

MEDIA RELEASE

10 MAY 2018

The RCPA supports the Government's announcement to focus on genomics and fund trials for genetic diseases

[The Royal College of Pathologists of Australasia](#) (RCPA) strongly supports the Government's greater focus on genomics in the Budget. This follows the RCPA's successful [application](#)¹ to the Medical Services Advisory Committee (MSAC) to introduce wider diagnostic testing in cystic fibrosis (CF).

Furthermore, the RCPA supports the establishment of the Australian Genomics Health Futures Mission and its aim to improve research for genetic disorders.

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

"The Government's decision to allocate funding to both testing and research for genetic diseases will improve general access to genetic testing and, as a result, will offer greater family planning assistance. CF, specifically, is one of the most common genetic diseases with 1 in 25 people carrying the defective gene – and since carriers usually do not display symptoms, most are unaware that they are carriers. In Australia, approximately one in 2,500 babies are born with CF, that's one every four days², therefore, offering the test provides the opportunity to make better informed reproductive choices."

The new test available in Australia will be used to determine the diagnosis of individuals suspected of having CF, as a prenatal test of the fetus, or as a preconception test for partners of CF carriers. This wider access to preconception CF testing will be available in order to determine the carrier status of couples and better information will be provided to assist in decision making. CF is a recessive disease, therefore to have a child that suffers from the disease both parents must be a carrier.

The main manifestation of CF is lung disease, however there are other manifestations, including the pancreas, liver, kidneys and intestine. The major cause of morbidity and mortality among young people with CF is progressive respiratory disease.

"The ability to test for genetic diseases such as CF enables individuals to make informed reproductive choices and certainly there are some potential treatments available which depend on knowing which mutations you have. Whilst treatments can ease symptoms, there is currently no cure. Major advances in medical research over the past several decades have made the condition much more manageable, however reduced life expectancy is still a reality for the majority of people living with CF, with an average life expectancy of 38 in Australia."

"The earlier that a decision is made regarding CF carrier status, the more options are available and, with this approval from MSAC, couples who have a child with CF, or who are at high risk because one or both parents is a carrier, will be provided with some improved access to testing via the MBS."

For further information on the RCPA, visit www.rcpa.edu.au. Or see our updates on [Facebook](#) or [Twitter](#) - [@PathologyRCPA](#) Instagram - [@the_rcpa](#).

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References:

¹ <http://www.msac.gov.au/internet/msac/publishing.nsf/Content/1216-public>

² <http://www.cysticfibrosis.org.au/all/learn/>

About the Royal College of Pathologists of Australasia:

The RCPA is the leading organisation representing pathologists 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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