

16 August 2017

Prof Gerald Watts  
Chair  
FH Australasia Network  
C/O PO Box 5436  
Chittaway Bay NSW 2661

Dear Gerald

**Re: MSAC Application**

The Royal College of Pathologists of Australasia (the College) is in the process of submitting an application to MSAC for a new MBS item number for Familial Hypercholesterolaemia (FH) genetic testing.

Because FH is an autosomal dominant disorder that results in increased levels of blood low density lipoprotein cholesterol (LDL-C), with LDL-C deposits in tissues and blood vessels that result in an increased risk of premature cardiovascular disease and death, genetic testing is a valuable pathology test in patient care. Although the severe phenotype is obvious clinically, milder manifestations are more difficult to identify.

Testing to identify individuals with a genetic cause for elevated blood cholesterol will enable early detection of those relatives who have inherited the same genetic cause for high cholesterol and are at high-risk of cardiovascular events. Identification of these individuals will allow commencement of lifestyle changes and drug therapy to lower blood cholesterol as early in life as possible, in order to prevent cardiovascular disease in a highly cost-effective manner and save patient lives.

The application proposes a new MBS item for the characterisation of germline gene variants (*LDLR*, *APOB* and *PCSK9*) in patients with hypercholesterolaemia where clinical and family history criteria place the patient at >10% risk of having a clinically actionable pathogenic mutation identified. The application also proposes an MBS item for cascade (predictive) testing of relatives identified with the FH pathogenic mutation.

As we recognise your important role in the care of FH patients and their families, we would ask for your support with this application. A letter from your organisation (via return email and addressed to me) affirming the clinical relevance of FH genetic testing and its importance in patient care would be valuable in achieving a successful outcome through the MSAC process.

Yours sincerely



Dr Michael Harrison  
**President**