

A finding that could help Alpha-1 sufferers breathe more easily

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Scientists have identified a new mutation in the gene that causes the inherited disease known as Alpha-1 Antitrypsin Deficiency (Alpha-1), which affects roughly one in 2,500 people of European descent.

Alpha-1 can lead to serious lung disease in adults, or [liver](#) disease at any age.

The finding extends understanding of Alpha-1 at the molecular level, potentially leading to new drug development and better diagnostic tools.

Dr Darren Saunders from Sydney's Garvan Institute of Medical Research and Professor Vanessa Hayes, from the J. Craig Venter Institute in San Diego, published their results in the international journal *PLOS One*, now online.

In healthy people, Alpha-1 Antitrypsin is a molecule, or 'protein', made in the liver then secreted into the blood. It helps keep the lungs healthy by blocking an enzyme that causes tissue breakdown.

The [genetic mutations](#) that trigger Alpha-1 disease cause the Alpha-1 Antitrypsin protein to accumulate in the liver, rather than being secreted into the blood. That in turn leads to tissue breakdown in the lungs.

[Alpha-1](#) is often first diagnosed as asthma or smoking-related [Chronic Obstructive Pulmonary Disease](#) (COPD). Typically, people experience symptoms like shortness of breath, wheezing, recurrent chest colds and

allergies. They might also develop unexplained [liver disease](#).

"We're interested in the fact that the mutation we've identified occurs in a completely different part of the gene from known mutations, yet it causes the same basic effect – the protein doesn't get secreted from [liver cells](#)," said Dr Saunders.

The research is still early stage, as the mutation has been identified in only one patient, and that patient is from a Middle-Eastern background.

"We have yet to establish the mutation's prevalence, and we can't do that until we have the funding to examine a wider population of patients."

"At the basic science level, it's a promising breakthrough because it helps us better understand how the protein misbehaves when it's mutated – and that's very important for drug development, because drugs have to try and minimise, or reverse, molecular damage."

"If the mutation turns out to be prevalent, then its inclusion in diagnostic tests will be important."

Provided by Research Australia

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