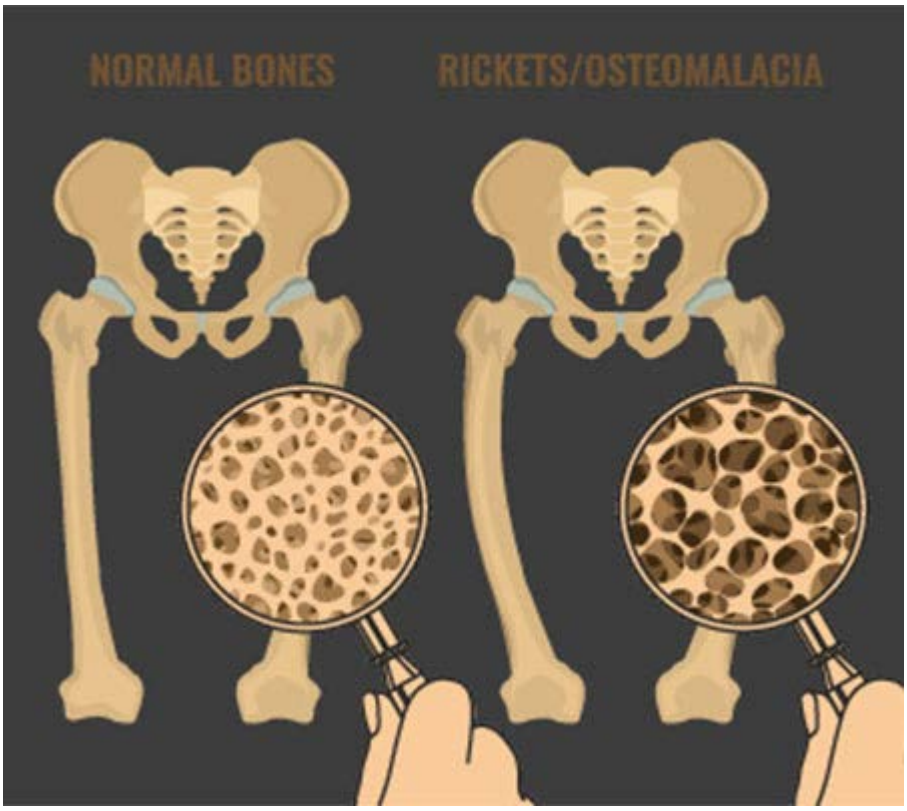
RCPA Manual

Lab Tests Online

Know Pathology Know Healthcare

Rickets – A preventable skeletal disorder



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The RCPA advocates for a collaborative approach for the successful introduction of pharmacogenetics in healthcare in Australia

The Royal College of Pathologists of Australasia (RCPA) has released a new position statement on the significance of genetic testing to predict a patient's response to drugs, also known as pharmacogenetics. The RCPA is advocating for a collaborative approach with medical colleges and the wider medical profession, the

Government, educators, and researchers, with the aim of creating national guidelines that will facilitate the safe and appropriate introduction of pharmacogenetics in Australia. By using pharmacogenetic testing, a clinician can align a prescription with the patient's potential for a beneficial or adverse response to a drug.

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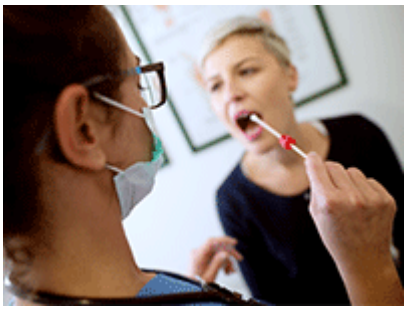
New RCPA position statement on direct-to-consumer genetic testing



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Pathology Update 2019, The Power of Personalised Pathology

Dr Lillian Siu

The Royal College of Pathologists of Australasia (RCPA) Pathology Update is the key pathology educational annual scientific meeting, and is one of the largest gatherings of pathologists in the world, now featuring ten concurrent sessions and 147 speakers including 11 international speakers. his year's conference, Pathology Update 2019, The Power of Personalised Pathology, took place at the Melbourne Conference and Events Centre, from 22-24th February 2019.

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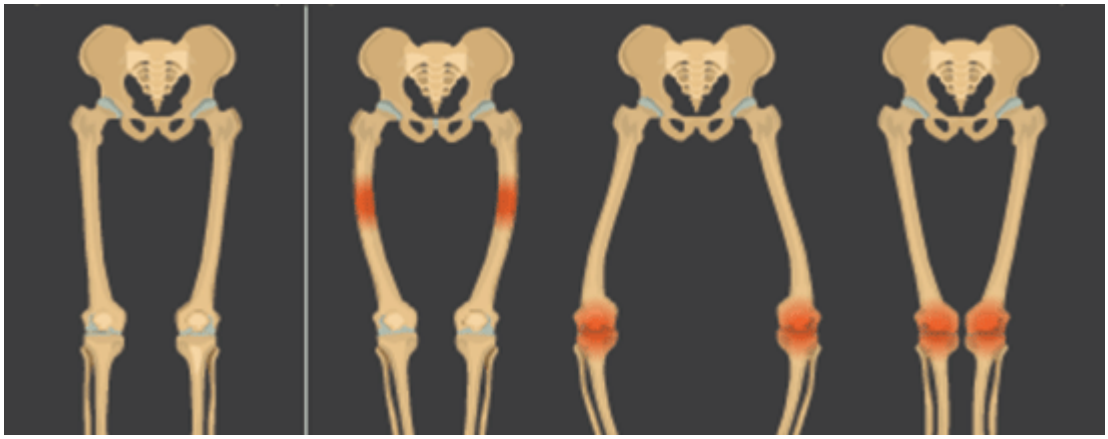
Rickets – A preventable skeletal disorder



Rickets is a preventable skeletal disorder that affects infants and young children, causing soft and weakened bones. We spoke to Dr Tony Huynh, a paediatric endocrinologist at the Queensland Children's Hospital and Chemical Pathologist at Mater Pathology in Brisbane, to find out more about this treatable disease and to discuss the role of pathology in the diagnosis, management and treatment.

"In paediatric terms, rickets is a disorder of defective chondrocyte differentiation and mineralization of the growth plate and defective osteoid mineralization. The adult equivalent is called osteomalacia, which is softening of the bones.

"There are genetic disorders of vitamin D and phosphate metabolism that cause rickets but these are relatively rare, and the vast majority of cases have a nutritional cause. Vitamin D deficiency and/or low calcium would be the predominant cause, which we refer to as nutritional rickets. An estimated incidence of vitamin D deficiency, from a 2012 paper published in the Medical Journal of Australia that defined a deficiency as 25(OH)-vitamin D levels <50 nanomoles per litre (nmol/L), was 4.9/100,000/year in Australian children less than 16 years of age. The majority of these cases had X-ray changes."

Most vitamin D is synthesised in the skin after exposure to ultraviolet B rays from direct sunlight, with dietary sources contributing little. Vitamin D deficiency and associated rickets are re-emerging as major public health issues worldwide, including in Australia. [1]

"The definition of vitamin D deficiency has been the subject of debate in recent years. A global consensus came out in 2016 on nutritional rickets. It defines "deficiency" as less than <30 nmol/L, "insufficiency" as 30-50 nmol/L and "sufficient" as >50 nmol/L in

children and adolescents. It is important that clinicians are aware that there are global consensus guidelines."

Rickets is most common in infants and young children. During this time, they usually experience rapid growth, meaning their bodies need high levels of calcium and phosphate in order for their bones to develop.

"Healthy children shouldn't be screened routinely because they are not at particular/much risk. The main risk groups are children in refugee populations/groups who may be dark-skinned, or people who are covered up, for example the Muslim population – that's probably the biggest group that we see with vitamin D deficiency rickets. The other risk group is infants that have been exclusively breast fed, because breast milk is low in vitamin D. Those with malabsorption form another risk group. Pancreatic insufficiency, for example, is an inability to absorb fat-soluble vitamins."

"Rickets can be asymptomatic. However, clinical symptoms include: bony features; swelling of the wrists and ankles; delayed fontanelle closing, which is normally closed by 2 years of age; and delayed tooth eruptions. The classic signs are leg deformities such as bowed legs, called genu valgum; and rachicitic rosary, which is widening of costochondral joints. You may also see frontal bossing, the term for a protruding forehead; and bone pain." "There are classical X-ray signs such as splaying or fraying of the metaphyses, widening of growth plates, and osteopenia". "There may be associated minimal trauma fractures". "Hypocalcaemia can also occur."

"Rickets is quite treatable. Obviously if it is really severe with hypocalcaemia then calcium needs to be replaced, sometimes intravenously. Rickets is really about getting enough vitamin D and calcium through daily safe sun exposure, and by eating foods that contain vitamin d and calcium such as fish, liver, milk, and eggs. In Australia we tend to use vitamin D3 or cholecalciferol as supplements, along with calcium supplements. There are minimum requirements at different ages, and the 2016 guideline goes through some of these. Even with significant deformities, such as leg bowing, once you correct the vitamin D deficiency and the calcium levels there tends to be a good recovery.

"I think the important public message is that if your child is healthy then there is a pretty low risk of rickets. From a breast-feeding perspective, we aren't saying don't exclusively breastfeed but there is the possibility that you may need to supplement your child with vitamin D during that time." Screening of at risk populations is appropriate.

To view the global consensus statement on rickets, please visit: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4880117/

[1] https://www.mja.com.au/journal/2012/196/7/incidence-vitamin-d-deficiency-rickets-among-australian-children-australian

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Following the new position statement, we spoke to one of Australia's most respected experts in the field of genetics, Professor Graeme Suthers,

"This is a rapidly growing and evolving discipline in healthcare and, when compared with other countries, Australia is under-represented in terms of pharmacogenetics education, guidelines, research, rebates, and regulatory guidance. Pharmacogenetics has potential application in many areas of healthcare. Patients metabolise medications differently, sometimes as a consequence of their genetic make-up, and this can create critical variation around whether a medication is beneficial. For patients in whom the metabolism of a drug differs from the average, the appropriate response to a pharmacogenetics test result can vary from modifying the dose of the drug to selecting a drug that is metabolised by a different genetic pathway."

"There is clear evidence that pharmacogenetics can assist in prescribing the right drug

at the right dose for a given patient. It is important to stress that it does not replace the professional responsibilities of a prescriber. Pharmacogenetics provides an additional source of information that informs the prescribing physician. A pharmacogenetic test is not required for every prescribing decision, and it is just one of the many factors to be considered by a prescribing doctor.

“There are only two pharmacogenetics tests on the MBS: these tests identify patients at risk of severe reactions to an HIV medication or a medication used in cancer and serious immunological conditions. The main source of funding for other pharmacogenetic tests is the patient, without there being support from Governments or insurers. Medical Colleges should seek MBS funding of those pharmacogenetics tests with sufficient evidence of validity and utility.

“Australia does not have a national vision about assessing and developing pharmacogenetics in healthcare. Many clinicians are unaware of recent advances in pharmacogenetics and their impact on clinical practice. Educational resources should be developed for students and practising clinicians, and we need national guidelines on how to use pharmacogenetic testing appropriately,” said Professor Suthers.

The College has identified a number of areas in relation to pharmacogenetics that should be addressed by the medical community, Government, educators and the public. Medications play a central role in the delivery of effective healthcare. However, medication can be costly and the benefits may be offset by side effects and risks of the medication. Pharmacogenetics provides a means of personalised prescribing, giving the right medication at the right dose to a patient. They deserve nothing less.

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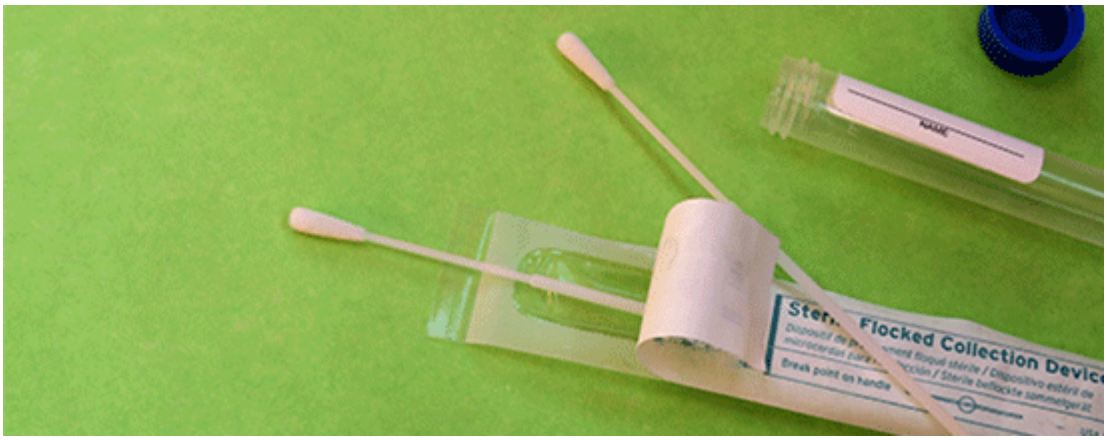
New RCPA position statement on direct-to-consumer genetic testing



The Royal College of Pathologists of Australasia (RCPA) has released a revised position statement on genetic tests that are marketed directly to consumers. The RCPA strongly advocates that complex medical tests should 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, however is particularly relevant for complex genetic tests that predict the medical future individuals, including children.

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

“We have had concerns over direct-to-consumer genetic testing, particularly where it was indicated that testing was taking place without the involvement of an experienced medical practitioner. This has prompted the RCPA to undertake a major revision of its position statement,

“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. 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. It is not appropriate for genetic tests that deal with significant clinical issues to be marketed directly to patients, or for professional support to be provided only after the testing has taken place.

“Many people are interested in using genetic tests, which can be utilised for medical and non-medical purposes, to determine ancestry, predict medication sensitivity, predict the

likelihood of developing particular diseases and of passing this predisposition to their children, and testing for acquired genetic changes in the oncology setting, which may help determine prognosis or treatment."

A table outlining broad categories for genetic testing can be viewed here. Using this table, categories one to five should be regarded as categories of 'Genetic testing for medical purposes', categories six to 10 regarded as 'Genetic testing for non-medical purposes' and category 11 as an example of a test for non-medical purposes that could never be regarded as a potential direct-to-consumer test.

"Importantly, it must be noted that labelling a medical test as 'For informational purposes only' (or similar wording) does not change its category of testing from that of a complex medical test, to a lesser non-medical category. It is the inherent nature of the genetic test that determines its categorisation, not the labelling that the provider places on it," said Dr Caramins.

For genetic tests that are used for any medical purpose, it is a legal requirement in Australia that most tests must be performed in a NATA/RCPA 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. Testing performed in an accredited pathology laboratory in Australia also ensures that the laboratory observes standards in relation to protecting patient privacy and confidentiality.

Many laboratories offering direct-to-consumer testing are not necessarily accredited to medical standards. Some laboratories are also based overseas and are not bound by Australian consumer protection laws. While this may not be relevant to the purpose of the tests being offered, it does mean that the laboratory might not be obliged to observe some of the customary safeguards medical laboratories adhere to in Australia.

"It is strongly recommended that the full 'Privacy Policy' (however named) and 'Terms and Conditions' of a direct-to-consumer laboratory service be carefully read and understood before providing any sample for testing. In particular, some of these laboratories reserve the right to release, forward or even sell samples or genetic information to external organisations. Once genetic information has been released to external parties, it is not usually possible to reverse or recover this information and it may have privacy consequences for the individual and their relatives," said Dr Caramins.

The statement also recommends that, for non-medical testing, scientific claims are reviewed for plausibility. While some testing laboratories offering non-medical genetic services use techniques based on sound scientific principles, others offering testing to predict physical beauty, athleticism, intelligence or romantic compatibility should have their scientific validity carefully considered before accepting their claims at face value. Consumer protection and truth-in-advertising laws may not necessarily apply for direct-to-consumer services from overseas through sources such as the internet.

This article appeared in the February 2019 Edition of ePathWay which is an online magazine produced by the Royal College of Pathologists of Australasia (http://www.rcpa.edu.au/Library/Publications/ePathway).

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Firstly, we would like to thank all of the international and local speakers who joined the conference this year, and the 1,370 delegates who travelled from far and wide to attend. Special thanks, of course goes to the dedicated team who made it all happen.

A particular highlight of the conference were the many international speakers who travelled across the globe to be with us, including those from New Zealand, the US, and throughout Europe. We were truly honoured to be joined by Dr Elaine Jaffe, who presented in the Anatomical, Haematology and Immunopathology combined scientific stream. Dr Jaffe presented the following topics at the conference: 'The revised WHO classification of lymphomas – what has changed for the pathologist and clinician'; 'Lymphoproliferative disorders (LPD) associated with immune deficiency'; 'Hodgkin's lymphomas – historical background and evolution'; and 'Peripheral T-cell neoplasms – current classification and differential diagnosis'.

Each year, the College invites one key speaker to deliver the Eva Raik plenary, named after the first female President of the College. This year's plenary was titled, "Precision Cancer Medicine – Cup half full or cup half empty?" and was presented by Dr Lillian Siu

who joined the conference all the way from Toronto. Dr Siu also presented in the Genetic Pathology scientific stream with presentations titled: ‘Molecular tumour board – interesting cases from a cancer genomics program’; and Building national and international genomic data sharing initiatives’.

In addition to the fascinating scientific sessions, the conference provided the perfect opportunity to meet with colleagues and share stories from mutual areas of interest. All delegates were invited to join the Welcome Cocktail Party on the Friday evening and, on Saturday evening, new College Fellows and Award Winners were celebrated.

This year the following fellows, and many more, were presented with awards:

Honorary Fellowship
Dr Elaine Jaffe

Distinguished Fellow Award
A/Prof David Ellis (SA)
Dr Colin Goldschmidt (NSW)

Meritorious Service Award
A/Prof Christine Hemmings (NZ)

Pathology Update 2019, in collaboration with major partner Roche and the Melbourne Convention and Exhibition Centre, was proud to support The Snowdome Foundation (https://www.snowdome.org.au) which was recognised as the Pathology Update 2019 Charity of Choice, The Foundation was chosen due to its synergy with the conference’s Scientific Program.

Snowdome has a deliberate mission to improve outcomes for Australian men, women and children with blood cancers. Its aim is to 'unlock new treatments' by channelling government and private philanthropic investment into early phase human clinical trials of next-generation drugs and therapies. Together with Roche and the Melbourne Convention and Exhibition Centre (MCEC), the RCPA raised a total of $7,000 over the Pathology Update 2019 weekend.

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