MEDIA RELEASE **11 MAY 2020**

Genetic testing for familial hypercholesterolemia listed on the Medicare Benefits Schedule

The Royal College of Pathologists of Australasia (RCPA) is pleased to announce that genetic testing for heritable mutations associated with Familial Hypercholesterolemia (FH) is now listed on the Medicare Benefits Schedule (MBS). This addition to the MBS follows an application by the RCPA to the Medical Services Advisory Committee (MSAC).

As a result of this change, in certain clinical situations patients with probable or definite FH will be referred for MBS-reimbursed genetic assessment. If a pathological genetic change is detected, cascade screening of close family members will also be available.

Spokesperson for the RCPA, NSW Health Pathology's Associate Professor David Sullivan, Head of Chemical Pathology, Royal Prince Alfred Hospital said,

"The RCPA is delighted that this application has been successful. Diagnostic testing for FH is a complicated and expensive test which analyses over 1,000 possible changes, to identify the gene pattern causing high cholesterol in the family. Once a causative change has been identified, other family members can be offered predictive testing, which can be done with much more certainty. At-risk individuals who are identified through genetic testing, and treated early, will likely decrease their risk of a cardiovascular event. Currently, for someone in whom the family genetic pattern is unknown, the cost of this test would be around AU$1,200.

"FH severely reduces the body's ability to remove low density lipoprotein (LDL) cholesterol from the blood, resulting in high levels of LDL cholesterol, which can form plaques known as 'atheroma' on the arteries of the cardiovascular system. The listing of these items enables earlier identifications of at-risk individuals with the aim of decreasing the risk of cardiovascular events, including myocardial infarction, unstable angina requiring hospitalisation, coronary revascularisation, stroke, transient ischaemic attack and hospitalisation for heart failure," 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 is a common genetic disorder and, in those whose family members are tested, around 50% are likely to also have the condition. Detection early in life allows those individuals to make lifestyle changes and also seek drug therapy in order 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.

"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 treatments to be approved is called the PCSK9 inhibitors, such as evolocumab and alirocumab.

[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.

"The associated cost of this genetic 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.

"The RCPA strongly supports this new MBS change and we expect to identify an increased number of individuals affected by these heritable gene mutations, in order to manage their treatments effectively, advise on preventative measures and ultimately save more lives," said A/Prof Sullivan.

For further information on the RCPA, please visit www.rcpa.edu.au or see updates on Facebook - @PathologyRCPA, Twitter - @RCPAPresident, @PathologyRCPA, or Instagram - @the_rcpa #RCPA #pathology #MedicineIsPathology.

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About the Royal College of Pathologists of Australasia:

The RCPA is the leading professional organisation representing pathologists, medical specialists and scientists who provide pathology testing in Australasia. Its mission is to train and support pathologists and to improve the use of pathology testing to achieve better healthcare.

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