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Study shows treatment for genetically caused emphysema is effective

May 28 2015

A landmark clinical study in the *Lancet* provides convincing evidence that a frequently overlooked therapy for genetically-caused emphysema is effective and slows the progression of lung disease.

Alpha-1 antitrypsin deficiency is an inherited disorder that can cause emphysema even without exposure to tobacco smoke. Alpha-1 antitrypsin (AAT) is a protein made in the liver that protects the lungs. With this disorder, the AAT protein builds up in liver cells and doesn't reach the lungs to protect them. Augmentation therapy involves regular infusions of purified AAT protein to raise the level of the protein in the blood and lungs. Although the therapy has been available for more than 25 years, it has seen limited use because doctors have been unsure that it works.

The study, "Intravenous augmentation treatment and lung density in severe α1 antitrypsin deficiency (RAPID): a randomised, double-blind, placebo-controlled trail," published online today will change how clinicians understand this treatment and encourage them to consider its early use before the condition causes severe emphysema.

By using CT scans to measure the lung density of patients in the trial, the researchers were able to overcome some of the challenges that have been associated with studying the effectiveness of the treatment. "This treatment has now been studied in our centre using the most sensitive measure of lung structure - a radiologic measurement of lung density - allowing us to detect changes far earlier than can be seen with standard

breathing tests," said Dr. Kenneth Chapman, Director of the Asthma and Airways Centre at Toronto Western Hospital and the Canadian research lead for the multicentre trial. "We can now say with certainty that augmentation therapy is effective and should be given to patients with emphysema caused by this deficiency."

According to the *Canadian Medical Association Journal*, up to five per cent of people with chronic obstructive pulmonary disease (COPD) are thought to have alpha-1 antitrypsin deficiency, yet only four to five per cent of those with a deficiency have been identified. Even when the deficiency is diagnosed, there has typically been a delay of five to 10 years before this specific genetic problem has been identified as the cause of respiratory problems.

"Augmentation therapy not only preserves lung structure, but likely adds years of life," said Dr. Chapman. "Patients with this condition need access to timely diagnosis and treatments to ensure they receive the best possible care". Dr. Chapman added that this treatment is used only for this specific type of emphysema and is not of benefit to those with more common types of emphysema, chronic bronchitis or COPD.

Patients, like Ken Bee, who have been diagnosed with α1 antitrypsin deficiency, welcome the news that augmentation therapy is an effective treatment. Ken was diagnosed seven years ago and since receiving augmentation therapy his lung function has not decreased. "Without this treatment my symptoms would have progressed," says Bee. "My hope is that more people will recognize the benefit it has to patients."

Provided by University Health Network

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